

Training in genetics and genomics for primary health care workers

The study presented in this thesis was performed at the EMGO Institute for Health and Care Research in collaboration with the Department of Clinical Genetics, section Community Genetics of the VU University Medical Center, Amsterdam, the Netherlands and the Departments of Family Medicine and Educational Development and Research, Faculty of Health, Medicine and Life Sciences at Maastricht University, Maastricht in The Netherlands.

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Contents

Chapter 1	General introduction and outline of the thesis	9
PART I	<i>Creating an agenda for effective genetic educational strategies: needs assessment and prioritization in primary care</i>	
Chapter 2	Genetic educational needs and the role of genetics in primary care: a focus group study with multiple perspectives <i>BMC Fam Pract.</i> 2011;12:5	25
Chapter 3	Prioritisation of future genetics education for general practitioners: a Delphi study <i>Genet Med.</i> 2012;14:323-9	45
PART II	<i>Development and evaluation of genetic educational modules for general practitioners</i>	
Chapter 4	Sustained effects of online genetics education: a randomized controlled trial on oncogenetics <i>Eur J Hum Genet.</i> 2013 Aug 14. Epub ahead of print	65
Chapter 5	Effectiveness of oncogenetics training on general practitioners' consultation skills: a randomized controlled trial. <i>Genet Med.</i> 2013 May 30. Epub ahead of print	91
Chapter 6	Effect of a comprehensive oncogenetics training intervention for primary care physicians on multiple performance levels <i>Submitted</i>	119
PART III	<i>Summarizing discussion and future perspectives</i>	
Chapter 7	Proposed roadmap to stepwise integration of genetics in clinical research and practice <i>Clin Transl Med.</i> 2013;2:1-5	141
Chapter 8	General discussion	151
Chapter 9	Summary Samenvatting	171 177
Dankwoord		185
Curriculum vitae		197
List of publications		201

Appendices	1: Website www.huisartsengenetica.nl	208
	2: Link to Genetic online Continuing Professional Development (G-eCPD) module on oncogenetics, in Dutch: "PIN Kanker en erfelijkheid"	209
	3: Links to: Nederlands Tijdschrift voor Geneeskunde, Weekboeken	210
	4: Links to: Reacties Medisch Contact	214

Chapter 1

General introduction and outline of the thesis



Primary care providers such as general practitioners (GPs) and midwives are considered gatekeepers of specialized care; in general they are the ones who initially deal with patient's concerns and questions. Because of the increasing availability of DNA-based tests (<http://www.ncbi.nlm.nih.gov/sites/GeneTests/?db=GeneTests>) and the growing possibilities for genetic screening, primary care providers need to be prepared for patients asking for information or advice on genetic testing.¹ Moreover, increasing knowledge of genetics of common disorders (diabetes, cancer, cardiovascular diseases), and its monogenic subtypes (e.g. Maturity Onset Diabetes of the Young (MODY), hereditary breast cancer (*BRCA 1/2*) and Lynch syndrome (hereditary nonpolyposis colorectal cancer (HNPCC), familial hypercholesterolemia (FH) and long QT syndrome) asks for urgent genetic literacy of primary-care providers.²⁻⁴ It has been shown that physicians lack knowledge of genetics relevant for daily practice, lack oversight of genetic testing and concerns about privacy and discrimination, and report inadequacy to deliver genetic services.^{5,6} For genetics to have an effect on clinical practice that is comparable to its impact on research, will require integration of genetic medicine in daily health-care.⁴ In a previously held large European study, GPs mentioned "genetics of common disorders" as a first teaching priority.⁷ GPs' willingness to carry out genetic tasks showed 61% would take a family history themselves but only 38% would explain an inheritance pattern and 16% would order a genetic test.⁷ To overcome barriers, it was argued emphasis should be on genetics of common disorders and on how to deal with genetic risk in daily practice, rather than on ethics, basic concepts or new technologies.⁷ Country specific approaches however should be kept in mind, when genetics education aiming at these affecting factors is organized. In this thesis we will explore primary care workers' genetics educational needs and priorities, followed by determining the effectiveness of needs-based genetic training for GPs, taking oncogenetics as an example.

Primary care in the Netherlands: a phenomenon

Primary care (synonymous with primary *healthcare*) was defined in 2004 by the Dutch Health Council Committee, with regard to organisation and significance.⁸ Primary care is considered to be generalist care. It consists of general medical, paramedical and pharmaceutical care, nursing and supportive care and non-specialized mental and social healthcare, including preventive and health educational activities. Additionally, care should be provided as close to home as possible, in the middle of the community. Primary care is provided in collaboration with other primary care providers such as midwives and physiotherapists and, if necessary, with secondary care. The Health Council also stated the healthcare system, as a whole, will function more effectively and efficiently with purposely functioning

primary care with the general practitioner as a central gatekeeper in the Netherlands. Although there are some differences internationally to how primary care is defined, a stronger primary care system leads to more effective and efficient health care.⁸

Unlike in many other countries, general practice in the Netherlands is an open access full-time service for every patient with any medical complaint, request, or question. The service includes a list system, implying that every person (with or without a disease) is on the list of one GP, thus guaranteeing optimal continuity of care. The GP handles more than 90% of all presented complaints and diseases.⁸ All referrals to secondary specialized care, including to the nine Dutch clinical genetics centers, and most referrals to other primary care services are managed by the GP. Therefore, the GP is the first to whom a patient will turn when he/she has questions on prevention, diagnosis, and treatment of disease. They are highly involved in care for patients with common disorders such as cardiovascular disease, diabetes mellitus, and cancer.⁶ It has been argued that the greatest public health benefit of advances in understanding the human genome may be realized for these diseases.⁶ Realistically however, translation into clinical practice is still in the early stages.^{6,9} Family history taking, thus understanding mode of genetic transmission and counselling, predictive DNA-based testing, knowledge of relevant guidelines and timely referral to clinical genetics for disease prevention and reproductive decision making are currently understood to possibly be of great value in primary care practice.¹⁰ Messy translation of genetic advances into the clinic and unrealistic promises are limiting potential preventive treatments and improved medical care.¹¹

Role of genomics in primary care

Genomics is defined by the World Health Organisation (2002) as “the study of genes and their functions, and related techniques”.

Genomics addresses “all genes and their interrelationships in order to identify their combined influence on the growth and development of the organism”, whereas genetics “examines the functioning of a single gene”.¹²

In the age of genomics both *genetics of common disorders* and large-scale applications in *screening* will become increasingly important, and primary care health workers (GPs, midwives) will have to be prepared to discuss these issues with their clients accordingly.^{1, 3, 6, 13-16} GPs may get more involved in preventive check-ups and develop a more flexible way to deal with patients’ requests for DNA-based

tests, in addition to the original role in an open access full time service for every patient.

For the assessment of possibilities for predictive testing (positive family history of common genetic disorders, in particular its monogenic subtypes, and DNA-based test results) and its implementation in preventive check-ups, other public health workers in primary care (well baby clinics, occupational health care and policy makers) need to increase their genetic skills and knowledge. Therefore, genomics could change the roles of all actors in the field of primary care for recognition and validation of patterns of possible familial common disorders through family history taking, registration of this information in Electronic Patient Records (EPR), timely referral to clinical genetics departments, counseling its familial consequences and vital support. The question is how primary care providers themselves view the role of genetics in primary care.

Creating an agenda for effective genetic educational strategies:

needs assessment and prioritization in primary care

Training needs can be assessed both based on perceived needs by the target group of the training and on the challenges related to future applications expected by genetic experts. The target group will only participate in training if they feel that the training could be useful for them, and suit in their learner-focused needs-led plans. Modules that fit in their regular training programs and have been approved by or co-developed with their professional organization might be better accepted than initiatives superposed. Changes will only be effective if they suit routine practice, including the EPR and GP information system (HIS: *huisarts-informatiesysteem* in GP practice). A developing routine practice, the prevention consultation, might include genomics. Organizers of education must be responsive to work patterns in planning training sessions, encourage supporters from previous courses and specialist areas to promote genetics education.¹⁴

Primary health care professionals increasingly perceive an urgency to deliver genetic training needs based on their every day practice. These needs concern *knowledge* on a diversity of topics (genetics of common disorders, psychosocial and counseling issues, ethical, legal and public health issues, techniques and innovations in genetics), as well as *skills* (identifying sources of information, drawing up pedigrees, referring to specialist genetics services, communicating genetic information).

Stakeholders to be engaged in facilitating agenda setting and attunement could be clinical genetics professionals (clinical geneticists or genetic counselors), primary care educators, and representatives of patient advocacy groups. Stake-

holders are therefore considered experts to have a broad view of the role of genetics in primary care and the need for genetics education i.e. what is needed, what works and what does not work. In this thesis the views of these stakeholders and primary care workers regarding primary care workers' needs for genetics education and the role of genetics in primary care is addressed. This information could help to develop effective genetics education as well as effective integration of genetics in primary care.

Urgent need for genetics training for non-genetic healthcare providers: development and evaluation of genetic educational training for general practitioners

The rapid development of (also commercially available direct-to-consumer) genetic tests changes the role of the GP from a gatekeeper for the medical field.¹⁷⁻¹⁹ The possibility of a more flexible role in patient requested genetic tests must be investigated. Informed medical care should also imply informing patients about the pros and cons of genetic tests. GPs are confronted with clients entering their consulting-room with a high-risk estimate provided by a genetic company and based on a whole genome scan.¹⁹ A detailed consumer report of a genome map will almost certainly be beyond current physicians skills. The current development of preventive counselling sessions (*e.g. PreventieConsult*) in primary health care might be a good moment for this, to facilitate engagement of multidisciplinary stakeholders involved in genetic education development.⁸

In curricula of Dutch medical schools, postgraduate training for GPs and public health professionals as well as the master program for midwives, genetics and genomics are not very well represented.^{5,16} Both in the domain of reproductive medicine (e.g. risks related to consanguinity, carrier screening, prenatal screening), and in diagnosis, therapy and prevention of common disorders, fast developments in genomics research increasingly make large scale health care applications in the near future possible. It is estimated, each GP has about 40 to 50 asymptomatic patients with relatively young first-degree relatives with common forms of cancers (breast, ovarian, uterine and colorectal cancer) who should pre-symptomatically be counseled by a clinical geneticist and screened according to recent guidelines.¹⁹⁻²³ Women carrying a *BRCA1* or *BRCA2* mutation (a familial breast cancer gene), for example, have a lifetime-risk of 60-80% of developing breast cancer and 36-63% and 10-27% to develop ovarian cancer in case of respectively *BRCA1* or *BRCA2* mutation. Timely identification and referral to the clinical geneticist could obviously enable them to benefit from otherwise unex-

ploited life-saving “risk-management options”, such as salpingo-oophorectomy and/or mastectomy, annual screening, and pharmaceutical chemo preventive options.^{9,24} Timely referral may benefit not only the individual patient but also other family members who might be at risk.

To prepare clinical practice for the rapid advances in genomics, education is urgently needed. Genetic literacy allows health care workers to engage in the debate on hopes and hypes, and to be able to distinguish between useful and useless applications in health care. Untimely implementation of DNA-based test results in genetic medicine takes place by commercial companies that refer to health care workers, who are not prepared for this task.^{19,26} Several of the tests offered, lack clinical utility.²⁶ In the Netherlands, however, patients only sporadically request DNA-based tests. Focus on two tracks of possible applications of genetics and genomics in daily practice is essential; one will be a *proactive* and the other will be a *reactive* track. The *proactive* track primarily supports GPs in their preventive work of timely recognition of common genetic disorders and its monogenetic subtypes throughout their daily problem based work, when a proactive initiative is requested. The *reactive* track primarily supports the patient when he/she asks the GP a certain question related to genetics or genomics.

Defining genetic core competences for non-genetic health care workers was considered prerequisite for implementing genetics education for general practice.^{6,28,29} Such education programmes should be based on an educational needs assessment of GPs referring to the three domains of educational activities: cognitive (*knowledge*), psychomotor (*skills*) and affective (*attitude*). Despite competence frameworks and increasing demands, GP educators are struggling to respond adequately.¹⁵ This thesis was designed to investigate whether effective and sustainable education for primary care physicians (GPs) can be organized and developed, taking oncogenetics as an example. The following paragraph describes the conceptual models used in the studies performed in this thesis.

Conceptual model

The shared goal of improving patient care and health outcomes are not met: 30-40% of patients do not receive care informed by best evidence and 20-50% receive inappropriate care.^{8,9,11} Improving educational strategies to enhance application of evidence in practice, through Continuing Professional Development (CPD) is essential if new clinical evidence is to lead to improved patient care and health outcomes, whether it is innovations in genetics or otherwise. The accrediting bodies now require medical professionals not only have to increase their

knowledge through CPD, but also their performance in daily practice through improved skills and attitude. Online Continuing Professional Development (eCPD) and other more traditional CPD activities (Printed Educational Materials (PEMs) and live CPD activities) are widely used to inform healthcare professionals.²⁹ Internet based CPD potentially reaches many users and showed to be similarly effective and could therefore be cost effective.^{30, 31}

The conceptual model presented here is based on Kern’s CPD curriculum design, derived from general education theory and practice and entails a six-step circular guide.^{32, 33} Similar to research outcomes, learner outcomes should be considered when the curriculum is defined (learning goals and objectives, teaching methods, content and assessment). Developing effective training, each step must show congruence with the other steps, which is demonstrated on the figure based on Kern’s design. Kern described the steps to come to the ultimate goal of CPD through education and learning: improved patient health and outcome. Actually, Kern’s *six*-step guide should rather be called *seven*-step guide, since this processes sustainable curriculum maintenance and enhancement.

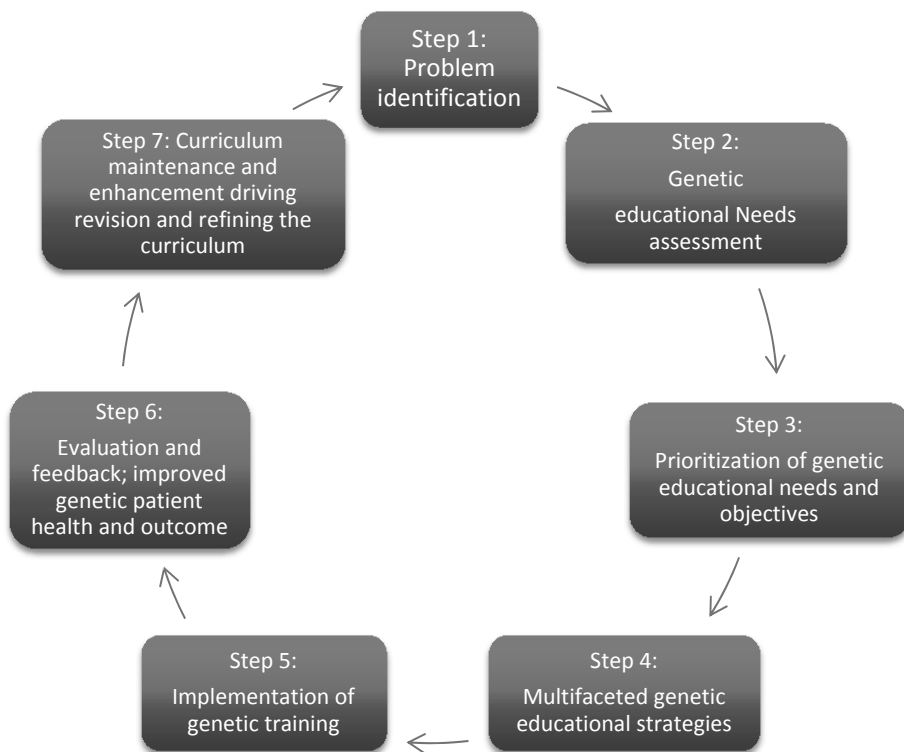


Figure 1. Conceptual model for genetic CPD curriculum development, evaluation and maintenance based on Kern ^{32, 33}

CHAPTER 1

The goal of CPD interventions is not necessarily change in translation of knowledge, but rather learning at various levels. Levels of educational outcomes can be demonstrated by Kirkpatrick's framework for evaluating educational outcomes, originally presented in 1967.^{32,34} The framework proposes four levels of outcome for educational interventions: valuation (level 1; satisfaction), learning (level 2; knowledge and knowledge retention), behaviour (level 3: applied knowledge on timely recognition of patients at risk and referral) and ultimately effects on patient health and organization and therefore impact on society (level 4: change in actual practice performance, results and effects on patient health and organization).

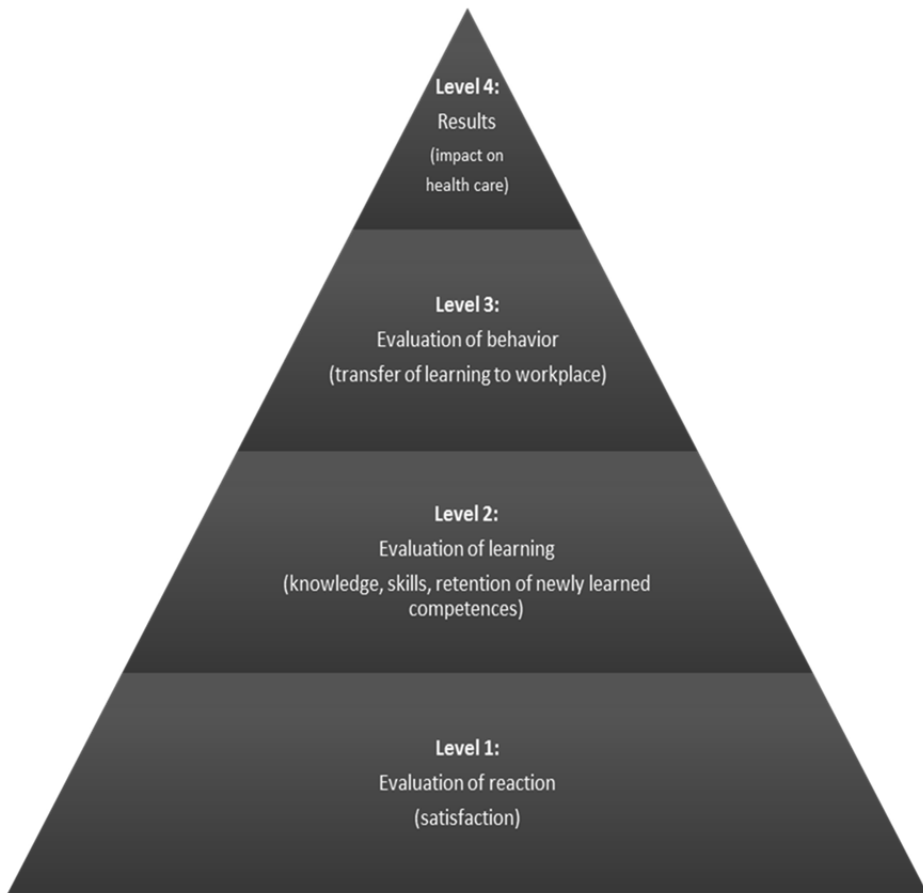


Figure 2. Genetic educational framework. Based on Kirkpatrick's Evaluation Framework for Educational Outcomes^{32,34}

In this thesis these four levels of Kirkpatrick's are assessed due to oncogenetics CPD activities organized for GPs. The format used to develop oncogenetic CPD training (online and live) and a website to use as a supporting tool for GPs (www.huisartsengenetica.nl). Evaluation is done on all four levels of Kirkpatrick: satisfaction with the oncogenetic training organized, usability and user-friendliness of a supportive website, knowledge and communication skills and change in daily practice performance (referral rates to departments of clinical genetics).

Outline of the thesis

This thesis describes the development and evaluation of oncogenetic CPD modules as a thematic example for general practitioners in collaboration with and joining The Dutch College of General practitioners (*Nederlands Huisartsen Genootschap, NHG*) based on previous assessments of educational needs and prioritisation of the genetic educational topics. Three genetic training modules endorsed by The NHG were developed: an online and a live interactive CPD module on oncogenetics and a new developed easily accessible website for daily use in practice (www.huisartsengenetica.nl). The effectiveness of educational outcomes of needs-based genetics training was examined according to Kirkpatrick's four levels.

Unfortunately training alone is often not effective to change clinical practice. Additional measures and actions targeting obstacles to change at the level of teams or organisations are needed when plans are developed enabling daily practice change.^{4,35-38} We also investigated key factors (e.g. collaboration with key actors in general practice) for successful future genetics training. Ultimately, gained knowledge, skills and attitude on genetics and cancer in family medicine could lead to better referral strategies to clinical genetics according to clinical guidelines. This would make recognizing familial forms of cancers, improved risk stratification in clinical practice, life saving "risk-management options" and timely referral feasible. The results may serve as a model for future GP genetics training on other common disorders (diabetes, cardiovascular disease), but may also serve as a model for genetics training for other primary and secondary care professionals and to improve the medical curriculum and biology lessons in high school curricula.

The results from this thesis could therefore serve for a step-by-step roadmap proposal, to effectively integrate genetics in daily general practice to its full potential.

The following research questions will be addressed in this thesis:

Q1. What are the current primary care educational needs with regard to genetics/genomics?

- a. What is the role of genetics/genomics in primary care according to primary care health workers and different stakeholders (i.e. clinical genetics professionals, primary care educators, and patient advocacy groups)?
- b. What are the genetic/genomics educational priorities according to primary care health workers and stakeholders?

Q2. Can effective and sustainable oncogenetics education for primary care physicians (GPs) be organized and developed?

- a. Is an online oncogenetic CPD training module (G-eCPD) effective in improving GPs' knowledge?
- b. Is a live oncogenetic CPD training effective in improving skills and attitude?
- c. Is there a change in actual practice performance (i.e. (self-reported) change in patient referral to the department of clinical genetics) as a result of the oncogenetic education modules organized
- d. Is a practice-based website for GPs (www.huisartsengenetica.nl) used as a supporting tool during daily primary care practice?

Q3. Is it possible to integrate genetics step by step in daily genetic primary care and therefore make operationalization possible?

Chapter 2 presents the results of a focus group study among GPs, midwives and experts and enables the exploration of the meaning and significance of the role of genetics and the need for education in that area as perceived by different stakeholders (**Q1a**).

Chapter 3 describes a Delphi study on prioritisation of topics in a "Top 10" for genetics education for general practice after exploring the themes found through the previously held focusgroup transcript analysis (**Q1b**).

Chapters 4 and 5 will answer whether useful and effective CPD modules on genetics for non-genetic healthcare workers in general practice can be organised and whether genetic competences and actual performance in daily general practice can be improved (**Q2a and b**).

Chapter 6 will give an overview of the results of the evaluation of effectiveness of all the genetics CPD modules organised and developed. This will answer the last research question (**Q2c and d**) and will determine whether changes in the organization of genetic health care have been achieved and whether new designs

of CPD can sustainably be developed that turn primary health workers in to learners.

Chapter 7 describes a 5-step roadmap, which will make it possible to integrate genetics information in the Electronic Patient Record (Q3). Through effective implementation of genetics education, operationalization of genetics innovation through adding relevant ICPC codes for simple registry of family history, could finally become reality. The next step in future research: improved genetic medical care in everyday medical practice.

Chapter 8 starts with a critical evaluation of the outcome of the studies, focusing on the established changes in the organization of genetic health care, followed by recommendations for possible changes and suggestions for implementation of genetics education both nationally and internationally and future research projects.

Chapter 9 summarizes the studies and conclusion presented in this thesis.

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GENERAL INTRODUCTION AND OUTLINE OF THE THESIS

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PART I

Creating an agenda for effective genetic educational strategies:

needs assessment and prioritization in primary care



Chapter 2

Genetic educational needs and the role of genetics in primary care:

a focus group study with multiple perspectives

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Geert Jan Dinant, Martina C. Cornel

BMC Fam Pract. 2011;12:5

Abstract

Background

Available evidence suggests that improvements in genetics education are needed to prepare primary care providers for the impact of ongoing rapid advances in genomics. Postgraduate (physician training) and master (midwifery training) programmes in primary care and public health are failing to meet these perceived educational needs. The aim of this study was to explore the role of genetics in primary care (i.e. family medicine and midwifery care) and the need for education in this area as perceived by primary care providers, patient advocacy groups and clinical genetics professionals.

Methods

Forty-four participants took part in three types of focus groups: mono-disciplinary groups of general practitioners and midwives, respectively and multidisciplinary groups composed of a diverse set of experts. The focus group sessions were audio-taped, transcribed verbatim and analysed using content analysis. Recurrent themes were identified.

Results

Four themes emerged regarding the educational needs and the role of genetics in primary care: (1) genetics knowledge, (2) family history, (3) ethical dilemmas and psychosocial effects in relation to genetics and (4) insight into the organisation and role of clinical genetics services. These themes reflect a shift in the role of genetics in primary care with implications for education. Although all focus group participants acknowledged the importance of genetics education, general practitioners felt this need more urgently than midwives and more strongly emphasized their perceived knowledge deficiencies.

Conclusion

The responsibilities of primary care providers with regard to genetics require further study. The results of this study will help to develop effective genetics education strategies to improve primary care providers' competencies in this area. More research into the educational priorities in genetics is needed to design courses that are suitable for postgraduate and master programmes for general practitioners and midwives.

Background

In the age of genomics, the genetics of common chronic disorders, pharmacogenetics and large-scale applications in screening are becoming increasingly important. Primary care providers (e.g. general practitioners and midwives) will have to discuss these issues with their patients, who are becoming increasingly aware of genetic contributions to disease and also have high expectations of genetic testing.¹ Consequently, primary care providers need to be educated to meet the needs of their patients that are created by rapid advances in genomics.² Genetics literacy among primary care providers needs to be improved to enable their participation in the debate on the hopes and hypes of genomic medicine and to distinguish between useful and useless practical applications in health care.³ Currently, genetics and genomics are rather underrepresented in postgraduate (physician) training programmes in general practice (here, the terms *general practice* and *family medicine* (commonly used terms in the Dutch health care system) are considered synonymous to the more commonly used term *family practice* in the U.S. healthcare system) as well as in master programmes in midwifery and public health.^{4,5} It is widely recognized that medical professionals and medical students should be educated about genetics.^{2,5-7}

Research into the perspectives of general practitioners and midwives on the educational priorities and attitudes in relation to genetics revealed a need for genetics education for primary care providers in areas like psychosocial issues and screening, assessment of the risk of genetic malformations and basic genetics.⁸⁻¹⁶ However, the educational needs of primary care providers and their views on the role of genetics in family practice are still under investigation, and international efforts to translate these needs into education programmes are still in their early stages.^{17,18}

Primary care providers have a unique role in the Dutch health care system and general practitioners are easily accessible to all patients for any complaint, request or question. Midwives provide obstetric and perinatal care and give advice and guidance to patients on pregnancy and childbirth. Genetics could have an effect on daily primary care practice if basic and clinical science advances in genomics of common chronic diseases in practice and midwifery care are successfully translated.^{2,3,19} Changes will only be effective, however, if they fit well into practice routines. It is therefore important to understand how these key professional groups conceive of their responsibilities and experiences in relation to genetics. For the implementation of genetics training in daily practice to be successful, it is important to identify factors that can enhance or inhibit effective genetic primary care.

Up till now, most studies of educational needs concerning genetics have been limited to the perspectives of target groups.^{1, 4-14} Professional training needs can be derived from those studies and from challenges posed by new applications as foreseen by experts. This study explored the views of general practitioners, midwives, patient advocacy groups and others involved in genetics in health care and education regarding their need for genetics education and the role of genetics in primary care. This information was collected to help develop effective genetics education and training as well as effective integration of genetics in primary care.

Method

Design

We used a qualitative study design with focus groups because this enables the exploration of the meaning and significance of the role of genetics and the need for education in that area as perceived by different stakeholders. A discussion with members of the research team and primary care providers revealed that general practitioners and midwives were the primary care providers most likely to be confronted with issues of genetics and genomics in their practices.

Participants

We used purposive sampling to recruit specific groups of professionals for focus group interviews in order to obtain rich, relevant and diverse data.

The participants were expected to provide complete and possibly complementary perspectives on genetics in primary care practice and education. Potential participants were named by key persons and network contacts at academic departments of general practice and the midwifery academies in Amsterdam and Maastricht, the Netherlands.

We convened three types of focus groups, (1) two groups of general practitioners, (2) two groups of midwives, and (3) three multidisciplinary groups composed of clinical genetics professionals (clinical geneticists or genetic counsellors), primary care educators, and representatives of patient advocacy groups. The participants in the multidisciplinary groups were considered experts who were expected to have a broad view of the role of genetics in primary care and the need for genetics education i.e. what is needed, what works and what does not work.

For each focus group, ten to fifteen professionals from one region were invited by email or telephone. Those who responded positively received an invitational letter, informing them that the purpose of the study was to explore their perceptions of the role of genetics and genomics in primary care and the related education needs. The term 'genetics' was commonly used during the focus group discus-

sions, and the term ‘genomics’ was used to denote a broad definition (e.g. common complex disorders and rapid technological developments).

Focus groups

Table 1 provides an overview of the participants. The interviews lasted approximately two hours and were held between March and August 2009. The discussions were facilitated by an independent and experienced moderator (SL), who encouraged the participants to participate actively and to openly state their viewpoints and engage in discussion. An assistant (IH) took notes and all the sessions were attended by one observer (LH). Participation was voluntary and participants received €100 plus travel expenses.

The study was approved by the Medical Ethics Committee of VU University Medical Center, Amsterdam and Maastricht University. All participants gave informed consent at the start of their focus group session.

Table 1. Characteristics of the participants in the focus groups

Type of focus group	N	Female (N)	Mean age in years (SD)	Mean work experience in years (SD)	Professional background
General practitioner group 1	7	3	40.6 (8.1)	11.6 (7.8)	general practitioner
General practitioner group 2	6	3	49.3 (10.2)	19.2 (12.3)	general practitioner
General practitioner group total	13	6	45 (8.7)	15.4 (9.7)	
Midwife group 1	8	8	39.3 (6.1)	13.5 (6.2)	midwives
Midwife group 2	6	6	32.2 (7.9)	7.8 (6.6)	midwives
Midwife group total	14	14	35.7 (6.7)	10.6 (6.1)	
Multidisciplinary group 1	6	5	52.2 (8.3)		3 midwives-midwifery teachers, 1 medical doctor-midwifery teacher, 1 researcher, 1 medical psychologist
Multidisciplinary group 2	7	5	49.7 (6.0)		2 patient organisation representatives, 2 medical doctors, 1 midwife-midwifery teacher, 1 policy advisor, 1 clinical geneticist
Multidisciplinary group 3	4	3	47.8 (14.6)		2 patient organisation representatives, 1 genetic counsellor, 1 policy advisor
Multidisciplinary group total	17	13	49.9 (8.7)		

Interview guide

An interview guide with open-ended questions was developed to ensure coverage of the major topics (table 2). The moderator opened each focus group interview with an introduction and a round-robin open question: “What are your experiences with genetics/genomics in primary care?” Further probing by the moderator was rarely required, since each group spontaneously talked about genetics education needs and the role of genetics in primary care. When needed, asking for clarification of the answers was sufficient to elicit ample additional information.

Table 2. Interview guide for the focus group discussions

Genetics in primary care; assessment of need for inclusion of genetics in primary care education

What comes to mind when you think of genetics in primary care?

Do you think primary care workers are capable of answering questions about genetics? Why (not)?

Genetics in primary care; assessment of the role of genetics in primary care

In your opinion, what is the role of genetics/genomics in primary care today and what will it be in the near future?

What is your opinion about the genetic knowledge and skills currently available to primary care providers to fulfill this role?

In your opinion, what are the most important genetic topics for the education of general practitioners and midwives?

Data analysis

The focus groups were audio recorded and transcribed verbatim. Member checking entailed sending a summary of the sessions to all participants and inviting their comments, which were then incorporated in the transcripts. Using Atlas.ti5.2 for data analysis, two of the researchers (IH and LH) independently coded the themes that emerged from the transcripts. They compared their coding for reliability and reached consensus on differences through discussion. Through a process of discussion and deliberation, connections between the codes were identified and categories and themes developed. The transcripts were read repeatedly to check the accuracy and completeness of the themes and subthemes. In the results section, representative quotes from the focus groups, translated from the Dutch, are presented to illustrate the themes.

Results

All participants acknowledged the importance of genetics education for primary care providers. A need for education was expressed more urgently by the general practitioners than by the midwives, while members of the multidisciplinary group generally indicated that both groups were deficient in genetics knowledge and skills. The extent and focus of the discussions differed between the general practitioners and the midwives, because the general practitioners saw themselves as generalists while perinatal care was the primary focus for the midwives. This difference was reflected in their priorities for education.

Four distinct themes emerged: (1) the need for genetics knowledge, (2) taking a family history, (3) ethical dilemmas and psychosocial effects related to genetics, and (4) insight into the organisation and role of clinical genetics services (table 3).

CHAPTER 2

Table 3. Themes and subthemes identified regarding the genetic educational needs of general practitioners and midwives

Themes	General practitioners	Midwives
1. The need for genetic knowledge	<p>Perceived need for genetic knowledge</p> <p>Aware of lack of knowledge</p> <p>Cannot know everything</p> <p>Knowledge should be aimed at prevention of common diseases</p> <p>Want to know where to find genetic information</p>	<p>No perceived need of genetic knowledge</p> <p>Not aware of lack of knowledge</p> <p>Knowledge should be aimed at prevention of perinatal diseases</p> <p>Want to know where to find genetic information</p>
2. Taking a family history	<p>Taking a family history and pedigree drawing is not routine</p> <p>Systematically taking a family history could be improved</p> <p>Registering family history is complicated</p>	<p>Taking a family history is routine</p> <p>Pedigree drawing is not routine; infrequently carried out and therefore skill not maintainable</p>
3. Genetic ethical dilemmas and psychosocial effects	<p>Increasingly confronted with genetic dilemmas and psychosocial issues</p> <p>Privacy issues and genetics complex</p> <p>Questions on what should be genetically identified and what should not</p> <p>Perceived difficulty of non-directiveness in consultations</p> <p>Discussing consanguinity is difficult</p>	<p>Genetic developments in perinatal screening are rapid; raises more ethical questions</p> <p>Discussing consanguinity is difficult</p> <p>Discussing preconceptional and prenatal screening is difficult due to inter-cultural differences</p>
4. Insight into the organisation and role of clinical genetics services	<p>Do not know when to refer to clinical genetics services</p> <p>Prefer to sort things out by themselves first</p> <p>Little insight into the organisation of clinical genetics centres and clinical trajectory</p>	<p>Consult clinical geneticists easily</p> <p>Little insight into the organisation of clinical genetics centres and clinical trajectory</p>

1. The need for genetics knowledge

General practitioners perceived deficiencies in their basic understanding of genetics, since they had never been taught this or because their knowledge had faded. They experienced their lack of knowledge as a barrier to the use of genetics in diagnosis, treatment and in consulting clinical geneticists, and they expressed a strong need for this knowledge, as reflected in the words of one general practitioner (male, 37 years):

"I think it's essential to know the basics [of genetics]... You should know what a gene is, that there are deviant genes and that genes can be turned on and off... These are basics that clarify genetics and make it understandable so that it can be translated to patients in relation to diagnosis or treatment or to advise them to refrain from something, smoking, for example."

Other participants acknowledged this need and argued that general practitioners should have more knowledge in order to provide general genetic information. Primary care providers wondered how much they really needed to know about such a complex field. All groups shared the view that primary care providers cannot be expected to know everything. Since many genetic diseases are rarely seen in primary care, there is no urgent need for them to be included in training programmes.

General practitioners appeared to be generally aware of their lack of knowledge, which made them ill equipped to identify genetic problems in their patients. In response to the question about experiences with genetics/genomics, one general practitioner (male, 52 years) said:

"I desperately hope the midwife or obstetrician will think of [prenatal diagnosis for women of advanced maternal age], because I don't always think of bringing it up. Also, I tend to think the specialist will consider all the genetic aspects of a clinical problem, but this often is not the case."

Midwives did not perceive a lack of genetics knowledge and said the midwifery master programme provided sufficient education on this topic. Multidisciplinary group members, however, said that both general practitioners and midwives needed education to increase their knowledge, as they observed a lack of knowledge in both groups.

There was a discrepancy in the content of the required knowledge as perceived by the participants. General practitioners mentioned a need for knowledge about prevention of common genetic diseases, whereas the midwives were primarily interested in prevention of perinatal diseases. To meet these educational

needs, general practitioners and multidisciplinary experts believed that genetics education should address family history and inheritance patterns.

Both general practitioners and midwives were interested to know where they could find more genetic information. There appeared to be a general need for easily accessible sources of information such as web-based education or websites with short and easy to understand information that could be applied in daily practice.

2. Taking a family history

Primary care providers believed that taking a family history was extremely important as it allows for familial risk stratification and identification of hereditary conditions. For midwives the importance of family history was limited to perinatal disease. The need to increase awareness of familial diseases in primary care was discussed by a midwifery educator (female, medical doctor, 56 years):

“We agree on when to think of some familial diseases. I mean, it’s clear when speaking of colon carcinoma or breast cancer that you realize [as a general practitioner] that it can be inherited. There should be a clinical guideline to help one decide when to consult a clinical geneticist for further diagnosis. Today, more general practitioners should realize this [colon carcinoma or breast cancer] could be familial.”

An educator, allied to a postgraduate general practice programme, thought that taking a family history was well covered during postgraduate training, but the general practitioners were not quite so sure. Although family history was part of some consultations, most general practitioners said that family history and pedigree drawing were not part of their daily routine. The midwives said that family history was something they did every day, but they lacked the skills for pedigree drawing. They thought this was not important, however, because they did not do it often enough to maintain this skill. They thought the main thing was to be able to recognize high-risk factors in a family history

“Midwives generally lack sufficient knowledge to draw a pedigree. I think the same applies for general practitioners, but it is more important for them [general practitioners] to detect high-risk family history criteria. How and when is something important? The signalling function, that is important and it should be taught. Counselling is important for both general practitioners and midwives. But above all it is important to be clear on when something is important, how important it is for you, what you want to do about it, and once this is clear you can take the next step” (a female GP and midwifery educator, 39 years).

General practitioners were uncertain about recording information from family history in the Electronic Patient Record. Participants said that measures should be taken to improve the Electronic Patient Record to include information from family history.

3. Ethical dilemmas and psychosocial effects related to genetics

Most primary care providers expressed concern about the surge of genetic testing which confronted them with ethical dilemmas and more profound psychosocial effects of genetics in their daily practice. One participant said he was faced with “an increasing amount of vague, worrying and inexplicable genetic information”. He referred to information about genetic risks provided to consumers by commercially available Personal Genome Services. Primary care providers felt unqualified to deal with these issues and thought that genetic ethical dilemmas should be part of genetics education.

General practitioners also wondered whether it was beneficial to their patients and themselves to know everything about a patient’s genetic background. They voiced concern about the possibility of unauthorized dissemination of genetic information and related privacy issues if it were to become obligatory for them to take a family history and record the information derived from it. They asked: “Who should you inform about genetic information and who should you not inform if you want to keep your patient’s best interest at heart, for example when a patient wants to take out life insurance or needs a mortgage to buy a new house?”

Midwives thought that developments in genetics were moving at a very rapid pace, giving rise to feelings of insecurity both in midwives and their patients. They discussed whether following the protocol for perinatal screening might be inappropriate and therefore not uniformly applicable. They preferred to adapt the application of genetic protocols to individual patients, because test results could have important genetic and personal consequences. One midwife, (female, 40 years) explained:

“Surely because this child is already in the uterus, the basic question is rather what do you really want to know? Because what are you going to do with this [genetic] information?”

Clear guidelines as to when a general practitioner should be proactive and bring up the subject of familial disease to patients and their families were non-existent but considered necessary. General practitioners were unclear about how to guide patients in their decisions around prenatal and genetic testing. As a result they perceived non-directive counselling as difficult because it was influenced by their personal opinions and sense of urgency. One general practitioner (male, 52 years) explained:

“I used to discuss prenatal screening with patients at first when it was a hot item. Everybody wanted the triple test, but this seems to change when you explain that it gives a probability that doesn’t offer any certainty and things can also happen after the baby is born or you cannot always see things on the outside and people then pull back automatically. I try to counsel nondirectively, because it promotes shared responsibility. I like to hold on to this nice shared responsibility, because it doesn’t make me feel that I am solely accountable.”

General practitioners and midwives alike mentioned consanguinity as a complex issue to discuss with patients and they raised the problem of how to deal with a potentially increased risk of congenital disease. One general practitioner (female, 52 years) said:

“When cousins get married, they are blissfully happy when presenting this news to their doctor. As a general practitioner I find it complicated, when there is a disease in the family, to confront these people with this problem in today’s society.”

Inter-cultural differences were considered a source of difficulties in discussing prenatal or preconceptional screening. Midwives sensed urgency early on in pregnancy when recommending this type of screening to members of ethnic minorities in order to prevent congenital disease. This feeling of urgency was sometimes enhanced by language barriers and time constraints.

4. Insight into the organisation and role of clinical genetics services

General practitioners expressed a need for education with regard to indications for referral to clinical genetics services. Some general practitioners preferred to first gather information from other sources (such as online websites) before turning to a clinical geneticist. Other general practitioners said it was better to refer than to do it all yourself. Midwives, on the other hand, said it was easy for them to consult clinical geneticists, whom they regularly telephoned for advice.

Some general practitioners and midwives said it was not clear to them what clinical geneticists do and what the clinical trajectory would be once a patient was referred to such a service. General practitioners mentioned their lack of familiarity with this type of service as a cause of inappropriate consultation strategies, which could result in untimely referrals. Geneticists argued that general practitioners should consult them more often, and that this should be stimulated by education.

“The relation between primary and secondary care is sometimes difficult. Once the clinical geneticist has diagnosed a certain genetic disease, which means more specialised information, the patient’s family members find themselves in a pickle together with the GP and then the whole process (of consulting) starts all

over again. [...] I hope education of primary care providers will result in accessible consultation services. Because I don't think the general practitioner should know everything about everything ... but they should at least know that help is easily available.” (Clinical geneticist, female, 45 years).

The role of genetics in primary care

The role of genetics in primary care was perceived to be unduly limited as a result of care providers' inadequate genetics knowledge and skills. Although care providers might show some interest in improving their knowledge, representatives of patient advocacy groups indicated that primary care providers were “not sufficiently proactive” in this area. They perceived an urgent need for inclusion of genetics in primary care guidelines in order to make genetics a “hot item”. General practitioners and midwives said they were unsure about their responsibilities in relation to genetics, perhaps because they lacked insight into the genetic background of diseases and its possible consequences. A representative of a patient advocacy group (male, 59 years) said:

“No knowledge (of genetics) and no interest (in genetics). It's not a hot item. It seems as if general practitioners are not interested in identifying patients with familial hypercholesterolemia (FH). We have an excellent screening programme for this in the Netherlands, which threatens to go under because too few index patients are put forward by general practitioners. General practitioners do not alert patients that they might have FH when there is a positive family history and high cholesterol levels, and patients are not advised to take part in a brief screening programme [...]. General practitioners often say they don't have the time or they're not interested or they see no benefit.”

Primary care providers noticed a change in their experiences with and views of the role of genetics in primary care, which led to an increased need for basic knowledge of genetics and family history taking. General practitioners felt their current knowledge was insufficient to meet these needs. Participants also said they noticed an increase in patients' questions about genetic issues. They perceived a change in the responsibilities of primary care providers that prompted increased attention for genetics. They also saw an urgent need for a description of the responsibilities of different disciplines in relation to genetic issues. A clinical geneticist (female, 40 years) put it as follows:

“When I think of genetics I think of monogenetic disorders, but of course disorders (seen in primary care) are often complex disorders or multigene or gene environment interaction disorders. Of course, clinical genetics cannot deal with all those problems, it's simply impossible. [...] I think it is the task of the general

practitioner, but I find this difficult... is it really the task of the general practitioner to deal with such complicated problems?"

Overall, participants were positive about the changed role of genetics in primary care. They said this change emphasized their role as an easily accessible source of information. However, there was also some criticism. General practitioners saw even more work coming their way, which caused some concern. Taking a family history, non-directive counselling, and unfamiliarity with recording information from family history in their electronic patient record were said to take up a great deal of time.

General practitioners indicated that they were aware of rapid developments in genetics and the subsequent lag in its application in primary care. They regarded this as important and pointed to two aspects of this change. Firstly, they said it was urgent for limits to be set in relation to required genetics-related knowledge and responsibilities in primary care. Secondly, education should include the clinical application of genetic developments and ways to communicate genetic information. A midwifery educator (female, medical doctor, 56 years) clarified these aspects:

"Highly educated people develop national genetics guidelines. Even the ethical issues involved in these problems and how to deal with them are prescribed. In primary care you are in close contact with patients and it can be difficult sometimes to apply theory-based guidelines in a way that can be understood by patients, it is difficult to do this appropriately."

Suggestions for strategies for effective genetic education

At the end of each focus group interview, the participants were asked to briefly consider effective strategies for teaching genetics in primary care education (table 4).

The following general considerations emerged: programmes should be relevant to primary care practice, participants in the multidisciplinary group emphasized the importance of assuring the quality of educational strategies and suggested that programmes should range in duration from brief sessions to ten-day programmes. Finally, strategies should be added to existing programmes or could be integrated with other topics, such as cardiovascular risk management or familial breast cancer as examples of common diseases.

Table 4. Suggested strategies, including details, for effective ways to incorporate genetics into primary care education

Strategies	Details
A short internship in the clinical genetics department to familiarise students with the specialty	Suggested by clinical genetics professionals, primary care educators and younger general practitioners
E-learning	Younger participants were in favour
Lectures	Preferred by older participants
New guidelines which should be easily accessible on a website	A decision tree should be attached to help GPs and midwives with busy schedules to quickly find genetic information about how, when and to whom to refer.
Workshops	Learning by discussing clinical cases with colleagues and attending clinical geneticists, paediatricians or other specialists depending on the topic under discussion. Basic genetic knowledge, skills, such as drawing a pedigree, and attitude, through discussing medical ethical topics, could be included.
Continuing Professional Development	CPD genetics sessions repeated yearly and accredited with CPD credits to promote attendance.

CPD = Continuing Professional Development

Discussion

The results of this study indicate that Dutch primary care providers need, and would welcome, more extensive education in genetics. Four major themes emerged in relation to the role of genetics in primary care and the related educational needs: lack of basic knowledge, need for education on family history taking and the potential clinical consequences, ethical dilemmas and psychosocial effects related to genetics and insight into the organization of regional genetics services and the referral system. There was general agreement that increased genetics knowledge and family history taking by primary care providers would require a better understanding of the organization of genetics services in order to promote more appropriate and timely referrals. In summary, the results point to a need for courses in genetics for master programmes in midwifery and postgraduate programmes in family medicine.

A similar need for genetics education in primary care was also found in other studies. The identified needs are in line with the learning outcomes and core competencies in genetics proposed by genetics experts for non-genetic health care professionals.^{17,18} Since there is little published research on the extent to which the need for genetics education matches the core competencies, we used a qualitative approach to explore the views of the target group. In this way we gained insight into the educational needs of this group with regard to genetics; general practitioners indicated that a paucity of knowledge can lead to poor recognition of and unresponsiveness to genetic problems in daily patient care.

The results of this study are in line with some studies and differ from others with regard to the need for increased genetics knowledge among midwives and general practitioners.^{5, 8, 10, 12, 13, 14, 16, 19, 20} The midwives in our study seemed more confident of their basic knowledge and did not perceive as strong a need to adapt existing educational programmes as was expressed by midwives in studies by Benjamin *et al.* and Metcalfe *et al.*^{8,13} This difference may be due to differences between master programmes in midwifery or between health care systems.

Our results support the outcomes of the afore mentioned studies regarding deficiency in skills (e.g. taking a family history, referral to appropriate regional genetics services and non-directive counselling).^{8, 13} It may be problematic for primary care providers to take appropriate steps in response to the perceived shift in the importance of genetics in primary care, such as taking enough time to discuss the family history or non-directive counselling. Another step to take would be to improve the Electronic Patient Record in order to achieve accurate documentation of family history information.

Martin and Wilikofsky reported on general practitioners' perceptions of their role in genetic counselling and their unwillingness to accept this role due to time and organizational constraints.²¹ Representatives of patient advocacy groups and genetic counsellors in our study emphasized the need to increase acceptance of the importance of genetics and genetic counselling in primary care. The responsibility, on the part of the patient or the doctor, to report data from the family history remains a topic of debate, however, even though the importance is clear and primary care seems well suited to include this role in daily practice routines.²² Perhaps a joint effort by all stakeholders would be realistic and useful.

General practitioners and other participants in our focus groups recognized the important role of genetics in primary care. This is in contrast to a study conducted by Fetters *et al.* in 1999, which found general practitioners reluctant to invest in self-education in genetics, because they felt genetic problems were not clinically relevant.¹² Our study suggests that today's primary care providers are aware of a progressive impact of genetics on primary care and therefore increasingly conscious of what they don't know. They recognize the need for attention to genetics

in educational programmes. Perhaps this is a reflection of family medicine finally becoming aware that genetics and genomics are an integral part of primary care.

Clinicians were seen to be uncomfortable in applying genetics in their daily practice, which resulted in difficulties in referring adult patients for genetic counselling.⁷ Our study showed similar results. Some general practitioners were reluctant to consult a clinical geneticist, whereas midwives seemed to be more comfortable with this. Representatives from patient organizations were also aware of this barrier and urged more genetic education for primary care providers, general practitioners in particular. Taylor *et al.* also suggested that insurance coverage of genetic consultation can be a problem. There is currently a paucity of published research on the clinical value of genetic evaluation in primary care.²³⁻²⁵ Genetic counselling could be of greater value and might be integrated in periodical check-ups more often if its results had greater practical applicability.

The educational strategies suggested by general practitioners and midwives in this study appear to be supported by Gaff *et al.*, who concluded "Program logic, adult learning theory, and evaluation theory together provide a useful and relevant theoretic framework for the development of genetics education programs for health professionals."²⁶

Limitations

The use of focus groups has engaged primary care providers of a potential genetics education programme in the Netherlands. A variation in concepts is possible, because it is unknown how far the themes reach in their contribution and interaction in real practice. The aim of this study was intended to yield results regarding the participants' particular views on knowledge, skills and attitudes in relation to genetics education in primary care. Apart from homogenous groups of general practitioners and midwives, we included participants from a variety of backgrounds to obtain input on broader and future developments in genetics in primary care. However, it remains to be investigated if the results have relevance beyond the Dutch health care system, since the nature of the sample was drawn from this particular health care system.

Together with previously published studies on various aspects of genetics in primary care education, our study offers a broad perspective on genetics education. We believe this information can be used to develop genetics education programmes in the near future. The inclusion of multidisciplinary focus groups which could provide meta views can be considered a strength but also a weakness of this study because of the unequal representation of different fields of expertise in these groups. Another limitation is that purposive sampling can result in self-selection, which can introduce bias. Our study revealed four major themes concerning the role of genetics in primary care. In order to ensure that our picture is complete and

usable for educational purposes, and possibly for policy makers as well, consensus has to be sought, for example by means of a Delphi procedure.

Conclusions

The results of this study suggest that postgraduate training in primary care could be enhanced by incorporating additional training in basic clinical genetics. For midwives and general practitioners there should be more emphasis on counselling using strategies that are clinically feasible and on ethical issues relating to genetic conditions. Insight into the organization of regional genetics services and the referral system should be enhanced to promote interdisciplinary collaboration. There is an urgent need for a clear description of responsibilities and guidelines to enable effective use of developments in genetics in primary care. Especially descriptions of the genetic responsibilities of primary care providers and their specific role in this area will have to be addressed by future research. Useful and effective application of genetics knowledge can only become a reality when genetics education is improved.

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Chapter 3

Prioritisation of future genetics education for general practitioners:

a Delphi study

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Abstract

Purpose

General practitioners (GPs) are increasingly expected to deliver genetics services in daily patient care. Education in primary care genetics is considered suboptimal and in urgent need of revision and innovation.

Aim of the study was to prioritise topics for genetics education for general practice.

Methods

A Delphi consensus procedure consisting of three rounds was conducted. A purposively selected heterogeneous panel (n=18) of experts, comprising six practising GPs also engaged in research, five GP trainers, four clinical genetics professionals and three representatives of patient organisations, participated. Educational needs regarding genetics in general practice in terms of knowledge, skills and attitudes, were rated and ranked in a Top 10.

Results

The entire panel completed all three rounds. Kendall's coefficient of concordance indicated significant agreement regarding the top ten genetic educational needs ($P < 0.001$). "Recognising signals that are potentially indicative of a hereditary component of a disease" was rated highest, followed by "Evaluating indications for referral to a clinical genetics centre" and "Knowledge of the possibilities and limitations of genetic tests".

Conclusion

The priorities resulting from this study can inform the development of educational modules, including input for case-based education, to improve GP performance in genetic patient care.

Introduction

It has been argued that the greatest public health benefit of advances in understanding the human genome may be realized for common chronic diseases such as cardiovascular disease, diabetes mellitus, and cancer.¹ International attempts to integrate such knowledge into clinical practice are still in the early stages, and as a result, many questions surround the current state of this translation.¹⁻³ Physicians lack knowledge of genetics relevant for daily practice, lack oversight of genetic testing and concerns about privacy and discrimination, and report inadequacy to deliver genetic services.^{1,4} For genomics to have an effect on clinical practice that is comparable to its impact on research will require advances in the genomic literacy of health-care providers.⁵

In the age of genomics both *genetics of common disorders* and large scale applications in *screening* will become increasingly important, and primary care health workers will have to be prepared to discuss these issues with their clients. GPs may get more involved in preventive check-ups and develop a more flexible way to deal with patient's requests for genetic tests, in addition to the original role in an open access full time service for every patient.

Defining genetic core competences for non-genetic health care workers was considered prerequisite for implementing genetics education for general practice.^{1,3,6,7} Such education programmes should be based on an educational needs assessment of GPs referring to the three domains of educational activities: cognitive (*knowledge*), psychomotor (*skills*) and affective (*attitude*).

Recently, a focus group study among participants from a variety of disciplinary backgrounds explored the genetic educational needs of GPs in the Netherlands.⁸ The results showed an urgent need for a genetics curriculum for postgraduate and continuing general practice education. Four overarching themes were identified with regard to educational needs: genetics knowledge, family history, ethical dilemmas and the role of clinical genetics services. These themes clarified genetics in general practice with implications for education.

The aim of the present study was to obtain consensus on prioritisation of GPs' educational needs regarding genetics, as identified in focus groups and focused on "knowledge", "skills" and "attitudes". The results are aimed to inform the development of effective genetics education for GPs.

Materials and methods

We used a Delphi method to operationalize the findings from our earlier focus group study and to obtain consensus on the prioritization of topics for GP genetics education. The Delphi technique has been widely used and is an accepted method for gathering data and achieving consensus from respondents within their domain of expertise.^{9,10} The technique was mainly developed by Dalkey and Helmer in 1963 at the Rand Corporation in the 1950s.¹¹

A panel of experts emailed their responses to a questionnaire about GPs' educational needs to the researchers in three rounds. The responses were fed back anonymously to all panel members in order to share answers and arguments thereby enabling the participants to reflect on different views and modify their own.

Panel selection

Eighteen purposively selected experts from the Netherlands responded to an invitation to participate in the study sent to 24 experts (response rate 75%). Of the invited experts, three did not participate due to time constraints, and three did not respond at all. Recruitment was guided by the researchers' network, and a snowball method was used. *Through the authors' [researchers] network, work in general practice and clinical genetics (Centre for Community Genetics in the Netherlands) we were familiar with key persons eligible for recruitment in our expert panel.* Representatives from patient advocacy groups were asked whether they were interested in participating or could refer someone else. We established a heterogeneous panel of experts who participated anonymously: six practicing GPs, also involved in research, five GP trainers, four clinical genetics professionals (one genetic counselor, three clinical geneticists) and three representatives from patient advocacy groups (See table 1 "Characteristics of the participants in the Delphi study"). The participants were considered to represent a complete overview, from different perspectives, of the importance of genetics core competences for general practice and the need of genetics education in general practice, i.e. what is needed, what works and what does not work. Eleven experts (61%) were female, and the average age was 51.4 years (SD 9.1). Seven panelists also took part in our previous focus group study.

Table 1. Characteristics of the participants in the Delphi study

Type of Delphi subgroup	N	Female (N)	Mean age in years (SD)	Mean work experience in years (SD)	Professional background
General practitioners	11	6	52.0 (9.4)	17.0 (11.0)	general practitioner;
Clinical genetic professionals	4	3	54.5 (8.6)	n.a.	6 practicing and involved in research, 5 trainers 1 genetic counselor, 3 clinical geneticists individuals representing: public health organization, patient advocacy groups for rare conditions and genetics and reproduction advocacy groups
Patient/consumer advocacy group representatives	3	2	44.7 (7.6)	n.a.	
Total	18	11	51.4 (9.1)		

n.a. not available

The Delphi procedure

The initial questionnaire consisted of 29 topics describing GP educational needs. In order to arrive at these needs, we first transformed all previously identified educational needs within four overarching identified themes in focus groups into learning outcomes.⁸ We then refined the list based on the proposed learning outcomes from the suggested core competences for GPs in Europe.¹² Topics relating to three domains of primary care genetics were presented to the participants: “knowledge” (7 topics), “skills” (12 topics) and “attitudes” (10 topics).

The flowchart in Figure 1 shows the phases and the (anonymous) process of the three consecutive Delphi rounds. After analyzing the responses to each round (EJFH, LH and MW) and discussing them with the other researchers (SjvL, MCC, GJD and CvdV), the researchers reworked the responses into a new questionnaire. The Delphi study was conducted between December 2009 and March 2010. At the start of the study, all experts were asked to complete at least three Delphi rounds. They received 100 Euro upon completion of the whole procedure.

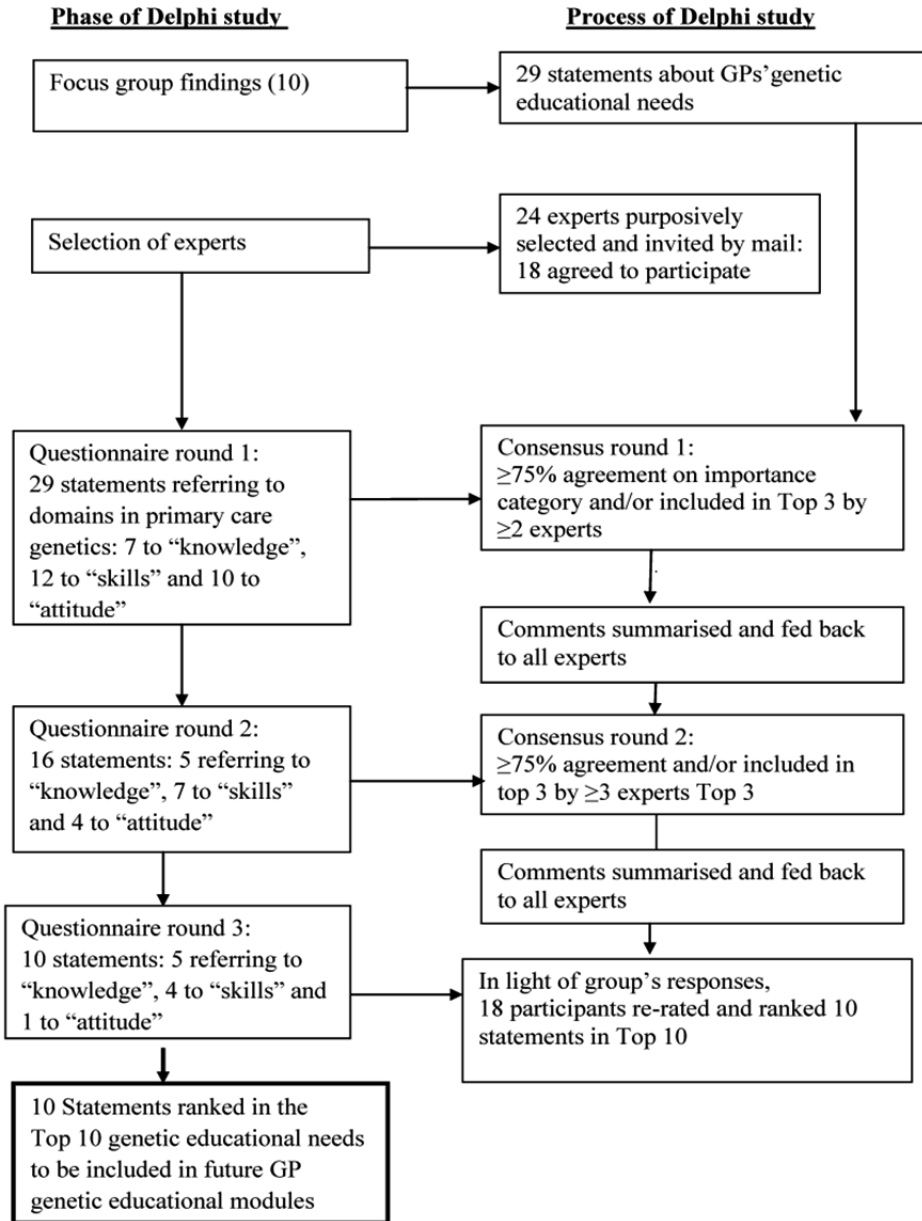


Figure 1. Flowchart Delphi consensus and prioritization procedure on GPs' genetic educational needs

Criteria for consensus

The research group discussed criteria for consensus on genetics education needs before the actual study was undertaken. The purpose of the study was to obtain consensus on and prioritize genetics educational needs in primary care. For this purpose, in the first two rounds, the experts were asked to prioritize the topics by ranking their importance and to give their Top 3 of topics for inclusion in educational modules. In the third and final round, ten items on which consensus was established in the first two rounds were judged. The definition of the inclusion criteria in a Top 3 in favour of a topic became more rigorous in the following second round, because in the first round at least two experts had to agree with a topic in the Top 3, whereas at least three experts had to agree in the second round. This will be explained in more detail below.

Round 1

In the first round, the experts were asked to rate the educational urgency for GPs of each of the 29 topics on a 7-point scale: "I believe that GPs have a strong need for education on [topic]", (totally disagree (1) - totally agree (7)). Experts were asked to comment on the topics they had given the lowest (1) and the highest (7), rating or about which they had doubts. The responses were converted into importance-based clusters of categories (low (1-2), medium (3-5) and high (6-7) importance category). The experts were also asked to indicate three (Top 3) of the 29 educational needs which they thought GPs most urgently wanted to be incorporated in an educational programme to be delivered within the next twelve months. In the first round, consensus in favour of a topic was defined as $\geq 75\%$ agreement regarding the "importance category" and/or inclusion among the Top 3 by at least two experts.

Round 2

The questionnaire for the second round consisted of the sixteen topics that had survived the first round. Some small adjustments were made to clarify topics which had been shown to be somewhat unclear. The inclusion criteria for the next round were more rigorous: $\geq 75\%$ agreement on importance category and/or inclusion in the Top 3 by at least three experts. An exception was made for topic #15 ("educating patients on the possibilities and limitations of genetic tests"), which despite 76% agreement was rejected in the second round, because the experts thought there was too much overlap with topic #4 ("knowledge of possibilities and limitations of genetic tests"), which did pass the round.

Round 3

For each topic the experts received a summary of the comments from the previous two rounds with the number of Top 3 ratings in round 2. The experts were asked to list their Top 10 of genetics educational needs for GPs, and Kendall's *W* (coefficient of concordance assessing agreement among raters) was computed for these rankings.

Results

After three Delphi rounds, 29 topics (table 2) were reduced to 10 priorities regarding genetic educational needs (table 3). All eighteen participants completed all three rounds. Response was high with many comments per round (table 2), indicating strong involvement of the experts. Of the 29 initial topics, ten remained after three rounds (text box 1 and 2). Of the 29 initial topics, three were modified after comments.

High agreement on a topic did not always imply high frequency in the Top 3. In fact, the reverse was true for some topics, which led to some unexpected results. Topic 1 ("Refreshing knowledge of basic genetic principles"), for example, showed only 39% agreement, but nevertheless made it through to the third round, because four experts placed it in their Top 3. In support of topic #1, some experts commented: "I think there are great differences [in competency] between younger and older GP generations" (active GP) and "without a proper knowledge basis, everything else will be futile". These comments underscored the notion improving genetics knowledge will pave the way for successful improvement of skills and attitudes for all GPs.

PRIORITISATION OF GENERAL PRACTITIONERS' GENETICS EDUCATION

Table 2. Number of comments (n), consensus (%) per round, frequency of inclusion in Top 3 (N) and final result (accepted/rejected) in terms of agreement or disagreement with the proposed topics

Topic	Round 1			Round 2			Result	In round
	n	%	N in Top 3	n	%	N in Top 3		
Knowledge								
1	18	39	4	18	39	4	Accepted	
2	17	53	1				Rejected	1
3	17	76	5	18	83	4	Accepted	
4	18	89	10	18	89	8	Accepted	
5	16	69	5	17	71	3	Accepted	
6	17	65	2	18	72	5	Accepted	
7	16	50	0				Rejected	1
Skills								
8	18	78	2	18	100	6	Accepted	
9	16	56	1				Rejected	1
10	17	71	4	18	50	3	Accepted	
11	16	75	3	17	71	7	Accepted	
12	18	61	1				Rejected	1
13	16	44	3	18	39	3	Accepted	
14	18	61	0				Rejected	1
15	18	61	2	17	76	2	Rejected	2
16	17	53	0				Rejected	1
17	16	56	2	18	61	0	Rejected	2
18	17	76	1	18	72	1	Rejected	2
Attitude								
19	18	56	0				Rejected	1
20	18	33	2	18	33	3	Accepted	
21	17	29	0				Rejected	1
22	16	50	0				Rejected	1
23	16	56	0				Rejected	1
24	18	61	1				Rejected	1
25	18	61	2	17	71	1	Rejected	2
26	17	71	2	18	56	1	Rejected	2
27	17	41	0				Rejected	1
28	18	67	0				Rejected	1
29	17	76	1	18	72	2	Rejected	2

Table 3. Overview of mean rank order1 (Top 10) and Kendall's coefficient of concordance assessing agreement among experts

Topic #	Mean rank order General	Mean rank order Active GPs	Mean rank order GP trainers	Mean rank order Clinical genetic professionals	Mean rank order Representatives patient organizations
11	2.9	3.0	4.4	1.5	2.3
8	3.1	3.7	3.8	1.8	2.7
4	3.2	4.7	1.2	4.3	2.3
6	4.6	5.3	4.4	5.0	2.7
3	5.2	4.9	5.6	4.5	6.0
1	6.1	5.3	6.6	7.5	5.0
5	6.6	6.0	6.0	7.5	7.0
10	7.3	6.6	5.0	10	8.7
13	8.0	7.5	8.6	7.3	9.0
20	8.1	8.0	9.2	5.8	9.3
N	18	6	5	4	3
Kendall's W	0.433	0.271	0.595	0.779	0.919
Chi-square ²	70.208	14.660	26.760	28.036	24.818
df	9	9	9	9	9
Sig	0.000	0.101	0.002	0.001	0.003

¹ The lower the score in mean rank order, results in higher ranking in the Top 10.

² Comparing score for topics within the group.

An example of a topic that was accepted in the first round (76% agreement, N=1 in Top 3) but rejected in the second round (72% agreement, N=1 in Top 3) is topic #18 ("Explaining the consequences of a genetic test for a patient and his or her family"). According to a Clinical genetic professional: "This task should be specifically assigned to the genetic counsellor. GPs should be able to generally evaluate whether a patient should be referred" and "The consequences [of genetic test results] are diverse. Generalization would be dangerous and might lead to misinformation. It seems therefore wiser for this kind of specific information to be delivered by a clinical genetics professional".

After round 2, there was consensus on ten topics, which increased for most after modification of the wording. In the end, it was not difficult to distinguish between accepted and rejected topics. The prioritized topics at the end of round 3 supports the development of educational modules with the main focus on skills and knowledge (Kendall's W=.43, $P<0.001$).

Text box 1. Accepted topics

Knowledge	
1	Knowledge of basic genetic principles (round 1) Refreshing knowledge of basic genetic principles (round 2 and 3)
3	Knowledge of the most important genetic disorders in the Netherlands (round1) Knowledge of the prevailing (most common) genetic disorders in the Netherlands (round 2 and 3)
4	Knowledge of the possibilities and limitations of genetic tests
5	Knowledge of the wide array of referral possibilities concerning genetics
6	Knowledge of the most important sources of genetic information
Skills	
8	Evaluating indications for referral to a clinical genetics centre
10	Discussing genetic risks with patients (risk communication)
11	Recognising signals potentially indicative of a hereditary component of a disease
13	Taking and interpreting a family history
Attitude	
20	Being aware of the fact that many symptoms/disorders can have a genetic component (round 1) Being aware of the possibility of a genetic/hereditary component of symptoms and disorders (round 2 and 3)

Although Kendall's *W* showed significant agreement among the respondents, there were also differences of opinion between subgroups of experts on different topics. For example participants from the active GP subgroup and Clinical genetic professionals subgroup commented differently on topic #10 ("Discussing genetic risks with patients (risk communication)"). Active GPs (mean rank order 6.6) commented "Risk communication is difficult, certainly in the case of genetic diseases" and "Risk communication is becoming more important, most GPs are not educated on this topic." Clinical genetic professionals however commented less supportive of adding this topic to the Top 10 of educational topics (mean rank order 10) "I think it depends on the [genetic] disease and the degree of difficulty. I prefer the GP to leave this up to the Clinical geneticist" and "If risk communication is meant as a means to support the patient in handling their genetic risk, this could be a GP's responsibility. However, if is meant the GP should be capable of calculating a certain genetic risk and discuss this with the patient, additional education would be necessary."

Relatively high agreement was found within the subgroup of GP trainers (Kendall's *W*=0.60, *P*=0.002), clinical genetics professionals (Kendall's *W*=0.78, *P*=0.001) and representatives of patient organisations (Kendall's *W*=0.92, *P*=0.003), whereas agreement was relatively low among practising GPs (Kendall's *W*=0.27, *P*=0.101).

CHAPTER 3

Text box 2. Rejected topics

Knowledge

- 2 Knowledge of different **hereditary patterns**
- 7 Knowledge of the **consequences** of genetic testing for obtaining a **mortgage and insurance**

Skills

- 9 **Discussing** with patients how to **cope** with (an increased **risk** for) a genetic disorder
- 12 **Explaining genetic information** in a way that is adapted to the patient's level of knowledge
- 14 Drawing and interpreting a **pedigree**
- 15 **Educating** patients on possibilities and limitations of **genetic tests**
- 16 **Explaining** the genetic aspects, except lifestyle, of **multifactorial disorders**
- 17 **Informing** parents about the possibilities and limitations of **prenatal and neonatal screening**
- 18 **Explaining** the possible **consequences** of a **genetic test** for a patient and his or her family

Attitude

- 19 **Recording** a family history in such a way that it can be easily retrieved
- 21 **Guiding** patients with genetic issues in a **non-directive** way
- 22 A medical practitioner's role in (actively) **suggesting the possibility** of having a genetic test
- 23 **Demarcating tasks** in the field of genetics in comparison with other caregivers
- 24 A GP's role in decisions about discussing (the chances of) a genetic disorder with the patient's **family**
- 25 **Offering support** to patients with (an increased risk for) a genetic disorder
- 26 **Dealing with** the choices relating to genetics, made by people from **different cultures**
- 27 Dealing with **sensitivities** surrounding genetic disorders in **families**
- 28 Informing patients about genetic **risks** in **consanguine** marriages
- 29 **Dealing with ethical dilemmas** in genetics

Discussion

Our study generated consensus on a Top 10 of prioritized topics for GPs' genetics education. The highest ranking topics were concerned with skill and knowledge competences: "Recognizing signals that can indicate a hereditary component of a disease", "Evaluating indications for referral to a clinical genetics center", and "Knowledge of the possibilities and limitations of genetic tests". These priorities could, in particular, be met by case-based education.

Strengths and limitations of the study

The Delphi procedure included eighteen selected experts, who completed all rounds. Despite the experts' differing backgrounds, it remains to be investigated if the results have relevance beyond the Dutch health care system, since the nature of the sample was drawn from this particular health care system. General practice in the Netherlands is an open access full-time service for every patient with any medical complaint, request, or question. The service includes a list system, implying that every person (with or without a disease) is on the list of one general practitioner (GP), thus guaranteeing optimal continuity of care. The GP handles more than 90% of all presented complaints and diseases. If genetic counseling as a primary care service is available in their particular region, the GP manages most referrals to this service as to most other primary care services and to all secondary care services. Therefore, the GP is the first to whom a patient will turn when he/she has questions on prevention and treatment of disease.

The results of this study are in line with some studies and differ from others with regard to the need for increased genetics knowledge in general practitioners.^{5, 12-16} This difference may be due to differences between health care systems. However, our previous focus group results are supported by the outcomes of the afore mentioned studies regarding deficiency in skills (e.g. taking a family history, referral to appropriate regional genetics services and non-directive counselling).^{8, 17, 18} It may be problematic for primary care providers to take appropriate steps in response to the perceived shift in the importance of genetics in primary care, such as taking enough time to discuss the family history or non-directive counselling.

Another possible weakness of our study is regression to the mean, although this is inherent to this consensus method: experts are inclined to adjust their opinions during a consensus process.¹⁵ Nevertheless, there was a high degree of agreement on the ten final topics, while nineteen topics were not accepted despite several adjustments. The procedure started with topics based on our earlier focus group research, and some experts were aware of these results although they were unaware of each other's identity.⁸ Although this may have biased their opinions, the validity of the focus group results was checked by comparing them with the results of the consensus procedure, a process commonly referred to as triangulation.¹⁹ Compared with the results of the focus groups, this study strengthened the prioritization of genetic educational topics for general practice. Also, the transparency of the way we dealt with comments and ratings and adjusted or rejected topics is expected to have improved the validity and reliability of the resulting consensus.

Comparison with existing literature

Our study resulted in a Top 10 of genetic educational topics in primary care through consensus and prioritization.^{1, 5, 7, 12, 13, 16, 20-22} The results also support the learning outcomes and core competences in genetics for non-genetic health care professionals as specified by genetic experts.⁶ A previous paper described the absence of genetics educational objectives for Dutch non-genetic health care providers.¹³ This Delphi study has laid a firm foundation, supported by experts' opinions, for the development of more appropriate genetics education for GPs.

In addition to the perceived inadequacy of primary care workers to integrate genetics in daily practice, Scheuner *et al.* identified deficiencies in primary care workers' basic genetic knowledge and ability to interpret familial patterns.¹ This is in line with our prioritized educational topics, which include knowledge of basic genetic principles, (the most common) genetic disorders and family history skills. Taylor and Edwards stated primary care should be encouraged to invest more time in family history data.²⁰ However, they also stressed identified barriers (e.g. time constraints) and the need to develop strategies to overcome difficulties preventing GPs from routinely obtaining family history information.²⁰ These barriers and strategies are still under construction, and may explain why topic #13 "Taking and interpreting a family history" ended ninth in the Top 10 list of genetic education priorities. In this study, we did not elaborate on these difficulties, but they should definitely be considered during the development of education modules concerning the integration of family history skills according to referral criteria.

Topic #8 ("Evaluating indications for referral to a clinical genetics centre") is similar to one of the priorities mentioned in Scheuner *et al.*'s systematic review, namely "referral guidelines would improve referral patterns", while (computerised) decision support might be helpful in familial risk assessment for common cancers (e.g. breast, ovarian and colon cancers) and would render many other genetics referrals more consistent with guidelines.¹ These results support the implementation of genetics education aimed at enhancing effective referral indications and options.

The results of this Delphi study differ from those of the GenEd study of 2004 by Calefato *et al.* and of previous focus groups.^{8, 12} Competences relating to attitudes received more attention in these studies, such as "ethical, legal and public health issues" and "psychosocial and counselling issues". This difference may be attributed to the fact the GenEd study was not limited to the Netherlands but encompassed five European countries with differing health care systems. In our study, the experts commented genetics education should first focus on "knowledge" before moving on to "attitudes". Some comments on this issue were rather ambivalent: "Attitude is not specific to genetics" and "A good attitude should be an intrinsic component of the GP's role". Thus, although it seems the

“attitude” domain is considered essential for GP genetics education (i.e. case based learning with medical ethical problems), effective implementation of genetics education may be jeopardized if too much attention is paid to this area.

The higher than expected number of topics in the Top 10 referring to “knowledge” (5 out of 7) exceeded the number of “skill” topics (4 out of 11). This may be explained by the experts’ perceptions genetic knowledge should be brought up to date before related skills can be learned. This unexpected result needs further research and probably explains a relatively low agreement among practicing GPs since some may find their genetic knowledge or skills sufficient and others not.

Implications for future research and clinical practice

Unless a scientific, logistical and ethical framework for the appropriate and effective use of genomic information is in place, the primary care workforce is unlikely to be adequately prepared to provide such information in general practice.^{1, 3, 5-7, 12, 20, 22-24} If GPs stay genetically uneducated and therefore incompetent related to the use of genomic information in general practice, individual genetic medical care provided will likely be unhelpful and possibly even harmful. We believe the results of this study should be used in the near future to guide the implementation of genetics education in the Netherlands and perhaps even internationally. Although the majority of the issues investigated cover genetics-related knowledge, skills and attitudes essential for every medical care provider, further studies will have to determine whether the results are relevant to other medical specialties as well.

We are currently working on developing genetics education for general practitioners in collaboration with the Dutch GP society. This entails a written educational module aiming for improving genetics knowledge, an informative website specifically aiming on genetics in general practice in the Netherlands and a live Continuing Professional Development module aiming for improving genetic skills and attitude. Hopefully this will improve (genetic) medical care in the Netherlands and will meet the needs expressed by GPs and experts in our previous work and the Top 10 of genetic educational topics presented in this paper.⁸

Preparation of health care providers for the future of genetic medicine, with personalized genomic information and education, will lead to effective use of genetics in daily primary care.

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PRIORITISATION OF GENERAL PRACTITIONERS' GENETICS EDUCATION

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PART II

Development and evaluation of genetic educational modules for general practitioners



Chapter 4

Sustained effects of online genetics education: a randomized controlled trial on oncogenetics

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Abstract

Medical professionals are increasingly expected to deliver genetic services in daily patient care. However, genetics education is considered to be suboptimal and in urgent need of revision and innovation. We designed a Genetics e-learning Continuing Professional Development (CPD) module aimed at improving general practitioners' (GPs') knowledge about oncogenetics, and we conducted a randomized controlled trial to evaluate the outcomes at the first two levels of the Kirkpatrick framework (satisfaction, learning and behavior). Between September 2011 and March 2012, a parallel-group, pre- and post-retention (6-month follow-up) controlled group intervention trial was conducted, with repeated measurements using validated questionnaires. Eighty Dutch GP volunteers were randomly assigned to the intervention or the control group. Satisfaction with the module was high, with the three item's scores in the range 4.1–4.3 (5-point scale) and a global score of 7.9 (10-point scale). Knowledge gains post test and at retention test were 0.055 ($P<0.05$) and 0.079 ($P<0.01$), respectively, with moderate effect sizes (0.27 and 0.31, respectively). The participants appreciated applicability in daily practice of knowledge aspects (item scores 3.3–3.8, five-point scale), but scores on self-reported identification of disease, referral to a specialist and knowledge about the possibilities/limitations of genetic testing were near neutral (2.7–2.8, five-point scale). The Genetics e-learning CPD module proved to be a feasible, satisfactory and clinically applicable method to improve oncogenetics knowledge. The educational effects can inform further development of online genetics modules aimed at improving physicians' genetics knowledge and could potentially be relevant internationally and across a wider range of potential audiences.

Trial registration trialregister.nl Identifier: NTR3322

Introduction

Although the dramatic surge in the volume and potential applicability of genetics knowledge in medical care is set to continue, there appears to be a marked underuse of this knowledge, in particular among primary-care physicians.^{1,2} This is probably largely because of physicians lacking sufficient knowledge about genetics for daily practice and failing to keep up with recent developments in genetic testing.³⁻⁹ It is therefore not surprising that there are inadequacies reported in the delivery of genetic services.¹⁰ In view of the ongoing rapid developments in genetics research, it is important that genomic literacy among healthcare providers be enhanced to ensure optimal translation to health-care delivery of research on common complex diseases, including familial cancers, such as breast and colon cancer. Previous studies have shown that as far as genetics is concerned non-genetic healthcare workers require not only education but also clear guidelines and definitions of their responsibilities.¹¹⁻¹³

Continuing Professional Development (CPD) seems the obvious vehicle for remedying deficiencies in practicing physicians' genetics knowledge and skills. In addition, e-learning appears to offer a cost-effective and time-efficient method of keeping physicians informed of new developments. In CPD, e-learning and other modalities (printed educational materials and face-to-face activities) are widely used and have been shown to be equally effective.¹⁴⁻¹⁶ In 2010, the Accreditation Council for Continuing Medical Education in the United States reported online (enduring materials) activities accounted for 28% of all CPD activities, with 4.6 million US physician participants (activity attendees). Online CPD (eCPD) activities represent by far the most popular form of CPD in the United States (40% of all CPD credits).^{17,18} Between 2003 and 2010, the number of physicians receiving credit for online CPD increased by 800%, compared with an 89% increase for all CPD programs.¹⁸ These findings and reported improvements in knowledge and clinical decision making following online case vignette courses suggest that online educational activities can offer 'a searchable, credible, available on-demand, high-impact source for physicians.'^{19,20} So far, however, there is a paucity of research into optimizing eCPD and its relevance to everyday primary care, with two small studies evaluating eCPD being methodologically weak and of uncertain clinical significance.^{16,21} Nevertheless, considering that eCPD is easy to deliver on a large scale and is relatively inexpensive to develop, it is important to determine the feasibility and effectiveness of accredited Genetics eCPD (G-eCPD) in keeping physicians abreast of new genetics developments affecting the delivery of (preventive) cancer care. We therefore conducted a randomized controlled trial (RCT) to investigate the effectiveness and applicability in daily practice of an oncogenetics eCPD module. We aimed to measure the educational outcomes of the module at the first two

levels of Kirkpatrick's four-level framework for evaluating educational outcomes (satisfaction, knowledge and knowledge retention, behavior, and actual practice performance and results).²²⁻²⁴ We investigated participant satisfaction (level 1), participants' gain in knowledge about oncogenetics and participants' self-reported application of newly acquired oncogenetics knowledge in daily practice (level 2). To our knowledge, the online module we developed was the first of its kind to be based on assessment of primary-care physicians' educational needs and priorities in relation to genetics knowledge and on core competencies for genetics education. Primary-care physicians' gains in knowledge about oncogenetics in family medicine are expected to improve referral strategies to clinical genetics services and adherence to clinical guidelines. This would increase the feasibility of identification of familial forms of cancer by primary-care physicians, which in turn would improve risk stratification in clinical practice and ultimately reduce morbidity and mortality.

Materials and methods

Experimental design

We designed an RCT to assess the outcomes of a G-eCPD module for primary-care physicians at the first two of Kirkpatrick's levels of educational outcomes. The intervention consisted of an oncogenetics eCPD module written by The Dutch College of General Practitioners (NHG; FJ) and the first author of this manuscript (EJFH). Two clinical geneticists and an educationalist (SvL) supported the development of the module. The trial was conducted between September 2011 and March 2012. To control for external effects, a control group was included and, to detect any changes over time, educational outcomes were measured by a pre- and post-test and a retention (6 months) evaluation trial. The study protocol was approved by the ethical review board of the Netherlands Association for Medical Education and the medical ethical review boards of the Maastricht University Medical Center and the VU University Medical Center in The Netherlands. Participation was voluntary and participants gave written informed consent before the start of the trial.

Study participants

General practitioners (GPs) working full time or part time in family practice were eligible for inclusion in the study. Out of 600 Dutch GPs who met the inclusion criterion according to the NHG, 80 responded to participate in the study. Two groups of 40 participants were estimated to be sufficient to detect a medium-to-large effect with a power of 90% and a significance level of 5%.²⁵ Figure 1 shows the randomization scheme and participation flow. Participants were recruited by

online mailings, informing them about the study and requesting informed consent. CPD accreditation points were awarded for completion of the module, the online knowledge tests and the online questionnaires. A book on genetics in general practice or a book voucher of equal value was offered as an extra incentive.

For sampling and random assignment of participants to the intervention and control group, a pseudo random number generator was used for which the operator was not otherwise involved in the intervention or data analysis. The results of the randomization were communicated to the NHG but not to the researchers.

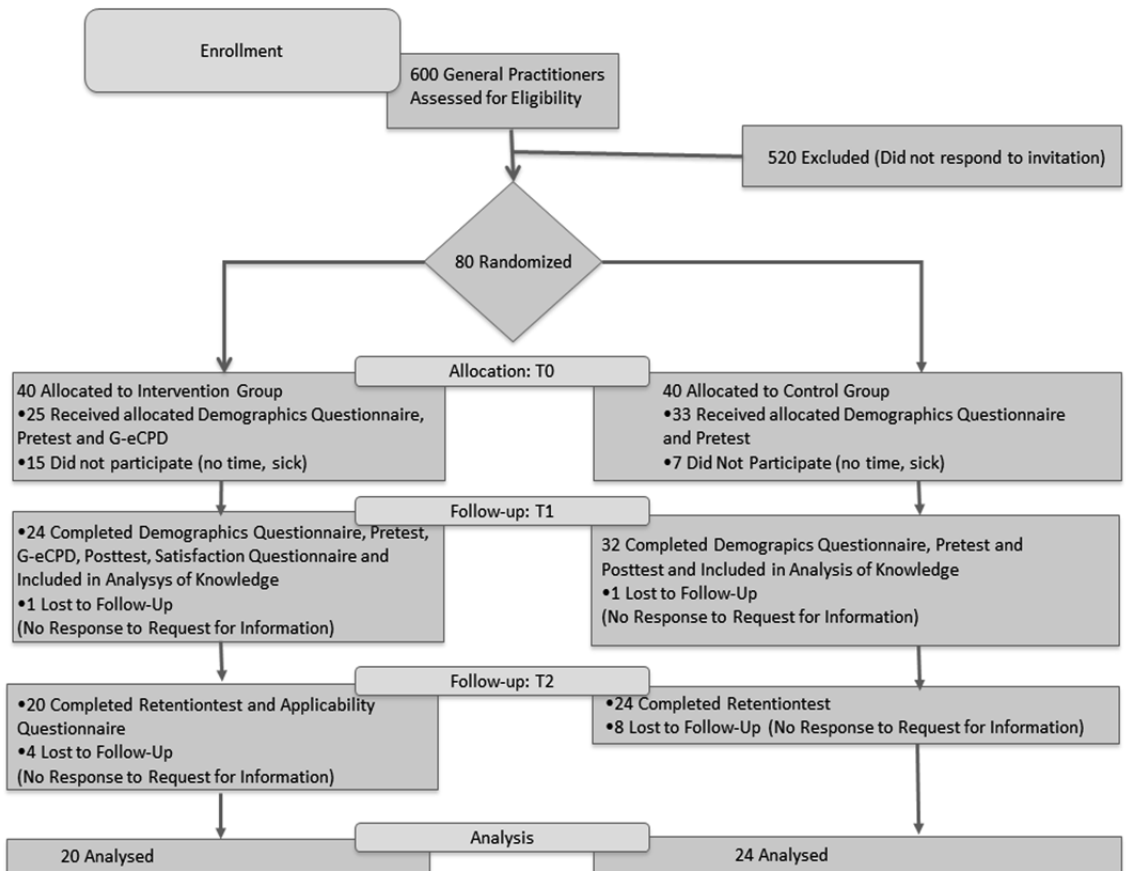


Figure 1. Randomization Scheme and Participation Flow of the online Genetics Continuing Professional Development (G-eCPD) study groups

Educational design and content

The module contained several didactic components with multimedia presentations, interactive cases with feedback, enabling tools (for referral, family history and online information searches) and other resources, such as step-by-step clinical practice guides and employable NHG guidelines. Common forms of oncogenetic diseases were presented in patient cases. The contents of the module were designed to include 10 items prioritized in a multidisciplinary Delphi study on core competencies of health professionals endorsed by the NHG.^{13,26} A multidisciplinary team consisting of FJ and EJFH who wrote the module, educationalists SvL and CvdV, and a clinical geneticist familiar with genetics in primary care, constructed the module and selected practical genetics information on common forms of cancer (such as breast and ovarian cancer, and colon cancer).

The aim of this module was to provide physicians with sufficient knowledge and skills to:

- Identify patients with an inherited predisposition to cancer.
- Draw a family tree as a tool for identifying patients at risk for hereditary cancer.
- Describe the most common types of hereditary cancer (ie, breast cancer and colon cancer) and the likely genetic mutations involved.
- Apply oncogenetics guidelines in identifying patients for whom referral is indicated or not, and find relevant information online.
- Explain the possibilities and limitations of oncogenetic testing.
- Discuss with patients periodic examinations and risk-reducing surgical options that are available to patients with hereditary cancer.

The online module provided access to didactic presentations, such as ‘a clinical genetic cancer consultation in daily practice’; interactive cases on breast cancer due to *BRCA* mutations and on colon cancer (eg, Lynch syndrome) due to *APC*/mismatch-repair gene mutations; and enabling tools, such as information about regional possibilities for referral and consultation. The educational sections were presented in the style the NHG usually uses when presenting online CPD modules for optimal recognition. The participants were free to revisit program sections as desired. The module was designed to enable completion within 2h. The online administration tools afforded monitoring of participant progress, including test and survey completion.

Data collection

Data were collected using four online instruments: online-only material contents section eTable 1, Questions and Answer Options of the Multiple-choice Knowledge Test; eTable 2, Satisfaction Questionnaire; eTable 3, Applicability Questionnaire;

and eTable 4, Demographics and Practice Characteristics Questionnaire. The test questions were based on a validated questionnaire that identified the genetic learning objectives and covered the oncogenetics topics of the G-eCPD.⁶ The instruments were developed and validated in collaboration with content experts (experts in daily clinical genetics, a GP, and an expert in education and questionnaire development) and pilot tested.

The knowledge test contained 20 multiple-choice items. Oncogenetics knowledge was measured as the proportion of correct answers. The satisfaction questionnaire contained 3 items (5-point scale: 1=completely disagree; 5=completely agree) (In the questionnaires, the coding was directed oppositely (1=completely agree, 5=completely disagree) in accordance with the conventions of the NHG. For ease of interpretation in the current paper, the ratings were recoded to comply with international conventions (1=completely disagree, 5=completely agree), related to different aspects of satisfaction, a global grading of the module on a 10-point scale (1=no value; 5=insufficient; 6=sufficient; 8=good; 10=excellent) and a question about the amount of time spent doing the module. The applicability questionnaire contained six five-point Likert scale items about the application of the newly acquired knowledge in daily practice and a multiple-choice question about the application frequency. The demographic survey asked about participants' general characteristics (eTable 4).

The interventions and measurements were conducted at and between time points T0, T1 and T2 (table 1). At T0, the intervention and the control group completed the demographics survey and the knowledge test. Between T0 and T1, the intervention group undertook the G-eCPD module, whereas the control group were free to spend the 2-h break any way they wanted, except by doing the module. At T1, both the intervention and the control group again completed the knowledge test in which the question and answer options had been randomly changed to correct for recall bias, and the intervention group completed the satisfaction questionnaire also. At T2, 6 months after T1, retention of knowledge was measured by administering the knowledge test to both groups, whereas the intervention group also completed the applicability questionnaire. After T2, in order to stimulate compliance the online module was made available to the control group, which also completed the satisfaction questionnaire.

Analysis

Knowledge gain immediately after the module was examined using regression analysis, with Knowledge Test Score at T1 (ScoreT1) as the dependent variable, test score at T0 (ScoreT0) as predictor and Training (0=no, control group; 1=yes, intervention group) as the indicator variable. In order to allow for different slopes for the relation ScoreT1 and ScoreT0, the interaction of Training and ScoreT0 (TrainingxScoreT0) was also included as an independent variable. To improve the

interpretation and numerical stability, the independent variable ScoreT0 was centered, and the resulting variable ScoreT0c, equal to $\text{ScoreT0} - \text{Mean}(\text{ScoreT0})$, was used in the analysis. In a similar analysis, retention of knowledge was analyzed using ScoreT2 as a dependent variable. The regression coefficient corresponding to Training represents the net gain in knowledge (proportion correct) after the intervention, and the standardized regression coefficient indicates the effect size. According to Cohen's categorization, 0.1, 0.3 and 0.5 indicate small, moderate and large effect sizes, respectively.²⁵ The final model included only predictors with a statistically significant contribution ($P < 0.05$).

The mean test scores and corresponding 95% confidence intervals (95% CIs) for the two groups at T0, T1 and T2 were calculated, and Student's *t*-tests were conducted to determine between-group differences. To determine satisfaction, mean scores, 95% CIs and SD were calculated for the pooled data of the intervention and control groups. For the applicability data (intervention group only), the same statistics were calculated. All statistical analyses were performed using the statistical package SPSS version 19 (SPSS, Chicago, IL, USA).

Table 1. Time table of the RCT

Instrument	Group	Time		
		0	2 h	6 months
		Pre-test (T0)	Post-test (T1)	Retention test (T2)
Knowledge test	Intervention	X ^a	Online oncogenetics training	X
	Control	X	X	X
Satisfaction questionnaire	Intervention		X	
Applicability questionnaire	Intervention			X
Demographics questionnaire	Intervention & Control	X		

Abbreviation: RCT, randomized controlled trial.

Time table of the RCT showing scheduled measurement times (columns 3–6), instruments (column 1) and measurements made (indicated with X in columns 3–6) in the intervention and control groups (column 2).

^a *Measurement made with the instrument indicated in column 1 in the group indicated in column 2.*

Results

Randomization and dropout comparisons

Of the total of 80 participating physicians (40 intervention group and 40 control group), 44 (20 intervention, 24 control group) completed all the learning activities, knowledge tests and questionnaires (Figure 1). Thirty-six participants were lost to follow-up; 22 did not participate because of time limitation or illness, and 14 did not respond to requests for information.

Participant characteristics

There were no significant differences between intervention and control group in age, gender, years of experience in primary care, type of practice and practice situation (eTable 5).

Knowledge

Figure 2 presents the results of the pre-test, post-test and retention test. The between-group difference was indifferent or in favor of the intervention group, starting from 0.034 (Student's *t*-test, non-significant, $P=0.34$) at T0, and increasing to 0.072 ($P<0.05$) and 0.084 ($P<0.05$) at T1 and T2, respectively. More precise estimations of knowledge gain were obtained by the regression analysis controlling for between-group differences in ScoreT0 and allowing for an interaction effect of intervention (group) and ScoreT0. The first numerical row of table 2 shows the regression results for ScoreT1. As the contribution of the interaction was found to be statistically non-significant, the interaction was excluded from the final model, leaving the intercept (Constant), and two additional independent variables (see second row) ScoreT0c, the centered version of the pre-test score (mean ScoreT0=0.66) and the indicator variable Training (0=no, control; 1=yes, intervention). The resulting regression coefficient, the corresponding 95% confidence interval (95% CI; low and high boundary) and the standardized regression coefficient are presented. Value $B=0.70$ for Constant indicates the expected proportion correct in the post-test for a participant in the control group with a ScoreT0 equal to the mean value (0.66). The regression coefficient (0.51) for ScoreT0c indicates the slope of the corresponding regression line for the control group, which was found to be statistically significant as is indicated. The effect of the intervention was found to be statistically significant, amounting to 0.055 on the proportion correct scale; the corresponding value for the standardized regression coefficient was 0.27, indicating an almost moderate effect size. The analysis for ScoreT2 also showed a non-significant interaction and in the final model the intervention effect was found to be 0.079 (standard regression coefficient of 0.31, moderate effect size), implying a further increase of the knowledge effect by 0.024 at 6 months after the intervention.

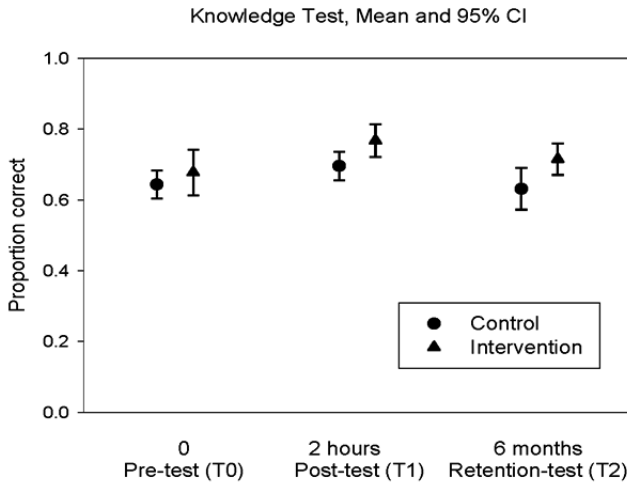


Figure 2. Knowledge test scores (mean and 95% CI) for the control group (circle) and the intervention group (triangle) at T0, T1 and T2, corresponding to pre-, post- and retention measurement, respectively.

Table 2. Effect of the oncogenetics training (G-eCPD module) on the performance of GPs. Regression results are shown for immediate gain of performance (ScoreT1) and retention of performance (ScoreT2), using the pretest score (ScoreT0) as a covariate and the control group score as a reference.

Dependent Variable	Independent Variables								
	Constant	Score T0			Training				
	regression coefficient	regression coefficient	95% Conf Int	standardized regression coefficient	Value	95% Conf Int	standardized regression coefficient	Value	95% Conf Int
		value	low high	value	low high	low high	value	low high	low high
ScoreT1	0.70***	0.51***	0.30 0.71	0.57	0.055*	0.006 0.103	0.27		
ScoreT2	0.64***	0.68***	0.43 0.93	0.62	0.079**	0.022 0.136	0.31		

Abbreviations: G-eCPD, Genetic online Continuing Professional Development; CI, confidence interval. Regression results are shown for immediate gain of performance (ScoreT1) and retention of performance (ScoreT2), using the pre-test score (ScoreT0c) as a covariate and the control group score as a reference. *P<0.05; **P<0.01; ***P<0.001.***

Satisfaction and applicability

Table 3 shows the results for satisfaction and applicability. The four satisfaction items had scores of at least 4.1 (95% CI lower boundary not <3.7) and a mean global score of 7.9 (95% CI lower boundary=7.5). The average time spent on the module (124 min) was very close to the recommended time. The applicability scores were more diverse: neutral scores (2.7–2.9) were obtained for self-reported recognition of disease, referral to a specialist and knowledge of possibilities/limitations of genetic testing. The scores on increased knowledge about genetic diseases, concepts and information sources were more positive (3.3–3.8). More than 90% of participants indicated applying newly acquired knowledge at least once a month, and 5% indicated a frequency of at least once a week. No participant reported daily application, and 5% indicated not having encountered any genetic problem in their practice in the last 6 months.

Table 3. Satisfaction (intervention+control; N=44) and self-reported applicability (intervention only; N=20) as a result of training with the G-eCPD module.

Variable	Mean	95% CI		SD
		Low	High	
<i>Satisfaction</i>				
Would recommend the module to a colleague ¹	4.3	3.9	4.6	1.1
Content of the module is relevant for a GP	4.2	3.9	4.6	1.1
Content of the knowledge test is relevant for a GP	4.1	3.7	4.4	1.0
Global score (1-10)	7.9	7.5	8.3	1.3
Time spent on the module (minutes)	124	115	132	27
<i>Applicability</i>				
Recognize patient with genetic disease sooner	2.8	2.3	3.3	.98
Sooner refer to or discuss with a genetic specialist	2.7	2.0	3.3	1.2
More knowledge of possibilities/limitations of genetic tests	2.9	2.4	3.4	.96
More knowledge of genetic diseases	3.6	3.1	4.1	.98
More knowledge of basic genetic concepts	3.3	2.8	3.8	.96
More knowledge of genetic information sources	3.8	3.4	4.3	.88
<i>Proportion of trainees applying the learned knowledge (%)</i>				
Daily	0			
Weekly	5			
Monthly	90			
Not (do not meet any genetic problems in our practice)	5			

Abbreviations: CI, confidence interval; G-eCPD, Genetic online Continuing Professional Development; GP, general practitioner. ¹ if not indicated otherwise results refer to scores of 5-point Likert scale items (1: completely disagree, 5: completely agree)

Discussion

To our knowledge, our study is the first to evaluate the outcomes of an online genetics CPD module at the first two levels of Kirkpatrick's framework, taking oncogenetics as an example. The results indicate that presenting a case-based oncogenetics module in an accessible online learning environment can result in sustained improvement of genetics knowledge for daily medical practice. Other topics, such as cardiogenetics (ie, long QT syndrome or hypertrophic cardiomyopathy) or diabetes (ie, maturity-onset diabetes of the young), could also be trained in this framework. The findings in the current study indicate that this approach may help to transfer urgently needed genetics knowledge on a broad array of issues, both in primary and secondary care. The participants were satisfied with the module and indicated that they actually applied their newly acquired knowledge in daily practice. However, self-reported applicability aspects focused on practice received neutral scores. This seems to indicate the G-eCPD mainly improved genetics knowledge rather than skills. A live training on oncogenetics may put more emphasis on these performance-oriented aspects reflected in increased consultation skills (ie, recognizing patient with genetic disease, how to refer to a Clinical Genetics center or to be able to explain possibilities/limitations of genetic tests).²⁷ Of course, the evaluation of the G-eCPD module should be an ongoing process, which can sustainably help to improve effectiveness. These findings are encouraging for future work in this challenging area of education.

The results indicate that significant knowledge gains of moderate effect size persisted for 6 months after the 2-h educational intervention. This is consistent with reports in the literature that most educational interventions lead to modest-to-moderate improvements in health care.²⁸ In addition to knowledge gains, the module showed relatively good cost effectiveness in terms of both finance and time, and it seems likely that it could easily reach large groups of physicians and possibly medical students as well.

Limitations

A limitation is the fairly large number of non-responders. Selection bias could have been caused by interested GPs who voluntarily participated and received financial incentives. Similar baseline characteristics of the two groups and comparability of the 55% participation rate to those reported for postal surveys among GPs (60% response rate) however, indicate that the participants were representative of GPs likely to attend oncogenetic training in the future.^{29,30}

It is possible that participating in the oncogenetics training might become a mandatory part of training for all GPs. Participants in the control group had to wait for training content, possibly causing resistance to finish all measurements. No specif-

ic reasons, such as fairly long duration of the study for drop out were reported, rather than general attributes (no time and sickness). It is therefore unlikely that self-selection in dropout negatively impacted the validity of the results. Although there were no significant differences in participant characteristics between intervention and control group, the physicians in our study appeared to be more number of women, younger and less experienced, compared with the general profile of Dutch GPs.³¹ This possibly reflects extra interest in genetics and/or using online learning modules by young woman GPs who recently graduated. Whether the results can be generalized within and beyond the Dutch healthcare system needs further investigation.

Although the results show substantive knowledge gains, it might be argued that we did not compare the online module to any other intervention or more traditional methods, such as paper format or live CPD modules. In education evaluation studies published, there may be publication bias: a wide variety of Internet-based interventions show effectiveness in medical education, perhaps leaving negative studies unpublished.³² Given recent rapid developments in genetics, there is not enough staff to provide nationwide traditional education activities. A meta-analysis suggested that Internet-based instruction would be similarly effective to non-Internet interventions. Moreover, Internet-based learning is associated with large positive effects compared with no intervention at all.³² Our study could therefore be seen as a proof-of-concept evaluation study and further research will be necessary to confirm comparable effectiveness on sustained knowledge.

Although our evaluation of the educational outcomes of the G-eCPD module by the questionnaire on the application of new knowledge in daily practice (eTable 6) closely approached Kirkpatrick's third level, assessment by observation of actual behavior was absent. We are currently undertaking a study to determine changes in referrals to Clinical Genetics centers after attending the G-eCPD module. Another study we have planned uses standardized patients to evaluate the effectiveness of face-to-face oncogenetics modules in terms of behavioral changes (level 3), such as family history taking and recognizing the need for referrals to the department of Clinical Genetics.²⁷ By applying an effective framework for genetics education and measuring outcome of education on various levels of Kirkpatrick, we might be able to initiate a change in organization (approaching level 4) and find barriers to implementation of genetics education.³³ Despite the limitations of the current study, however, the results suggest that the module presents a promising innovative educational approach in CPD for health professionals. Despite the obvious importance of evaluation at higher Kirkpatrick levels, the results we found for lower outcome levels are also important to build a solid basis for an advanced impact.

Generalizability

The results of the present study may contribute to the development of genetics educational programs, and online CPD in particular. Online modules once created could potentially reach a large group of physicians in primary and secondary care (non-clinical geneticists, such as oncologists, cardiologists, pediatricians, etc), and across large geographical areas. In addition, possibly those in nursing professions, medical school students and those attending biology classes could benefit from this framework for online genetics education. Costs are likely to be less than multiple face-to-face sessions; however, the time and expertise to create an effective tool is not insignificant.³²

General practice in the Netherlands is an open-access full-time service, available to all patients with any medical complaint or question. As, under the Dutch system, the entire population, irrespective of the presence of disease, is on the list of a GP, optimal continuity of care is guaranteed. If genetic counseling is available in the region, the GP manages most referrals to this service for healthy family members of cancer patients. The GP is therefore likely the first healthcare professional to whom a patient will turn with questions about genetics.

It is also important to consider whether the current results can be generalized to future effects. Obviously, the results are representative for those GPs who participated in the trial on a voluntary basis. It is reasonable to assume that the participants are representative of the group of GPs who, in the future, would be willing to make use of online CPD modules. In other words, the results seem to be generalizable to future users of online CPD modules.

Voluntary participation may have led to self-selection of participants with a special interest in genetics or in clinical leadership qualities. Furthermore, the online module may be made mandatory for all Dutch GPs in the near future. However, it seems plausible that accidental factors, such as time and health problems, rather than specific attributes of participants were responsible for participant dropout. However, specific attributes should be investigated further, for there was a relative high dropout rate (30%).

Studies have reported satisfaction and knowledge gains as a result of online modules on other topics and have suggested that course outcomes may benefit when a course is designed in accordance with a prior educational needs assessment.^{15,19,34-36} The advantages of online CPD have also been broadly discussed and supported.³⁷ Various reviews, however, have pointed to heightened effects on physician behavior of multiple interventions compared with single episodic interventions. Multifaceted interventions can tackle several common barriers to change and this combined operation may ultimately lead to improved practice performance. This aspect deserves further study.

Interpretation

Our online oncogenetics module proved to be a satisfactory and feasible method to achieve urgently needed knowledge improvement in a rapidly evolving field. Web-based genetics education can be a valuable tool to provide physicians, in general, with applicable genomics knowledge, and the long-term educational effects show great promise with respect to practical and strategy implications.^{2,38} The results suggest that relatively simple and low-cost online educational activities can have a pivotal role in urgently needed genetic health-care improvement.

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Online-only material contents

eTable 1. Questions and answer options of the multiple-choice knowledge test¹

-
- 1) In an autosomal-dominant inheritance pattern, who can pass on the disorder?
- Men
 - Women
 - Men and women
-

- 2) In which category do the most common disorders with a genetic component fall?
- Monogenetic
 - Chromosomal
 - Multifactorial
-

Case Study: Ms. van Aalst

Ms. van Aalst comes to your practice. Her father's sister was diagnosed with breast cancer at age 40 and died of it. Her daughter (Ms. van Aalst's cousin) recently received the same diagnosis at age 35. Answer questions 3, 4 & 5 using Case Study: Ms. van Aalst.

- 3) What is the degree of the relationship between Ms. van Aalst and the sister of her father: 1st, 2nd, 3rd or 4th?
- The sister of the father is a first-degree relative of Ms. van Aalst.
 - The sister of the father is a second-degree relative of Ms. van Aalst.
 - The sister of the father is a third-degree relative of Ms. van Aalst.
 - The sister of the father is a fourth-degree relative of Ms. van Aalst.
- 4) Which possible influence does breast cancer in the family of the father have on the risk of Ms. van Aalst for inherited breast cancer?
- Increased
 - Decreased
 - None
- 5) Is there an indication to refer Ms. van Aalst to a clinical genetics department based on the available information?
- No

Yes

Case Study: Ms. Brederode

Ms. Brederode comes to your practice. The father of her mother was diagnosed with colorectal cancer and died of it shortly after diagnosis, at the age of 50. The sister of her mother was diagnosed with colorectal cancer at age 39 and died of it a year later. Ms. Brederode is 30 years old and has two children. Answer question 6 using Case Study: Ms. Brederode.

- 6) Is there an indication to refer Ms. Brederode to a clinical genetics department based on the available information *according to CBO-guidelines*?
- There is an indication to refer Ms. Brederode to a clinical genetics department
 - There is no indication to refer Ms. Brederode to a clinical genetics department

A decision cannot be made based on the available information.

¹The order of the questions and possible answers was changed from T0 to T1 to T2.

-
- 7) HNPCC (or Lynch syndrome) is form of inherited colon cancer in which one gene plays a great role, i.e. it is a monogenetic subtype. Which portion of colon cancer cases involves a recognizable monogenetic subtype?
- Less than 1%
 - Approximately 5%
 - Approximately 30%
 - Approximately 50%
- 8) An increased chance of colon cancer caused by one gene mutation exists in other disorders as well. In which of the following disorders is that most likely?
- tuberous sclerosis
 - adenomatous polyposis coli
 - retinoblastoma
- multiple endocrine neoplasia
-

Case Study: Ms. Crynen

Ms. Crynen (28) has received test results from the department of clinical genetics indicating that she has a BRCA2 gene mutation. She considers her family to be complete. Ms. Crynen is now considering her options, such as breast cancer screening, mastectomy, oophorectomy and hormonal treatment. Answer questions 10, 11 & 12 based on Case Study: Ms. Crynen.

- 9) Which of the following breast examinations is the most sensitive in young BRCA2 patients (≤ 30)?
- Palpation by an experienced surgeon
 - Mammogram
 - MRI
 - Sonogram
- 10) And if Ms. Crynen had been 40 years old, which breast exam would be the most sensitive:
- Palpation by an experienced surgeon + MRI
 - Mammogram + MRI
 - Sonogram + MRI
 - CT + MRI
- 11) How should the Family Physician approach the options mentioned by Ms. Crynen?
- The FP should discourage breast cancer screening and encourage postmenopausal oophorectomy.
 - The FP should encourage bilateral mastectomy and premenopausal oophorectomy.
 - The FP should encourage premenopausal oophorectomy.
- 12) Ms. Crynen is considering prophylactic oophorectomy. BRCA2 patients have the following prognosis:
- Following the oophorectomy there will be no menopausal symptoms.
 - Following the oophorectomy there will be menopausal symptoms, which can be managed with non-hormonal treatment.

Hormonal treatment to limit menopausal symptoms should follow the oophorectomy.

- 13) In comparison to hereditary cancer, sporadic cancer usually develops at an:
- earlier age
 - later age
-
- 14) Which website presents the general guidelines for referrals to clinical geneticists, for advice related to inherited forms of cancer?
- www.erfelijkheid.nl
 - www.oncoline.nl
 - www.kankerrichtlijn.nl
 - www.erfelijkkekanker.nl
-

-
- 15) Multiple patients in the same family are diagnosed with colorectal cancer at a young age (age ≤ 50). Many of these patients have polyps. In the majority of cases, this indicates a mutation in:
- One of the genes that can cause Lynch syndrome (aka HNPCC)
 - The *APC* gene that can cause FAP
 - The BRCA1/ BRCA2 gene
-
- 16) Multiple patients in the same family are diagnosed with colorectal cancer at a young age (age ≤ 50). These patients have few or no polyps. In the majority of cases, this indicates a mutation in:
- One of the genes that can cause Lynch syndrome (aka HNPCC)
 - The *APC* gene that can cause FAP
 - The BRCA1/ BRCA2 gene
-
- 17) Multiple patients in the same family are diagnosed with endometrial cancer. When a genetic predisposition such as this exists, it indicates a mutation in:
- One of the genes that can cause Lynch syndrome (aka HNPCC)
 - The *APC* gene that can cause FAP
 - The BRCA1/ BRCA2 gene
-
- 18) Patients can screen themselves for increased risk of inherited breast cancer or colorectal cancer via one of the following websites. This screen provides an immediate initial risk estimation, but cannot determine if hereditary cancer exists. Those who are found to have increased risk are advised to visit their family physician to discuss a referral to a clinical geneticist for further testing. Which website is it?
- www.erfelijkheid.nl
 - www.oncoline.nl
 - www.kankerrichtlijn.nl
 - www.erfelijkekanker.nl
-
- 19) In a certain family a genetic disorder is passed by women to their sons and daughters, and by men to their sons and daughters, and the chance of repetition is 25-50%. Which inheritance pattern is most likely?
- autosomal dominant with increased penetration
 - autosomal recessive with increased penetration
 - autosomal recessive with decreased penetration
 - autosomal dominant with decreased penetration
-
- 20) In a certain family with an inherited form of cancer, many family members have questions about their chance of getting the cancer. What is the usual form of clinical genetic care in such families?
- The first patient visits the clinical geneticist, the family members are informed via telephone by the clinical geneticist.
 - The first patient visits the genetic counselor, the family members are informed via telephone by the clinical geneticist.
 - The first patient visits the clinical geneticist, the family members are seen by the genetic counselor.
 - The first patient visits the genetic counselor, the family members are seen by the clinical geneticist.
-

eTable 2. Satisfaction questionnaire

1. I would recommend this internet PIN to my colleagues. (On a scale of 1=Totally Agree to 5=Totally Disagree, 6= Not applicable/ No opinion.)

2. In general, I judge the topics presented in the internet PIN as relevant for family practice. (On a scale of 1=Totally Agree to 5=Totally Disagree, 6= Not applicable/ No opinion.)

3. In general, I judge the questions presented in the knowledge test as relevant for family practice. (On a scale of 1=Totally Agree to 5=Totally Disagree, 6= Not applicable/ No opinion.)

4. Which grade would you give this internet PIN, on a scale of 1= Bad to 10= Perfect?

5. How much time did you spend on this internet PIN?

- Less than an hour
- 1 - 1.5 hours
- 1.5 - 2 hours
- 2- 2.5 hours
- 2.5 - 3 hours
- More than 3 hours

6a. Which topic(s) appealed to you the most? (Tick one or more boxes please.)

- Genetic tests
- Hereditary breast- and colorectal cancer
- Referral and cooperation with specialists
- No preference

b. Why?

7a. Which topic(s) appealed to you the least? (Tick one or more boxes please.)

- Genetic tests
- Hereditary breast- and colorectal cancer
- Referral and cooperation with specialists
- No preference

b. Why?

8. Space for optional extra comments:

eTable 3. Applicability questionnaire

1. I apply the knowledge gained from this internet PIN:

- Daily
- Weekly
- Monthly
- I don't come across any genetic issues in my practice and therefore do not apply the knowledge from the PIN.

2. I recognize patients with a genetic condition much earlier than I did before I completed the internet PIN. (On a scale of 1= Totally Agree to 5= Totally Disagree, 6= Not applicable/ No opinion.)

3. I refer to or consult with a clinical geneticist much earlier than I did before I completed the internet PIN. (On a scale of 1= Totally Agree to 5= Totally Disagree, 6= Not applicable/ No opinion.)

4. I have more knowledge about the possibilities and limits of genetic testing than I had before I completed the internet PIN. (On a scale of 1= Totally Agree to 5= Totally Disagree, 6= Not applicable/ No opinion.)

5. I have more knowledge about the most common genetics conditions in the Netherlands than I had before I completed the internet PIN. (On a scale of 1= Totally Agree to 5= Totally Disagree, 6= Not applicable/ No opinion.)

6. I have more knowledge about fundamental concepts of genetics than I had before I completed the internet PIN. (On a scale of 1= Totally Agree to 5= Totally Disagree, 6= Not applicable/ No opinion.)

7. I have more knowledge about important sources of information about genetics than I had before I completed the internet PIN. (On a scale of 1= Totally Agree to 5= Totally Disagree, 6= Not applicable/ No opinion.)

8. Space for optional extra comments:

eTable 4. Demographics and practice characteristics questionnaire

1) You are:

- Male
- Female

2) Age: ... years old

3) Number of years experience as General practitioner: ...years

4) In which type of practice do you work?

- Solo practice
- Duo practice
- Group practice
- Community Health Center
- Other

5) Degree of Urbanization of Practice Area:

- Metropolitan area (>100.000 residents)
 - City (30.000 - 100.000 residents)
 - Small Town (5.000 - 30.000 residents)
 - Rural area (<5.000 residents in largest village)
-

eTable 5. Demographical and practice characteristics of participants

	Control Group (n=24)	Treatment Group (n=20)	Chi-square/ Mann-Whitney Test (P values)
<i>Sex</i>			
Female	20	19	.23
Male	4	1	
<i>Age in years</i>			
30-39	13	10	.70
40-49	7	5	
50-59	3	5	
60-69	1	0	
<i>Professional experience in years</i>			
less than 10	13	12	.92
between 10 and 19	7	4	
between 20 and 29	3	1	
between 30 and 39	1	3	
<i>Practice Type</i>			
Solo practice	4	0	.29
Duo practice	3	5	
Group practice	4	5	
Community Health Center	4	2	
Other	9	8	
<i>Practice Setting</i>			
Metropolitan area (>100,000 residents)	7	4	.82
City (between 30,000 and 100,000 residents)	7	5	
Small Town (between 5,000 and 30,000 residents)	7	7	
Rural area (<5,000 residents in largest village)	3	4	

eTable 6. Evaluation of G-eCPD¹ according to two levels of Kirkpatrick

Instrument	Item description	Kirkpatrick level of evaluation
Knowledge test	20 Multiple Choice items; on knowledge of basic genetic principles, genetic disorders, possibilities and limitations of genetic tests, referral possibilities concerning genetics and most important sources of genetic information	KP ² 1 and 2; evaluation of participation and learners behavior (effects on knowledge and skills)
Satisfaction questionnaire	3 items on user relevance with a 5-point Likert scale. 1 item global evaluation rating score (1-10). 1 item on amount of time spent on the online module 2 items on user opinion on personal preference of the topics with multiple choice options and explanation	KP 1; evaluation of satisfaction and opinions of participants
Applicability questionnaire	1 item on frequency of application of knowledge acquired through the online G-eCPD with a 4-point ordinal scale. 6 items on applied knowledge in daily practice (recognizing patients with a common genetic form of cancer, referral to secondary care, knowledge of genetic tests and common genetic diseases, basic genetic terms, usage of important resources of genetic information) with a 5-point Likert scale.	KP 2; effects on attitudes and perceptions, knowledge and skills, effects in change of behavior and possibly evaluation on the effects on patient genetic Health
Demographics and Practice Characteristics Questionnaire	5 items on age, gender, years of experience as a FP ³ , practice situation (urban/rural practice) and practice type (group/single practice).	Not applicable

¹ Genetic online Continuing Professional Development; ² Kirkpatrick levels of effect of Continuing Professional Development (CPD) programs (Kirkpatrick 2006); ³ Family Physician

Chapter 5

Effectiveness of oncogenetics training on general practitioners' consultation skills: a randomized controlled trial

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Abstract

Purpose

General practitioners (GPs) are increasingly called upon to deliver genetic services and could play a key role in translating potentially life-saving advancements in oncogenetic technologies to patient care. If GPs are to make an effective contribution in this area, their genetics competencies need to be upgraded.

The aim of the study was to investigate whether an oncogenetics training for GPs improves their genetic consultation skills.

Methods

In this pragmatic, blinded randomized controlled trial, the intervention consisted of a four-hour training (December 2011 and April 2012), covering oncogenetic consultation skills (family history, familial risk assessment, and efficient referral), attitude (medical ethical issues), and clinical knowledge required in primary care consultations. Outcomes were measured using observation checklists by unannounced standardized patients and self-reported questionnaires.

Results

Of 88 randomized GPs who initially agreed to participate, 56 completed all measurements. Key consultation skills significantly and substantially improved; regression coefficient post-intervention equal to .34 and .28 at 3-month-follow-up indicating moderate effectsize. Satisfaction and perceived applicability of newly learned skills were highly scored.

Conclusion

The GP-specific training proved to be a feasible, satisfactory and clinically applicable method to improve oncogenetics consultation skills and could be used as educational framework to inform future training activities with the ultimate aim of improving medical care.

Trial Registration: trialregister.nl Identifier: NTR3323

Introduction

Genomics holds great promise to improve human health. Genetics of common disorders (diabetes, cancer, cardiovascular diseases), and monogenic subtypes (Maturity Onset Diabetes of the Young (MODY), *BRCA 1/2*, familial hypercholesterolemia (FH) and long QT syndrome) in particular, are expected to come increasingly to the forefront in primary care. Consequently, general practitioners (GPs) are facing a daunting informational challenge to keep abreast of the expanding body of genomics knowledge and attain competencies for informed use of its potential for personalized patient care.¹ In view of increasing requests for DNA-based predictive testing arising from a positive family history and GPs' increasing involvement in preventive check-ups, it is important for GPs to be competent to take and interpret a family history and deal appropriately with patients' questions and concerns.² Each family practice has a substantial number of unidentified asymptomatic patients with relatively young first-degree relatives with familial or hereditary forms of cancer (breast, ovarian, uterine, and colorectal cancer), and such patients should be referred to a clinical geneticist for counseling and/or screening according to guidelines.^{3,4} Women carrying a *BRCA1/2* mutation, for example, have a lifetime-risk of 60-80% of developing breast cancer (accounting for 5-10% of all breast cancer cases), and timely identification enables them to benefit from otherwise unexploited life-saving "risk-management options", such as salpingo-oophorectomy and/or mastectomy, annual screening, and pharmaceutical chemo preventive options.⁵ Assessing familial risk by taking a family history can be a reliable method to improve outcomes of hereditary forms of cancer with targeted cancer prevention strategies.⁶⁻⁹ Taking an adequate family history however is difficult and takes time. Insufficient genetics knowledge and consultation skills to actually conduct an initial oncogenetics risk assessment and its interpretation pose a barrier to appropriately recognize and elicit details to assess the features of potential oncogenetics risk.⁶ This could warrant timely referral to oncogenetics services for further assessment and genetic testing (referral-level competences). Moreover, lack of computerized decision support implies that GPs themselves need to learn adequately interpreting family history and act on it.¹⁰ Educational innovation therefore seems imperative, including genetic risk ascertainment and prevention.¹¹ Unless GPs receive proper education and training, individual genetic care by GPs will likely be unhelpful and possibly even harmful.^{3,12,13} Considering the urgent need for and the potentially huge benefits to be gained from genetics education for GPs, we embarked on an educational project aimed at strengthening the role of genetics in family medicine.

Well-defined core genetics competencies for non-genetic health care workers are considered a precondition for the development of effective genetics educa-

tion.^{14,15} Educational activities should be responsive to GPs' assessed needs in respect to cognitive (*knowledge*), psychomotor (*consultation skills*), and affective (*attitude*) aspects of genetics competence. Previous studies have shown that as far as genetics is concerned non-genetic healthcare workers require not only education but also clear guidelines and definitions of their responsibilities.^{16,17}

This article reports a study in which GPs attended a needs-based, interactive oncogenetics training aimed at enhancing insight, consultation skills, and attitudes relevant to the identification of oncogenetic disease in family practice consultations. We evaluated the effects of the training in two ways: 1. Office visits by standardized patients (SPs) to determine the extent to which GPs synthesized and applied the newly learned behaviors; and 2. Questionnaires to determine GPs' satisfaction with the training and perceived applicability of the new genetics consultation skills in their practice.

Materials and methods

Experimental design

We conducted a pragmatic, blinded randomized controlled trial (RCT) with parallel repeated measurements using a performance checklist and questionnaires. Kirkpatrick's four level framework for evaluating educational outcomes entails: 1. valuation (satisfaction), 2. learning (knowledge and knowledge retention), 3. behavior (applying knowledge about timely identification of patients at risk and referral), and 4. effects on patient health and organization (change in practice and results).^{18,19} The design included an innovative measurement method with office visits by SPs aimed at the third level. Unannounced SPs are a proven method to collect data about real practice in a direct and reliable way.²⁰

Participating GPs were randomly assigned to a training date: December/January for the intervention group (four sessions) or March/April for the control group (three sessions). The trial ran from December 2011 to April 2012.

The RCT involved an intervention (oncogenetics training) and repeated measurements before (T0), one month (T1) and three months (T2) after the intervention (table 1). All participants completed a demographics survey at T0. Between T0 and T1 the intervention group attended the training, while the control group received no intervention. For the evaluation of genetic consultation consultation skills at T0 (pre-test), at T1 (post-test) and at T2 (retention test), SPs completed checklists after consultations with both GP groups (level 4)²¹. The SPs were blinded to GPs' group assignment. To measure satisfaction with the training (level 1), the intervention group completed a questionnaire at T1. To measure participants' perceived applicability of the training content (level x), the intervention

group answered a questionnaire at T2. To stimulate compliance of control group participants, they were invited to attend the training after T2.

The ethical review boards of the Netherlands Association for Medical Education, Maastricht University Medical Center, and VU University Medical Center Amsterdam, the Netherlands approved the study protocol. All participants gave written informed consent before the trial.

Table 1. Time table of the randomized controlled trial showing scheduled measurement times, instruments, and measurements made (indicated with X) in the Intervention and Control groups.

Instrument	Group	Time			
		0 Pretest (T0)	2 weeks Oncogenetics training	1 month Posttest (T1)	3 months Retention test (T2)
Standardized patients' checklist	Intervention	X ¹		X	X
	Control	X		X	X
Satisfaction questionnaire	Intervention			X	
Applicability questionnaire	Intervention				X
Demographics questionnaire	Intervention & Control	X			

¹ measurement made with the instrument indicated in column 1, in the group indicated in column 2.

Participants

The project team collaborated with the Dutch College of General practitioners (NHG) and local training providers to recruit GPs working full - or part time in general practice.

For logistic reasons, recruitment was limited to all GPs practicing in two Dutch provinces, who received an invitational online mailing with information about the aim of the study, the contents of the face-to-face training, and the evaluation procedure. Accreditation points were offered to GPs completing the study. A book on genetics in general practice or a book voucher of equal value was offered as an extra incentive. Four email or telephone reminders were sent to non-responders.

Eighty participants were needed to detect a medium- to large-sized effect with a power of 90% and significance level of 5%.²² Figure 1 shows the randomization scheme and participation.

Intervention

The intervention group attended a four-hour face-to-face evening training covering oncogenetic consultation skills (family history, familial risk assessment, and efficient referral), attitude (medical ethical issues), and clinical knowledge required in primary care genetic consultations. More specifically, the training comprised the following educational content aimed at equipping GPs to:

- recall clinically relevant information about types of hereditary cancer (breast, ovarian, colon, skin) including genes associated with oncogenetics syndromes most commonly tested for;
- recognize patients with features suggesting inherited predisposition to cancer;
- draw a family tree as a tool to identify patients at risk;
- discuss (possible) familial and hereditary cancer risks, management of potentially developing hereditary cancer (i.e. surveillance and risk-reducing surgical options) and related ethical issues;
- identifying patients for referral for risk assessment and find relevant information online using oncogenetics guidelines;
- explain the possibilities and limitations of oncogenetic testing;
- know when to consult and/or refer to a genetics specialist.

The training was developed by a multidisciplinary team consisting of an NHG educational expert (EJvdJ), GP researcher (EJFH), two clinical geneticists, and two educationalists (SvL, CvdV). The focus was on oncogenetic diseases with relatively high prevalence in family practice (breast cancer due to *BRCA* mutations, colon cancer (e.g. FAP, Lynch syndrome) due to *APC*/mismatch-repair gene mutations, and skin cancer (e.g. Familial Atypical Multiple Mole Melanoma (FAMMM) Syndrome due to *CDKN2A* (p16) gene mutations)). The training started with a one-hour interactive theoretical session on hereditary forms of cancer lead by a clinical geneticist from a local academic hospital who was familiar with the aims of the program, followed by a one-hour session with two patients of the Dutch BRCA patient organization, who talked about their experiences, discussed ethical issues and answered questions. A short break was followed by a two-hour workshop in which participants in small groups engaged in three role-played consultations for three oncogenetic problems in the presence of experts (clinical geneticist, patient representatives, and two trainers). Patients and GPs were role-played by participants and the others gave feedback.

Measurements

Standardized Patients

For a detailed description of the training sessions with SPs preceding the practice visits, clinical case scenarios (eTable 1) and development and finalization of checklist (eTable 2), see Supplementary Materials and Methods information.

Questionnaires

Three online self-reported questionnaires were used to collect data on satisfaction, applicability of new consultation skills, and demographics and practice characteristics, (see eTables 3, 4 and 5). The instruments were developed and validated in collaboration with the research team. The questionnaires were developed and validated in collaboration with content experts (experts in daily clinical genetics, a GP, and an expert in education and questionnaire development) and pilot tested.”

The satisfaction questionnaire contained two items with five-point Likert scales (1: completely disagree; 5: completely agree) and an item with a global rating on a ten-point scale.⁵ The applicability questionnaire contained six items with five-point Likert scales and one item with a four-point ordinal scale.

Regression analysis

For detailed description of Regression analysis to investigate improvement of genetic consultation behavioral skills, see online-only material contents (*Details of background of Methods –Analysis of Regression*).

Satisfaction with the intervention and applicability scores were analyzed by calculating mean scores, 95% confidence intervals, and standard deviations for the pooled data from the satisfaction questionnaire.

All analyses were performed using SPSS version 19 (SPSS, Chicago, IL).

Results

Randomization and dropout comparisons

Of 88 randomized GPs who agreed to participate in the training in December/January 2011/2012 (intervention group) or March/April 2012 (control group), 56 (38 intervention, 18 control group) completed the entire procedure, and 32 were lost to follow-up due to lack of time or sickness (Figure 1).

Participant characteristics

Participants in the intervention and control groups did not differ significantly in age, gender, years of experience, type of office, and office situation (See table 2).

Table 2. Demographics and Practice Characteristics of Participants (n)

	Control Group (n=18)	Intervention Group (n=38)	Chi-square/ Mann-Whitney Test (P values)
<i>Sex</i>			
Female	12	29	.446
Male	6	9	
<i>Age in years</i>			
30-39	3	14	
40-49	5	6	
50-59	7	12	.318
60-69	2	4	
Unknown	1	2	
<i>Professional experience in years</i>			
Less than 10	4	9	
Between 10 and 19	3	7	
Between 20 and 29	7	6	.679
Between 30 and 39	0	2	
Between 40 and 49	0	1	
Unknown	4	13	
<i>Practice Type</i>			
Solo practice	1	5	
Duo practice	4	7	
Group practice	2	3	.364
Community Health Center	5	3	
Other	4	12	
Unknown	2	8	
<i>Practice Setting</i>			
Metropolitan area (>100,000 residents)	10	11	
City (between 30,000 and 100,000 residents)	1	6	
Small Town (between 5,000 and 30,000 residents)	3	10	.145
Rural area (<5,000 residents in largest village)	1	0	
Unknown	3	11	

Effects of the intervention on oncogenetics-related learned consultation skills

Each of the 56 GPs was visited by three SPs, portraying different cases, resulting in 168 first visits (Figure 1).

EFFECTIVENESS OF ONCOGENETICS TRAINING ON CONSULTATION SKILLS

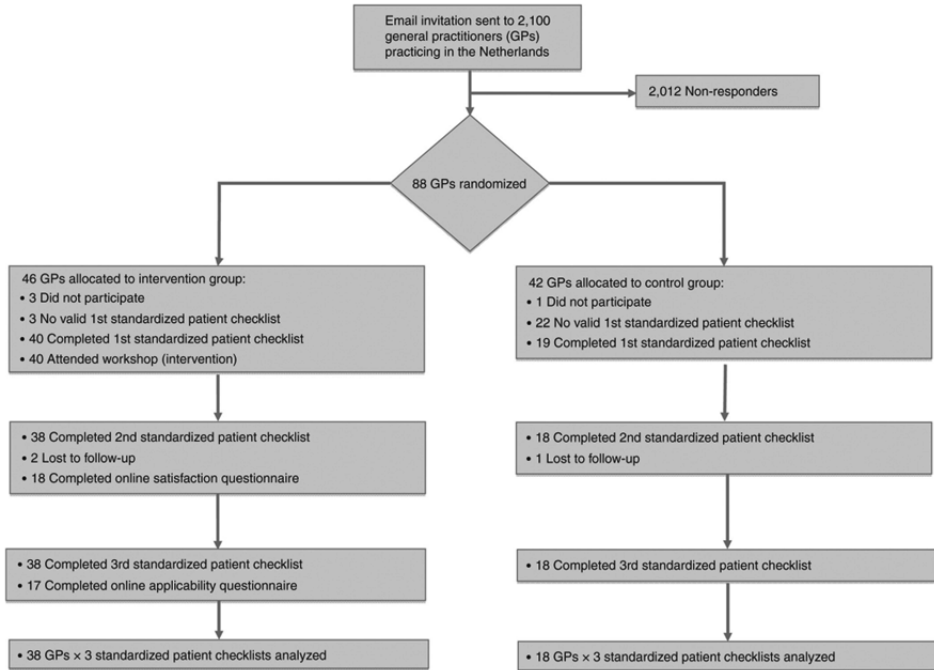


Figure 1. Consort Diagram of Randomization and Participation

Figure 2 shows the raw mean performance scores (proportion correct) for the control and intervention groups at times T0 (pre-test), T1 (post-test), and T2 (retention-test). Between-group differences were found to be non-significant for pre-test and retention-test, but the post-test difference of 0.19 in favor of the intervention group was found to be significant (t-test, $p < 0.0005$). These estimations, however, are based on raw means and may be biased due to differences in difficulty between the three SP cases. More precise and unbiased estimations were obtained by the regression analysis (table 3). The regression results for ScoreT1 showed that the effect of the intervention (the coefficient for Train) was statistically significant and amounted to .14 on the proportion correct scale; the corresponding value for the standardized regression coefficient was equal to .34 indicating a moderate effect size. The analysis for ScoreT2 showed that the significant intervention effect persisted until the retention measurement at T2 (two months later) and amounted to .11 (standardized regression coefficient=.28, moderate effect size). Hence, the performance improvement due to the intervention was still substantial at T2, being equal to 80% of the immediate effect at T1.

Effect modification of the treatment effect by baseline value was tested for the T1 and T2 scores; for both variables, the effect modification was found to be non-significant.

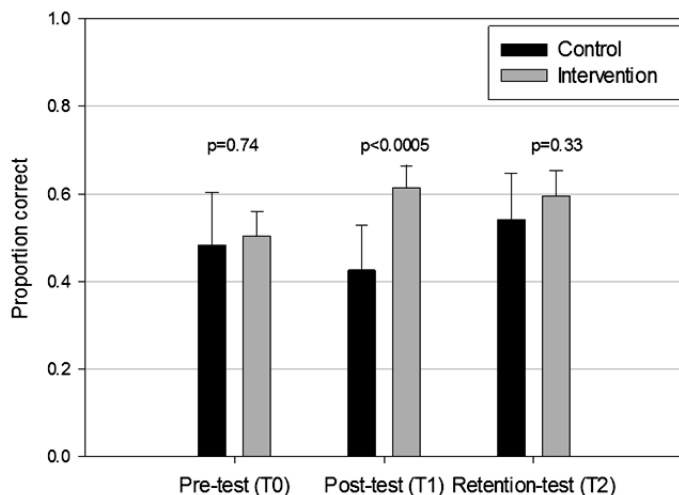


Figure 2. Performance scores for oncogenetics consultation skills as measured by proportion correct on SP checklists (mean and 95% confidence interval) for control group (black) and intervention group (gray) at T0, T1, and T2, corresponding to pre-, post-, and retention-measurement, respectively.

Table 3. Effect of the oncogenetics training on the performance of GPs. Regression results are shown for immediate gain of performance (ScoreT1) and retention of performance (ScoreT2), using the pretest score (Score T0) as a covariate and the control group score as a reference.

Dependent Variable	Independent Variables								
	Constant	Score T0				Train			
		regression coefficient	regression coefficient		standardized regression coefficient	regression coefficient		standardized regression coefficient	
			value	95% CI		value	95% CI	value	
		low	high		low	high			
ScoreT1	.58 ***	.23	-.02	.47	.23	.14 **	.05	.23	.34
ScoreT2	.52 ***	.15	-.07	.38	.16	.11 *	.03	.20	.28

*** p<0.001; ** p<0.01; * p<0.05; CI: confidence interval

Satisfaction and applicability

The satisfaction questionnaire resulted in high scores for the two items (both 4.4) and a global score of 7.7; when applicability is also considered, favorable scores were found for all six items (3.5-4.5). Overall, 65% of the trainees reported applying the newly learned skills monthly, and 35% weekly (table 4).

Table 4. Satisfaction (intervention only; N=18) and self-reported applicability (intervention only; N=17) as a result of oncogenetics training.

Variable	Mean (min - max)	95% CI		SD
		low	high	
<i>Satisfaction</i>				
Would recommend the module to a colleague ¹	4.4 (2 - 5)	3.9	4.9	0.98
Content of the module is relevant for a GP	4.4 (1 - 5)	3.8	5.0	1.10
Content of the knowledge test is relevant for a GP	4.1 (2 - 5)	3.7	4.5	0.83
Global score (1-10)	7.7 (1 - 10)	6.7	8.6	1.90
<i>Applicability</i>				
Recognize patient with genetic disease sooner	4.1 (2 - 5)	3.6	4.5	0.90
Sooner refer to or discuss with a genetic specialist	3.9 (3 - 5)	3.5	4.4	0.77
More knowledge of possibilities/limitations of genetic tests	4.0 (3 - 5)	3.7	4.3	0.61
More knowledge of genetic diseases	3.7 (1 - 5)	3.2	4.2	1.00
More knowledge of basic genetic concepts	3.5 (2 - 5)	3.2	3.9	0.72
More knowledge of genetic information sources	4.5 (2 - 5)	4.0	4.9	0.87
<i>Proportion of trainees applying the learned knowledge (%)</i>				
daily	0			
weekly	35			
monthly	65			
not (do not meet any genetic problems in our practice)	0			

CI, confidence interval; GP, general practitioner; max, maximum; min, minimum.

¹ if not indicated otherwise results refer to scores of 5-point Likert scale items (1: completely disagree, 5: completely agree); CI: confidence interval

Discussion

Summary of main findings

To our knowledge this is the first RCT to use SPs to investigate improvement of GPs' oncogenetic professional behavior after attendance of an oncogenetics training. The results show sustained improvement 3 months after the training, as well as high satisfaction with the training and positive perceptions of the practical applicability of training topics.

Immediate and long-term training effects were evaluated at Kirkpatrick's level 3 (behavior showing evidence of learning), which enhances the value of the findings.^{22,23} The results indicate that case-based oncogenetics education can achieve sustained improvement with a moderate effect size in urgently desired genetics competencies for GPs, while the positive results for satisfaction and applicability may reflect a move toward a culture of genetic medical practice improvement. Educational interventions likely have a small to moderate effect on physician knowledge and performance, and patient outcomes.²⁵ A few factors that were applied could have supported this result, such as active and interactive sessions, single group and smaller group sessions. Whether there is a sustainable impact on applicability of the training in practice, including timely identification of patients with a possible cancer predisposition syndrome and appropriate referral, will need further longer-term studies. Designed to fill gaps in physicians' competencies and boost their confidence in using basic clinical genetic principles and activities, the oncogenetics training addressed previously prioritized key features of genetic consultation skills and attitude, but not basic science knowledge.³¹⁻³⁵

SPs have been used successfully before to assess changes in clinical competence and performance and sustained effectiveness of behavioral training, but not to evaluate attitudinal factors cited as directing practice performance, such as patient satisfaction.^{21,26,27}

After the training the participating GPs seemed to be more comfortable incorporating oncogenetics aspects in patient consultation skills as reflected in their high perceived applicability. It seems plausible that this, in turn, will enhance efficient and effective referral for genetic counseling. Whether the latter effect will materialize, however, remains to be examined in future studies. Taylor et al. discussed barriers to effective primary care involvement in the expanding field of adult genetics, arguing that genetic medicine should be part of integrated medical care and therefore of primary care medicine.²⁸ We agree with this viewpoint and feel that the training we designed shows promise to enhance communication between GPs and the genetics community, identification of high-risk patients, and timely referral to genetics services.

Methodological considerations

One of the aims of including real patients and simulated consultations in our training was to promote a favorable attitude among GPs to the application of genetic competencies. A study by Carroll et al. measured intent to use clinical genetics scenarios and increase competence due to a multifaceted knowledge translation intervention but used questionnaires and not ratings of observed practice behaviors.²⁹ Patient and societal perspectives on legal consequences of DNA-based testing results (for example being able to find a genetics information source or ability to obtain a mortgage or life insurance) however, demand that physicians' effective use of genetics be demonstrated by actual performance in health care practice.^{7,21} We therefore deliberately deployed trained and blinded SPs to optimize the value of the measurement. Repeated SP visits may have impacted the outcome of learning effects in both study groups, as the GPs would have had a higher level of awareness of being critiqued and could have felt a certain pressure to perform appropriately, but this is controlled for in the current study thanks to its RCT design.³⁰

Rollnick et al. suggested that learner-directed and context-bound consultation skills training should be integrated in everyday practice in a way that is acceptable to clinicians.³¹ Based on this principle we had physicians identify their training needs and tailored our training to the practice context by patient centered consultation skills training. Based on the results of our earlier studies we emphasized everyday genetic clinical experiences more than consultation skills and attitude alone.^{16,17} Our strategy could therefore be described as an “*enriched* context-bound consultation skills training”. Informal comments after the training by participants made clear that this format had a positive effect on their learning.

Potential oncogenetic problems are considered very personal topics to discuss between a patient and their own general practitioner (GP).¹⁶ This determined why it was not discussed in an incognito setting with a so-called “new” patient or unannounced, concealed simulated patient.³²

Strengths and limitations

Strength of the study was measuring change in consultation skills after the training by using SPs in particular, as opposed to using computerized case-based testing for example. A variation of measurement instruments was proposed to predict practice performance.³³ SP-based measurement is relatively unobtrusive, highly authentic and based on patient perception. Another strength of the study is the fact the educational intervention was tailored to the learners' needs.^{6,16} Because the current study is confined to one health-care setting within one country, the generalizability of the results may be limited. The training's demands on resources, facil-

ities, and logistics may limit the feasibility of training delivery in many different settings. Nevertheless, SP-based assessment is a valid instrument to describe what happens in real practice and can therefore provide valuable information for advanced development of genetic trainings. The study design introduces the possibility of bias by virtue of each GP seeing three different case presentations in different orders. This potential limitation was acknowledged and statistical accommodations were made.

Using comparable case scenarios in this study, it was possible to measure change in checklist scores over time. However, it remains to be investigated whether it is possible to use different scenarios, for example based upon a family history alone. This would be a scenario seen in daily GP practice and the time when timely referral could be of benefit to the pre-symptomatic patient in regards to preventing or reducing familial cancer risk. Future studies could include assessing the issues addressed in the study by Culver et al, namely satisfaction with the time to address concerns, acknowledgements of patient concerns about cancer risk by physician and offering reassurance.³⁴ Using the validated checklist, the current study measured GPs' genetics consultation skills, thereby reflecting that training outcomes' covering the full scope of good practice consultations: key ingredients related to family history taking, genetic risk assessment, and referral to genetics specialists. The SP requested standard 10-minute appointments. This may seem short for a first consultation, however this is standard duration the Netherlands. If requested, in "real world" clinical practice, a follow up appointment could be made to adequately address all of the issues concerned further. However, in the study design, the SPs came in with a concern possibly related to an inherited form of cancer. The extent to which GPs synthesized and applied the newly learned behaviors was assessed by long-term changes in a 28-item checklist score, not by whether all issues would be discussed. Performance assessment is considered representative of a product of competence, influences of individual (e.g. health, relationships) and organization (e.g. facilities, practice time). GPs were therefore similarly assessed for performance under equivalent conditions (e.g. appointment time limitation focused entirely on the sole reason for visit, without distraction or delay).^{21,27,35}

Although the three-month study period may have been too short to detect sustainable practice improvement long term, repeated measurement of consultation skills predicts practice performance long term.^{21,26}

Voluntary participation by interested GPs could have caused selection bias. However, similarity of the baseline characteristics of the two groups and comparability of the 60% participation rate with that of other studies among GPs indicate that the participants were representative of GPs likely to attend oncogenetic training in the future.^{36,37} Furthermore, it is possible that participating in the oncogenetics training might become part of standard training for all GPs. There was an

imbalance however between the drop out rate in the intervention and control group and the reason for this is not clear. Attending the training in the beginning of the trial period could have provided the urgent information to be able to satisfactorily finish all measurements long-term. Participants in the control group on the other hand had to wait for training content, possibly causing resistance to finish all measurements resulting in drop out. General reasons for drop out were reported (no time and sickness), not specific attributes. It is therefore unlikely that self-selection in dropout negatively impacted the validity of the results.

A pragmatic and blinded study design has known limitations.³⁸ Obviously, it is preferable for an RCT where participants are blinded to inclusion in the intervention or control group, that those conducting the measurements are blinded as well.³⁹ We achieved this by blinding the SPs to the GPs' group allocation and by having two independent researchers (AMMM and SRvT) analyze the checklist scores in a blinded manner.

The results indicate that an oncogenetics training designed to meet GPs' educational needs can be a satisfactory and feasible method for sustained improvement of competencies to ensure appropriate application in family medicine of developments from the rapidly evolving field of genetics. Learner-directed and context bound genetics education appears to be a valuable tool to stimulate GPs to deliver genetic services.⁴⁰ We plan to use the results to inform the design of new trainings on complex genetic diseases, including hereditary forms of cancer, cardiovascular disease, and diabetes, in our continuing efforts to improve referral strategies and timely recognition of high-risk genetic patients. Large-scale international randomized controlled trials with adequate power are warranted to further assess how genetics education can improve health care.

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Online-only material contents

eTable 1. Description of cases presented by standardized patients

eTable 2. Standardized patient checklist items assessing genetic consultation skills

eTable 3. Satisfaction questionnaire

eTable 4. Perceived applicability questionnaire

eTable 5. Demographics and practice characteristics questionnaire

Supplementary Materials and Methods: Details of background of Methods –
Measurements of genetic consultation skills through standardized patients and
Analysis of Regression

EFFECTIVENESS OF ONCOGENETICS TRAINING ON CONSULTATION SKILLS

eTable 1. Description of Cases presented by Standardized Patients

	Skin cancer case	Colon cancer case	Breast cancer case
Biography	Male or female, age 31. Born and raised in suburb of major city. Went to culinary school, works at restaurant. Lives in apartment downtown, romantic relationship ended two years ago.	Male or female, age 41. Born in small city, moved to outskirts 10 years ago. After high school, first worked in do-it-yourself store, past 15 years as taxi driver. Relationship with spouse is stable.	Female, age 41. Lives in quiet suburb with husband, daughter age 13 and son age 10. After college began working at bank. Family situation is stable, spends a lot of time with children in the weekend.
Medical complaint	Spots on back which bleed, itch and are getting bigger. Last month doctor called it a regular birthmark. Spots noticeable for at least 3 months.	Pain in lower left abdomen and watery brown bowel movements. Symptoms began 3 weeks ago after trip to Thailand. Obesity, but recently lost 1 kilo.	Lump in left breast, noticed last week. The 4 cm irregular swelling not painful but sensitive. The skin on the swelling is a little red and dimpled.
Lifestyle	Smokes half a pack of shag a day since age 17. Drinks heavily twice a week after work. No drugs, no coffee, tea once a day. Sunbathing vacation in southern France every year.	No history of smoking. Drinks wine occasionally. On Mirtazipine (anti-depressant) for a year. Drinks 4 cups of coffee and 2 cans energy drink per day. Fresh vegetable is absent from diet, does eat 2 fruit servings.	Heavy smoker from age 16 up to 2 years ago. Drinks wine with dinner and social drinker in the weekend. Drink 2 cups of coffee a day.
Medical history	No medical history. Family history: Sister: skin cancer at age 23, died two years later of brain tumor. Father: skin cancer at age 40, treated and cured. Grandfather, father's father: skin cancer at age 43, died 5 years later of brain tumor. On mother's side no one has cancer.	Mild depression Obesity;BMI 35 Family history: Father diagnosed with colon cancer at age 40, died of it a year later. No contact with rest of family.	No medical history. Family history: Grandfather, mother's father: unknown cancer and died, age 55. Mother: breast cancer at age 35 and is now 75. Sister: ovarian

	Skin cancer case	Colon cancer case	Breast cancer case
Motivation for appointment	You want a thorough examination, because you know how quickly cancer can develop. If the doctor's diagnosis isn't cancer, you will need convincing. You know that your sister had the same symptoms back then and it took a lot of persistence to get a referral and diagnosis.	You want a solution to your symptoms, and clarity on whether it's cancer or not. You know your father got cancer at your age and it killed him. You don't mention this until later; you don't like to talk about family. You don't have much contact with relatives due to conflicts.	cancer at age 30 and died at age 33; other 2 sisters seem healthy. On father's side no one has cancer. You're scared of cancer because there's a lot of it in the family. You want to know whether the lump will get larger and if it's malignant. You really want to be referred to the specialist. If the doctor doesn't do this, let him or her know you're really scared and be disappointed.
Physical examination results (given in paper format by the SP to the GP during the visit if requested)	<i>Inspection:</i> A lesion is visible on the right shoulder blade. The lesion has an erratic surface, irregular border and color, and is 2 cm in diameter.	<i>Inspection:</i> no irregularities. <i>Auscultation:</i> plumbing sounds. <i>Percussion:</i> normal sounds in all four quadrants of abdomen. <i>Palpation:</i> no palpable irregularities. Rectal exam: normal.	<i>Inspection:</i> no irregularities. <i>Palpation:</i> a palpable mass in the left breast. Size: 2cm. Consistency: solid-elastic. Irregular shape. Not painful, no inflammation. No discharge from nipple.

EFFECTIVENESS OF ONCOGENETICS TRAINING ON CONSULTATION SKILLS

eTable 2. Standardized patient checklist items assessing genetic consultation skills

	Score as yes:	YES	NO	?
1. Responds to my concern about possible genetic cancer.	"I can imagine you are worried about that", "what are you concerned about?", etc.			
2. After expressing my concern, the GP confirms that this form of cancer can be hereditary.	"This form of cancer can be hereditary", "this form of cancer is often passed in families", etc.			
3. Asks about cancer in my family.	"Do any of your relatives have cancer?", etc.			
4. Further asks which kinds of cancer occur in my family.	"Which kind of cancer did they have?", "Can you remember what kinds of cancer your relatives have?", etc.			
5. And asks at which age the cancer was diagnosed.	"At which age did the doctors discover that your relative had cancer?", "How old were they when they got the diagnosis?", etc.			
6. Asks who was diagnosed.	"Can you remember who had this kind of cancer?", "Which relatives have this kind of cancer?"			
7. Asks specifically if this kind of cancer is present in my children, siblings or parents (first -degree relatives).	"Have your children, parents or siblings had this kind of cancer?"			
8. Draws a family tree to clarify possible inheritance of cancer.	A family tree is drawn.			
9. Explains the possibilities and limits of genetic testing.	"What we can/can't determine by genetic testing is...", "With genetic testing we can diagnose X but not Y."			
10. Explains that a genetic test for this kind of cancer does not prevent that I may or may not get that cancer in the future.	Makes it clear that interpretation of tests is still limited. "This test doesn't offer certainty that you will or won't get this form of cancer at some point in your life."			
11. Explains what the consequences could be for me if a certain genetic test for hereditary cancer comes back positive.	"If the test indicates that you have this inherited defect, then...", "A positive test result would mean for you..."			
12. Asks about my expectations regarding the possibilities and limits of genetic testing.	"what do you expect from the genetic test?", "Do you expect to have certainty after having the test?"			
13. Explains why referral to the clinical geneticist is or is not useful.	"Referral to a clinincal geneticist can be useful in your case", "a referral in your case isn't useful because..."			

CHAPTER 5

	Score as yes:	YES	NO	?
14. Considers looking up referral criteria on the internet (possibly with me)	"I'll look online if there is an indication to refer you to the clinical geneticist", "to be sure that it's useful to refer you to the clinical geneticist, I'll look up their website"			
15. Knows about relevant clinical geneticists in the region (possibly by looking with me online)	"By this department of clinical genetics are the following possibilities...", "Let's together look at what the clinical geneticists can do for you."			
16. Considers contacting (by phone) the clinical geneticist to discuss possible referral (possibly with me)	"I will contact the specialist in genetics to discuss your situation", "I'm calling the clinical geneticist to be sure we make the right decision"			
17. Explains when I would meet the criteria for referral to clinical genetics	"The following criteria must be met for a referral ...", "If this happens, then I can refer you to clinical geneticist"			
18. Discusses with me whether relatives may be informed that referral is useful for them	"Perhaps it's a good idea to inform your relatives and have them tested as well", "It's important that your family members also get tested for this condition"			
19. Discusses with me whether relatives should make a GP appointment, if it turns out it is hereditary cancer	"Your relatives who also come to this practice, could also have a higher risk and should be tested", "Because your family members could have the same condition, I would like to also ask them to have a genetic test."			
20. If yes, discusses with me whether relatives may receive information about me.	"Do you have a problem with that?", "What do you think about that?"			
21. Indicates that cancer indeed occurs more often in my family than one would expect	"cancer indeed occurs more often in your family than I would expect", "Your family does have a lot of this kind of cancer"			
22. Indicates that this could point to a hereditary form of cancer.	"We do see this kind of cancer in hereditary form", "It's possible that this kind of cancer is inherited"			
23. Registers the genetic risk (family history) of this kind of cancer in the computer system	"I'm registering this genetic risk of this kind of cancer in your record", "I see you typing, what are you entering, is it in my record?"			

EFFECTIVENESS OF ONCOGENETICS TRAINING ON CONSULTATION SKILLS

	Score as yes:	YES	NO	?
24. Mentions possible consequences of the genetic risk for my children, cousins/nieces and nephews	"The chance that your children get this condition is...", "There's a higher chance that your nieces will develop this condition"			
25. Refers me to the closest clinical genetics outpatient clinic	"I would like to send you to the clinical genetics outpatient clinic", "They can help you better at the clinical genetics outpatient clinic, so I'm sending you there."			
26. When I ask for more information to read at home, the doctor mentions www.erfelijkheid.nl (a Dutch website on general genetics topics) or another website	"If you want to read more info on the internet at home, you can look at www.erfelijkheid.nl ", "At (another site) you can find more info"			
27. Gives me a patient information letter	"In this letter you can read more information"			
28. Mentions possible support from the relevant patient organisation when I ask where else I can turn to with my concerns	"You could find support by a patient organisation", "A specific patient group could support you with your condition"			

eTable 3. Satisfaction Questionnaire

1. I would recommend this training to my colleagues.

(On a scale of 1=Totally Agree to 5=Totally Disagree, 6= Not applicable/ No opinion.)

2. In general, I judge the topics presented in the training as relevant for family practice.

(On a scale of 1=Totally Agree to 5=Totally Disagree, 6= Not applicable/ No opinion.)

3. Which grade would you give this training, on a scale of 1= Bad to 10= Perfect?

CHAPTER 5

eTable 4. Perceived applicability questionnaire

-
1. I apply the knowledge gained from this training:
- Daily
 - Weekly
 - Monthly
 - I don't come across any genetic issues in my practice and therefore do not apply the knowledge from the training.

(On a scale of 1=Totally Agree to 5=Totally Disagree, 6= Not applicable/ No opinion.)

2. I recognize patients with a genetic condition much earlier than I did before I completed the training.
3. I refer to or consult with a clinical geneticist much earlier than I did before I completed the training.
4. I have more knowledge about the possibilities and limits of genetic testing than I had before I completed the training.
5. I have more knowledge about the most common genetics conditions in the Netherlands than I had before I completed the training.
6. I have more knowledge about fundamental concepts of genetics than I had before I completed the training.
7. I have more knowledge about important sources of information about genetics than I had before I completed the training.
8. Space for optional extra comments:
-

eTable 5. Demographics and Practice Characteristics Questionnaire

-
- 1) You are:
- Male
 - Female
-
- 2) Age: ...years old
-
- 3) Number of years experience as General practitioner: ...years
-
- 4) In which type of practice do you work?
- Solo practice
 - Duo practice
 - Group practice
 - Community Health Center
 - Other
-
- 5) Degree of Urbanization of Practice Area:
- Metropolitan area (>100,000 residents)
 - City (30,000 – 100,000 residents)
 - Small Town (5,000 – 30,000 residents)
 - Rural area (<5,000 residents in largest village)
-

Supplementary materials and methods

Details of background of Materials and Methods; Measurements of genetic consultation skills through standardized patients and Regression Analysis

Methods

Measurements of genetic consultation skills

Standardized Patients

Visits

Twelve experienced SPs (5 male/7 female) were trained by JJR and EJFH to each play one of three oncogenetic cases. All of the four SPs trained to play the breast cancer case were female, while one of the familial colon cancer case SPs and two of the melanoma case SPs were female. They received a comprehensive written account of the clinical scenario and a full briefing of their role. Thereafter, a training session with two of the researchers (EJFH and JJR) clarified the purpose of the project and the standardized roles. The three oncogenetic cases were comparably recognizable and urgent to be able to train all the SPs the first day together on presentation and checklist items. The second training day the SPs were trained separately for each one of the three cases on specific clinical history. In case the GP would indicate to proceed to a physical examination, the SPs handed over to the GP a standardized leaflet summarizing the main PE findings fitting the specific oncogenetic case (see eTable 1. Description of Cases presented by Standardized Patients).

At each measurement time (T0, T1 and T2) each GP was visited by one SP. Each SP presented a different case. The GPs were therefore confronted with three different standardized cases, played by three different SPs. The SPs were blinded to whether the GP belonged to the intervention or control group. The GPs were informed an SP would consult during office hours; however they were unaware of exact date and time. After telephoning the office assistant the SP made a standard ten-minute appointment, insisting that their identity would not be disclosed to the GP; however they revealed their identity on entering the GP's office. At the beginning of each simulated encounter, the SPs gave instructions asking the GPs to conduct the consultation authentically. Immediately after each visit, the SPs completed a uniform, predefined checklist to assess GPs' genetic consultation skills.

Cases

Experts in family medicine, clinical genetics, and education selected the oncogenetic cases and wrote the clinical scenarios (EJFH, JJR, EJvdJ, CvdV) using the following criteria: relatively high prevalence in family practice; diagnostic features identifiable through family history and family tree drawing (eTable 1); suitability for a discussion on efficiency considerations (efficient referral to clinical genetics specialist and timeliness of referral); suitability for scoring with one uniform checklist; the presence of important features for physician education; feasibility of realistic SP performance; and coverage of a broad spectrum of oncology cases commonly presented in family practice.²³ Using comparable case scenarios according to the criteria mentioned, enabled comparing potential changes in checklist scores between the three different measurement times.

Checklist

The research team developed a 28-item checklist to quantify evidence judged necessary to assess GPs' genetics consultation skills reflecting training outcomes' covering the full scope of good practice consultations: key ingredients related to family history taking, genetic risk assessment, and referral to genetics specialists. To validate the checklist, three experts in family medicine education and four clinical geneticists evaluated the applicability of the items and cases (eTable 2). Items judged not suitable for scoring (i.e. less than 75% of experts agreed on inclusion) were removed. Items were rated as "yes, observed" (score=1), "no, not observed at all" (score=0), or "?, unclear" (not scored = missing). The scores were summed and the proportion of the maximum possible score (1 for each item, ignoring the missing items²⁴) was determined.

Training of standardized patients and finalization of checklist

One month before the first office visit, 12 SPs were trained, in groups of four, to play the three cases (one case consistently per group of 4) and score the checklist on GP's performance consistently. Training took 16 hours spread over two days. Realistic portrayal was promoted by SPs practicing ten-minute consultations with ten real GPs (not training participants) in a studio in a GP practice setting. Training focused particularly on the use of introductory phrases and on helping them avoid giving away clues as to their role. SPs were also trained to fill in the checklist, to be completed immediately after a consultation during the trial, and their colleague SPs practiced their same role. Their checklist scores were compared with those of the SP trainers (EJFH and JJR), the supposed "gold standard". The practice-consultations were videotaped, scored, and judged to have sufficient face validity for each SP by a panel of four GPs who did not participate in these training sessions and were not otherwise involved. Differences in rating between the panel members were discussed and final alterations made to the measurement scale and

checklist. The panel and the SPs independently re-rated the transcripts using the modified checklist. Agreement between SPs and panel ranged from 87% to 98%, with Cohen's kappa 0.87, which was deemed satisfactory for commencement of the practice visits.

Regression Analysis

Regression analysis was used to investigate improvement of genetic consultation skills immediately after the course using the checklist scores at T1 (ScoreT1) as dependent variable and the checklist scores at T0 (ScoreT0) and the indicator variable Train (0: control group; 1: intervention group) as independent variables. To improve interpretation and numerical stability, the independent variable ScoreT0 was centered and the resulting variable Score T0, equal to ScoreT0-Mean (ScoreT0) was used in the analysis.

Each participant was rated on one of the three SP cases at T0, T1, and T2. As the roles might vary in difficulty and the case order varied between participants, we corrected for these differences by extending the regression equation with four terms corresponding to four indicator variables (values: 0, 1), the dummy variables D_1 - D_4 . This suffices to represent the effects of the five possible different role pairs in comparisons at T1 and T0. Role pair 1 is indicated by ($D_1=1$, $D_2=D_3=D_4=0$), role pair 2 by ($D_2=1$, $D_1=D_3=D_4=0$), etcetera, and role pair 5, is indicated by ($D_1=D_2=D_3=D_4=0$).

Thus, the effect of the intervention was assessed using the model:

$$\text{ScoreT1} = C + b_1 \text{Train} + b_2 \text{ScoreT0} + b_3 D_1 + b_4 D_2 + b_5 D_3 + b_6 D_4 + E$$

In the model C (Constant) represents the intercept of the regression equation, i.e. the predicted score at T1 for a participant in the control group (Train=0) with a score at T0 equal to the mean score (Score T0=0), and being visited by role pair 5 ($D_1=D_2=D_3=D_4=0$). Regression coefficient b_1 represents the effect of the intervention (Train=1), i.e. the increase of the score at T0 due to a participant being a member of the intervention group. Coefficient b_2 is the effect of the pretest (Score T0), b_2 Score T0 being the part of the score at T1 that can be predicted from the score at T0. Regression coefficients b_3 - b_6 indicate the increase of the score at T1 when a participant was visited by role pair 1, 2, 3, or 4, respectively, instead of being visited by the reference role pair (role pair 5). Term E (Error) is the part of the score at T1 that cannot be explained by the predictors in the regression model (the residual).

In a similar procedure, using ScoreT2 as dependent variable, retention of knowledge was analyzed.

The regression coefficient b_1 corresponding to Train represents the net gain in performance (expressed as proportion of the maximum score) due to the intervention. The corresponding standardized regression coefficient indicates the effect size. According to Cohen's categorization (Cohen, 1988) values 0.1, 0.3, and 0.5 indicate small, moderate, and large effect sizes, respectively.

The mean checklist scores and corresponding 95% confidence intervals were calculated for the two groups at T0, T1, and T2. Because the raw means are not corrected for varying role orders, the differences of the raw means may differ considerably from the intervention effect found in the regression analysis. It is important to note that the intervention effect inferred from the raw means is biased, whereas the regression analysis provides an unbiased estimator.

Satisfaction with the intervention was analyzed by calculating mean scores, 95% confidence intervals, and standard deviations for the pooled data from the satisfaction questionnaire. The data from the applicability questionnaire were analyzed in the same way. All analyses were performed using SPSS version 19 (SPSS, Chicago, IL).

Chapter 6

Effect of comprehensive oncogenetics training interventions for primary care physicians on multiple performance levels

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Submitted

Abstract

Purpose

General practitioners (GPs) are increasingly called upon to identify patients at risk for hereditary cancers. Therefore, their genetic competencies need to be enhanced. Three oncogenetics modules were developed based on the priorities identified: an online Continuing Professional Development (G-eCPD) and live genetic CPD module, and a website (*huisartsengenetica.nl*) with supporting genetics information applicable in daily practice. The study aimed to determine: 1) long-term (self-reported) genetic consultation skills (i.e. increased genetics awareness and referrals to Clinical Genetics centers) among GPs participating in the training, and 2) interests in and satisfaction with the website.

Methods

1. A (1-year follow-up) online questionnaire on self reported applicability of genetic competences and on change in referral behavior.
2. Referral numbers from GPs to Clinical Genetics centres.
3. Satisfaction questionnaire and visitor count analytics of genetics website.

Results

The genetics CPD modules achieved sustained improvement of oncogenetic knowledge and consultation skills. Participants reported to be more alert of genetic problems. 68% of the respondents who attended the live training reported to more frequently refer patients to the Clinical Genetics centers, compared to 29% of those who attended the online oncogenetics training. No significant change in referral numbers however was reported by the Clinical Genetics centers one year after the training. Website visitor numbers are still increasing. The page most often consulted is “family tree drawing”.

Conclusion

Self-perceived genetic consultation skills increased long-term and there was interest in and satisfaction with the supportive website. The study results presented suggest we have provided an adaptable and effective framework to answer to the need for effective educational programs for non-genetic healthcare-providers enabling improvement of genetic medical care.

Trial Registration: trialregister.nl Identifier: NTR3322 and NTR3323

Introduction

Genomic innovations are increasingly applicable in daily medical care. General practitioners (GPs) are confronted with challenging genetic information, patients' requests for genetic tests and its diagnostic and therapeutic consequences. For successful implementation of genetic innovations several barriers have to be overcome, including the fact that physicians lack knowledge of genetics relevant for daily practice, lack oversight of genetic testing, and report inadequacy to deliver genetic services.^{1,2} For genetics to have an effect on clinical practice comparable to its impact on research will require genetic literacy of health-care providers.³

Physicians in general wish to be educated in a practical manner, which means genetics education should be applicable in daily practice through exploiting case based learning.⁴⁻⁶ Genetic core competences for non-genetic health care workers have been developed⁷⁻¹⁰. Competences in three domains are needed: cognitive (knowledge), psychomotor (skills) and affective (attitude).¹¹ Combining the educational competences in training is regarded to have more impact on a clinical situation than training competences separately.¹²

We set out to provide an adaptable and effective framework for genetics education for primary care health physicians based on multiple methods and assessable at the highest possible level of evaluating the learning process and its effects on genetic performance in daily practice. We started by exploring the needs and the role of genetics in primary care and assessed priorities in genetic education mentioned by GPs.^{5,6} Top three of prioritized genetic competences were "Recognizing signals that can indicate a hereditary component of disease", "Evaluating indications for referral to a Clinical Genetics center", and "Knowledge of the possibilities and limitations of genetic tests".⁶ It was expected that training focusing on these topics would lead to higher quality consultations between medical professionals and patients, reflected in timely referrals to the specialized departments of Clinical Genetics. The genetic competences were applied in oncology, as this was the yearly theme of the Dutch College of General practitioners (NHG).

Based on the priorities and integrating genetic core competences we developed three training modules executed by the NHG (Department of Education):

1. a Genetics online Continuing Professional Development on oncogenetics,
2. a live training (interactive program taking oncogenetics as a model condition), and
3. a supportive website (www.huisartsengenetica.nl, "*GP and genetics*").^{13,14}

The website was developed and is kept up-to-date by the research team in collaboration with the Erfocentrum (Dutch Information Center on heredity and genetic disorders) and NHG, with on demand supportive information to be able to work on the learning tasks and apply genetic competences in daily general practice. The easily accessible website gives GPs on demand information on e.g. basic genetics information and how and which Clinical Genetic center to refer to.

To our knowledge, this was the first time a series of oncogenetic modules were organized and evaluated based on prioritized topics and effects on genetic performance were assessed.

Kirkpatrick's framework for evaluation educational outcomes

For effect evaluation we considered Kirkpatrick's framework for evaluating educational outcomes, originally presented in 1967, describing four levels: valuation (level 1; satisfaction), learning (level 2; knowledge and knowledge retention), behaviour (level 3: applied knowledge and consultation skills on timely recognition of patients at risk) and effects on patient health and organization (level 4: change in actual practice performance (i.e. referral) and results).¹⁵ Also the impact on society, or patient safety in genetic medical care would be part of level 4 (Figure 1). We used Moore's model for CPD curriculum design identifying individual learning steps with its educational objectives and the Kirkpatrick framework as a model to evaluate our oncogenetic modules (table 1).¹⁶

Table 1. Matrix levels of oncogenetics modules evaluation according to Kirkpatrick and Moore (Adjusted to Davis et al., 2008 ⁴)

Kirkpatrick/Moore levels of Education and Evaluation	Kirkpatrick Definition	Oncogenetic module format	Assessment	Educational objective
I	Satisfaction	G-eCPD, ¹³ live training, ¹⁴ supportive website	Satisfaction surveys and website visitor count	Information, comprehension
II	Knowledge, Self-reported applicability of newly learned consultation skills	G-eCPD, ¹³ live training ¹⁴	Multiple choice questions, open ended questions, Vignettes: Pre/post-and retention-test	Information, comprehension
III	Behavioral change	Live training ¹⁴	Responses to SP- encounters in actual practice: Pre/post- and retention-assessment	Synthesis, application, performance, attitude
IV	Organizational change, health gain	G-eCPD, ¹³ live training, ¹⁴ supportive website	GP Referral data of Clinical Genetics centers	Analysis, synthesis, evaluation: health gain through timely (increased) referral to Clinical Genetics centers

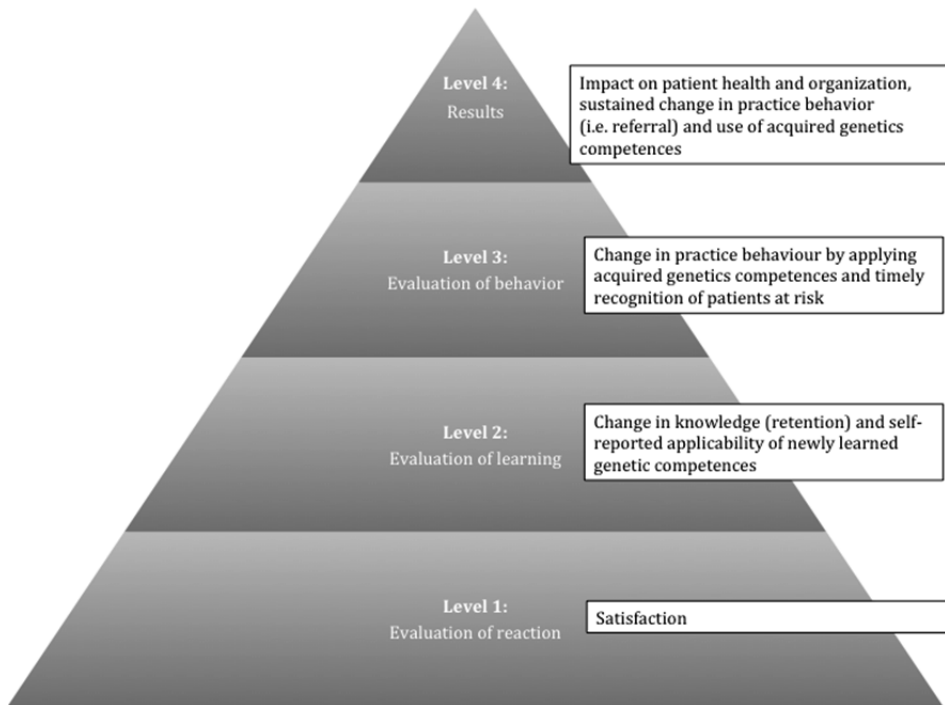


Figure 1. Genetic educational framework;
Based on Kirkpatrick's Evaluation Framework for Educational Outcomes^{15,17}

After the needs-based development and shorter-term evaluation of our genetic education modules, the goal was to answer the following research questions:

1. To determine long-term genetic consultation skills among GPs participating in the training by assessing self-reporting skills (i.e. increased genetics awareness and referrals to Clinical Genetics centers), and by comparing referral data from GPs to Clinical Genetics centers before and after training. An increase in number of referrals was expected to reduce the number of missed cases.¹⁸
2. To determine interest in and satisfaction with the website, website visitor count was analyzed and satisfaction with the website was determined through a pop-up questionnaire. Visitor count was expected to increase with oncogenetic programs organized and increasing media attention such as links on social media, newsletters and newsflashes in the media.

Materials and methods

Four instruments were used to answer the research questions:

1. An online questionnaire for determining long-term self-reported genetic consultation skills was emailed to those who had previously attended the oncogenetics CPD modules. See online-only material contents for Details of background of Materials (Questionnaire to determine self reported applicability of an Online Continuing Professional Development (G-eCPD) module and a Live training).
2. Referral numbers by GPs were requested from the Clinical Genetics centers, in the Northern and Southern parts of the Netherlands (VU University Medical Center (VUMC), Amsterdam and Maastricht University Medical Center (MUMC), Maastricht, The Netherlands) from the two years before (2010 and 2011) and the year after (2012) launch of the oncogenetics CPD modules and website *huisartsengenetica.nl* ("GP and genetics"). Increase in referral numbers was presumed to estimate short-term improvement of GPs' synthesis and application of the newly-learned oncogenetics knowledge and consultation skills with increased awareness of oncogenetics problems in daily practice.
3. Website visitor analytics roughly determine GPs' sustained interest in the supportive website one year after introduction, suggesting change in organization and consequently health gain. Visitor count was expected to go up with oncogenetic programs organized and increasing media attention such as links on social media, newsletters and newsflashes in the media.
4. To more specifically determine whether there was an interest in and satisfaction with the website, an online pop-up questionnaire was requested for one month when visitors would visit the website. See Online Only Text for Details of background of Materials (Website Satisfaction questionnaire).

The ethical review boards of the Netherlands Association for Medical Education, Maastricht University Medical Center, and VU University Medical Center Amsterdam, the Netherlands approved the study protocols. All participants gave written informed consent before the trials.

Participants

The project team collaborated with the NHG on providing genetics CPD modules.^{13,14} The GPs who previously participated in these studies, working full or part time in family practice, were all followed up to participate long term in the online questionnaires. Recruitment for participation in the live oncogenetic CPD module was limited to GPs practicing in two Dutch provinces where the previously held live training sessions were given (n=88). The online oncogenetics training

required recruitment of participants outside these two provinces to be able to assess effects separately (n=80).

Participants of the website evaluation were all recruited online, when they visited the website a pop-up invitation would come up. The pop-up came up during the whole month of February 2013.

Analysis

The answers to the online questionnaire (effects of online and live training) and the pop-up questionnaire (appreciation of the website) were investigated in a similar way. For ease of interpretation the 5-point scales were transformed into 2-point scales (binomial) by merging the lower three categories (disagree completely, disagree, not disagree /not agree) and the upper two (agree, agree completely). The proportion answers in the upper category of the binomial scale and the associated 95% confidence interval were calculated to indicate the effect of the training as reported by the GPs, and the appreciation of the website by its visitors.

For the 10 point global rating the mean score and the associated 95%-confidence interval were calculated to indicate the respondents' level of appreciation of the genetics website (the third training module).

Website visitor counts were obtained by Google Analytics in the period September 2011 until March 2013. Time series for the Number of visits per month, the Number of pages viewed per month, and the Number of pages per visit per month were obtained. Where relevant the mean trend in the time series was estimated by fitting a straight line (linear regression) to the data, and using the slope of the line as indicator of the trend.

Data were analysed using SPSS20.

Results

Two oncogenetics CPD modules were developed aimed at improving competences:

1. A G-eCPD module aimed at improving GPs' oncogenetics knowledge, and
2. A four-hour interactive live training module covering oncogenetic clinical skills (family history, risk assessment, and efficient referral).^{13,14}

Two parallel-group pre-post-retention (6-month follow-up for G-eCPD, 3-month follow-up for live module) controlled group intervention trials (standardized patients, checklists and validated questionnaires) were conducted to assess effectiveness of the CPD modules developed. 168 GPs working in the Dutch primary care setting responded to an email invitation and were randomly assigned to intervention or control groups, evaluating the G-eCPD module (n=80, 44 GPs completed all measurements) or the live module (n=88, 56 GPs completed all measurements).

Results showed there was a significant follow-up improvement in oncogenetic knowledge (G-eCPD) and consultation skills (live module) after the intervention. Satisfaction and self-reported applicability was high for both modules.^{13,14}

GPs self-reported skills and referral

Participant characteristics

42 GPs (52%) who participated in the G-eCPD evaluation study and 50 GPs (57%) who participated in the live training program, responded to the online questionnaire on long-term effects on Kirkpatrick's second level of educational outcome. 88% of the respondents who attended the live training reported to more frequently consider referral of patients to the Clinical Genetics centers, compared to 64% of the respondents who attended the online CPD module. Respectively 68% and 29% reported to actually refer patients more frequently (table 2).

EFFECT OF COMPREHENSIVE ONCOGENETICS TRAINING INTERVENTIONS

Table 2. Self reported applicability of an Online Continuing Professional Development (G-eCPD) module and a Live training on Oncogenetics by GPs who participated in one of these CPD modules

Statement/Question	Response Category	Online Continuing Professional Development module (G-eCPD)				Live Interactive Program on Oncogenetics			
		Total number of respondents	%	95%-CI ¹		Total number of respondents	%	95%-CI	
				lo	hi			lo	hi
I am more alert on genetic problems	Agree, Agree completely	42	69	53	82	50	92	81	98
I have treated more patients with genetic problems	Agree, Agree completely	42	12	4	26	50	46	32	61
I have more frequently considered to refer patients to the Clinical Genetics department	Agree, Agree completely	42	64	48	79	50	88	76	96
I have more frequently referred patients to the Clinical Genetics department	Agree, Agree completely	42	29	16	45	50	68	53	81
I am better able to explain possibilities/limitations of genetic tests to patients	Agree, Agree completely	42	50	34	66	50	72	58	84
How frequent do you use the genetics website	Once upto Daily	38	18	8	34	47	47	32	62
Will you keep on using the genetics website	Yes	31	81	63	93	25	96	80	100
Did you ever consult the genetics website when referring patients to the Clinical Genetics department	Yes	7	71	29	96	22	91	71	99
I would recommend the genetics website to my colleagues	Agree, Agree completely	7	86	42	100	22	91	71	99
Global rating of the genetics website	10-point	7	7.7	7.0	8.4	22	8.1	7.8	8.3

¹95%-CI: 95%-Confidence Interval

Changes in referral to Clinical Genetics centers

In table 3 the results of the referral rate from GPs to Clinical Genetics centers found through a search in the ICT system at the Clinical Genetics centers in Amsterdam and Southern part of the Netherlands are shown for the years 2010-2012. No change in number of referrals was seen in the year after presentation of the oncogenetic modules and website.

Table 3. GP Referral rates at the Clinical Genetics Medical Centers in the Northern and Southern part of the Netherlands the years 2010-2012.

Site	Year		
	2010	2011	2012
Maastricht University Medical Center	1549	1590	1508
VU Amsterdam Medical Center	961	1350	1367
Total	2510	2940	2875

Website interest and satisfaction

38 visitors (12 (32%) aged 31-40 years, 27 (71%) female) to the website answered the popup questionnaire (results of the questionnaire in table 4). Figure 2 (upper panel) shows website visitor numbers are steadily increasing each month with almost 60 new visitors. The percentage of returning visitors (Figure 2) is stable around 20% each month demonstrating sustained interest in the website. Website visitor analytics showed a top 10 of most frequently visited webpages: drawing family trees, hereditary diseases, family history taking and consanguinity and pregnancy wish pages suggest increased application of genetic knowledge and consultation skills conceivably reflecting increased genetic health.

EFFECT OF COMPREHENSIVE ONCOGENETICS TRAINING INTERVENTIONS

Table 4. Self reported satisfaction and applicability of the genetics website by general visitors and GPs only.

Statement/Question	Response Category	All respondents (resp.)				GPs only			
		Total number of resp.	%	95%-CI ¹		Total number of resp.	%	95%-CI ¹	
				lo	hi			Lo	hi
Is this your first visit to the genetics website	No	38	21	10	37	22	18	5	40
The content of the website is helpful	Agree, Agree completely	19	68	44	87	10	70	35	93
The content of the website is up to date	Agree, Agree completely	19	68	44	87	10	70	35	93
The content of the website is apprehensible	Agree, Agree completely	19	74	49	91	10	70	35	93
The content of the website is up to my expectations	Agree, Agree completely	19	63	38	84	10	70	35	93
The content of the website is attractive	Agree, Agree completely	19	53	29	76	10	60	26	88
The content of the website is up to professional standards	Agree, Agree completely	19	74	49	91	10	80	44	98
The content of the website is clearly structured	Agree, Agree completely	19	63	38	84	10	70	35	93
The content of the website is simple to use	Agree, Agree completely	19	58	34	80	10	70	35	93
I would recommend the genetics website to my colleagues	Agree, Agree completely	19	68	44	87	10	80	44	98
Was your current visit successful	Completely, or partly successful	19	84	60	97	10	100	69	100
Global rating of the genetics website	10-point	38	6.3	5.5	7.1	22	7.2	6.5	7.9

¹95% -CI: 95%-Confidence Interval

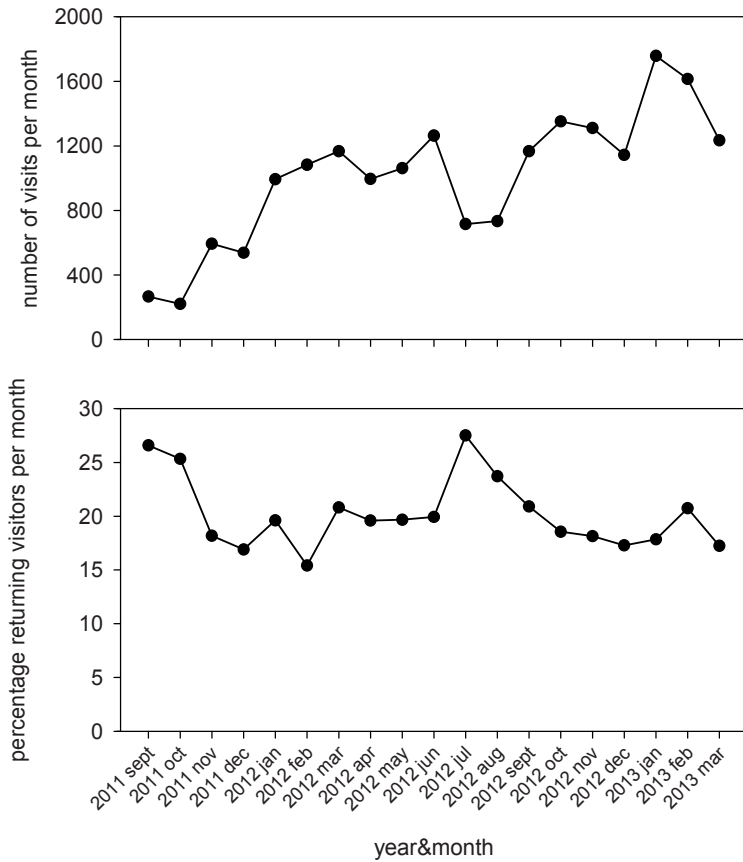


Figure 2. Website visitor numbers and percentage returning visitors per month

Discussion

Self-reported genetic consultation skills increased long-term, including increased consideration of referral to Clinical Genetics centers. However, regional GP referral numbers did not change accordingly the year after oncogenetic CPD module presentation. A reason could be the limited number of GPs who participated in the oncogenetic CPD modules. Also, these CPD modules' topic was specifically oncogenetics, GPs referral numbers may increase if genetics CPD modules also involve other topics such as cardiogenetics, diabetes or reproductive genetic counselling. GPs are gatekeepers and play a key role in the Dutch health care system when appropriate and timely referral to medical specialists is warranted. Their role could be enhanced with increased awareness and alertness if more GPs are effec-

tively educated.¹⁸ There could also be a difference in concept, for improved genetics knowledge and consultation skills should not necessarily result in more referrals. This improvement could also lead to more accurate referrals and could consequently show a decrease in number of referrals. Direct measurement of change in referral numbers or efficiency and effectiveness of referral at this time however was impossible, since the ICT tools to register family history in the Electronic Health Registry is not in place.

Although there was only a small number of website visitors who answered the pop-up questionnaire, those who did indicated to be interested in and satisfied with the supportive website. This converged with long-term website visitor counts and percentage returning visitors estimating adequate website user-friendliness and usability. Website visitors often looked for application of basic genetics (drawing family trees, family history taking), while initially we had not expected needs for these topics. However, at the time no website visitor analytics were available from comparable websites. Some website visitor analytics were available from websites with information aiming for (non) genetic medical professionals, general public, patients, families and others, which is incomparable to the data provided on the supportive website *huisartsengenetica.nl*, which mainly aims to inform GPs. With 8747 GPs actively working in the Netherlands, around 1800 visitors (presumably GPs) returning each month is promising. Updating the website is an ongoing process which should sustainably increase website visitor numbers. Future studies could possibly translate the website for GPs internationally and where needed (such as specific information on referral and insurance) adapt the information to that specific country.

The study results presented suggest we have provided an effective framework to answer to the need for effective genetic educational programs for non-genetic healthcare-providers. The training proved to be a feasible and satisfactory method to achieve long-term improvement of applicable oncogenetic knowledge and consultation skills. This educational framework can inform future training activities for GPs and potentially other medical professionals to enhance genetics-related consultation and decision making with the ultimate aim of improving medical care.

Comparison with other studies

A meta-analysis, which examined the effect of moderator variables on physician knowledge, performance and patient outcomes showed similar results compared to our study. It suggested a larger effect size of CPD outcome in case of interactive interventions, using multiple methods, designed for small groups of physicians from a single discipline.¹¹ However, the meta-analysis showed a negative correlation between effect size and the outcome assessment time ($r=-0.31$). This would imply, increasing assessment time between CPD intervention and impact evalua-

tion could impair sustainability of learned genetic competences. Longer-term outcome assessment of the oncogenetic training program on referral numbers and change in organization would need further study to be able to say anything about true impact on patient genetic health. Possibly repeating the oncogenetics training programs on a yearly basis for local GP groups, may show better outcome effectiveness longer term. Effective impact of new implementations of change in patient care is possible to change referral rates or improve outpatient referral appropriateness.¹⁹ However, limited rigorous evaluations on referral processes from primary to secondary care are available to base policy on.²⁰ A few promising interventions which we incorporated already in our live training were found to positively influence the referral process, such as local educational intervention involving secondary care specialists and structured referral sheets. This will need further studies in case of referral to Clinical Genetics centers. Clear guidelines for referral should be distributed and should be part of training. Genetic counsellors may also be beneficial to the referral process for they can provide a second opinion before referring to the specialist. Financial compensation to optimize referral has been studied, but there is insufficient evidence to draw firm conclusions.²⁰

Methodological considerations

A model for the development and evaluation of genetics education programs was previously described and was informed by three theories: *adult learning theory*, *program logic modeling and evaluation theory*.²¹ In short, *adult learning theory* proposes for an education program to be effective, it should be responsive and enable learners optimally. Recognizing a need for learning about a certain topic, was previously accomplished.^{5,6} The first steps towards an effective genetics education program were taken involving the GP learners, focusing on the content and process, using a range of multifaceted teaching strategies namely experiential and interactive.

Program logic modelling is a theory useful to define and plan program design and possible evaluation, which inspired the previously held Delphi study.⁶ With the use of experts topics for a genetics program were prioritized based on the previously held focus group studies.⁵ The Delphi study was used as a tool to develop our hypothesis that with the use of experts a top ten of genetics topics applicable in daily GP practice could be found which would inform further development of effective genetics education. By letting the health professional participate in the education program by assessing their learning needs, awareness of the relevance of genetics in daily practice was considered to increase and GPs were expected to learn best and apply learnt competences accordingly in daily genetic medical care.

The *evaluation theory* rigorously determines impact of the training programs. Beneficial effects on process outcomes are usually shown (e.g. increase of genetic

competences), however evaluations of patient outcomes (e.g. genetic health care improvement) are often lagging behind. Several terms explaining genetic educational program evaluation apply, to ultimately make evaluation of patient outcome possible: *formative evaluation* (needs assessments by stakeholders involved). The second term *process evaluation* explains the manner in which a genetic educational program is implemented in daily practice (i.e. family history taking and registration in GP computer information system) and whether it reaches its intended audience on time (i.e. pre-symptomatic referral to the clinical geneticist). The last term refers to *summative evaluation*, which sums up the impact of the program on the users and audience involved (i.e. improved genetic competences, awareness and behaviour in trained GPs and genetic healthcare outcome).²¹ Results of the summative evaluation reported, show that applying the adult learning theory and program logic model, in our project assessing the needs and subsequently defining the program design, is effective.

Strengths and limitations

Voluntary participation by interested GPs could have caused selection bias. However, similarity of the baseline characteristics of the two groups in the RCTs and comparability of the participation rate (previous RCTs 55% (G-eCPD) and 60% (live training), for this study 52% (G-eCPD) and 57% (live training)) with that of other studies among GPs (60%) indicate that the participants were representative of GPs likely to attend oncogenetic training in the future.^{13,14,22,23} Furthermore, it is possible that participating in the genetics training might become part of standard training for all GPs. By applying an effective framework for genetics education and measuring outcome of education on various levels of Kirkpatrick, we were able to initiate a change in organization and find barriers to implementation of genetics education.²⁴ Further assessments are necessary however to make additional assumptions on patient health impact. Rigorous assessment of appropriate referrals was not feasible within the project, for data were based on self-reported competencies, increase in referrals and website analytics, not registered referrals in Electronic Patient Record (EPR). We plan to assess change in referral to the departments of Clinical Genetics more rigorously by looking at the registered number of referrals in the EPR based on added ICPC codes.²⁴ Furthermore, to truly assess user-friendliness and usability of our website we plan to assess the website through qualitative research methods.

Conclusion

The results presented provide an adaptable and effective framework for genetics education of health professionals possibly across national borders. The suggested training tools guide future development of curricula that are appropriate to the national context, educational system and healthcare setting of the professional involved. It is therefore possible, to optimize genetic educational materials as a multifaceted approach to implementing the genetic educational needs and prioritized topics and genetic education core competences. This educational framework therefore has the potential to improve the quality of genetic healthcare for patients.

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CHAPTER 6

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Online-only text

Details of background of Materials-Questionnaire to determine self reported applicability of an Online Continuing Professional Development (G-eCPD) module and a Live training

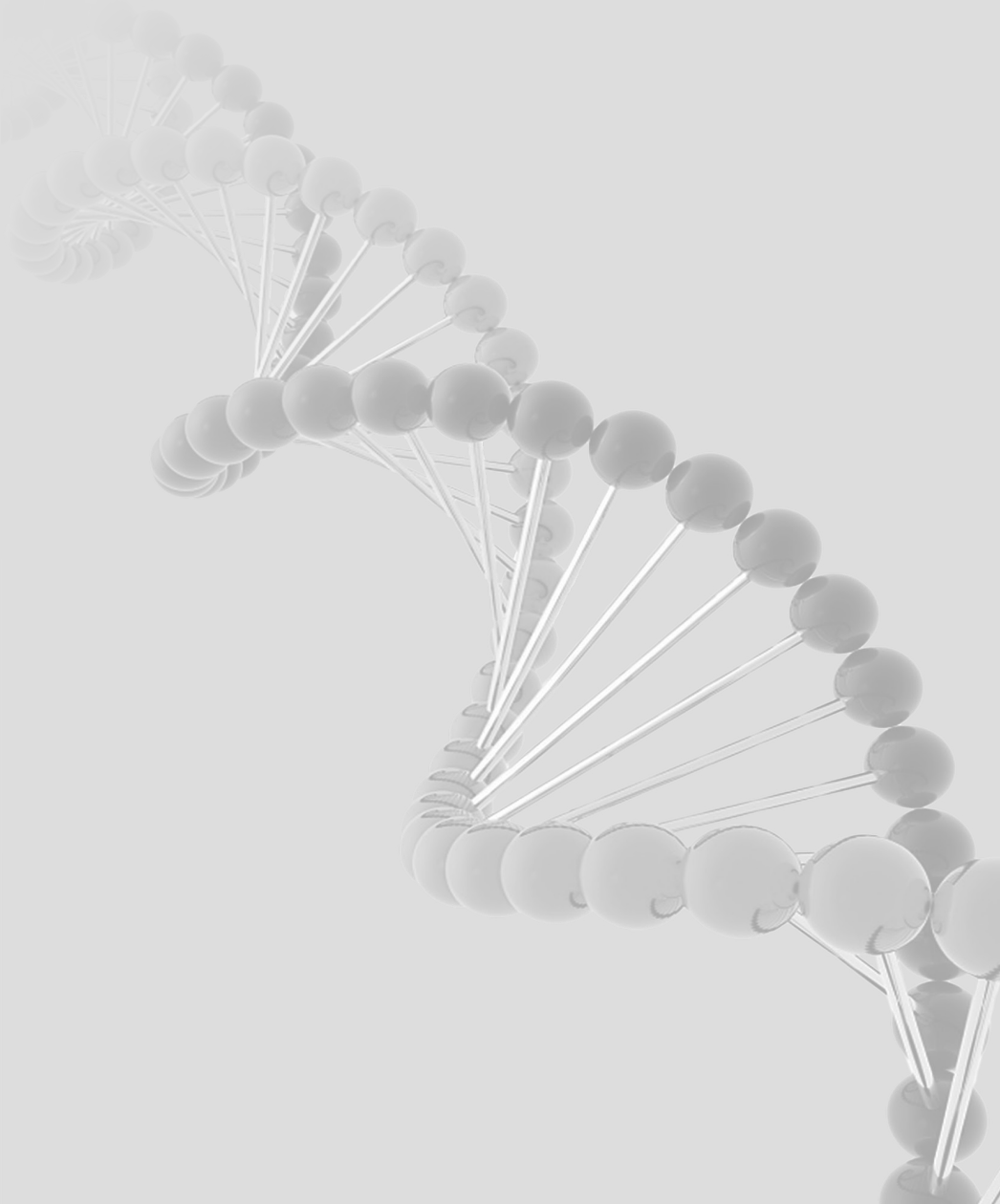
An online questionnaire was emailed to those who had previously attended the oncogenetics CPD modules determining long-term self reported genetic consultation skills (i.e. increased awareness of possible genetic predisposition of diseases, discuss (possible) familial and hereditary disease risks, management of potentially developing hereditary diseases, more frequently consider referral and concrete referral to Clinical Genetics centers and explain the possibilities and limitations of oncogenetic testing. The questionnaire contained five items (with a 5-point Likert scale: 1=completely disagree; 5=completely agree) related to different aspects of, self-reported applicability of genetic competences. A global grading of the website on a ten-point scale (1: no value; 5: insufficient 6: sufficient; 8: good 10: excellent), and a question about the frequency of using the website. The applicability questionnaire contained two 4-point scale items about the need for the supportive website and application of the website on referral in daily practice and a multiple-choice question about which pages were used to benefit referral to Clinical Genetics center. One question was on whether the participant would recommend the website to colleagues (yes/no).

Details of background of Materials-Website questionnaire determining self reported satisfaction and applicability of the genetics website by general visitors and GPs only.

The satisfaction questionnaire contained four items on website content (helpful, recent information, understandable language, responding to expectations) and four on usability (attractiveness, professional, structure, easy to use) on a 5-point Likert scale (1=completely disagree; 5=completely agree) related to different aspects of satisfaction, a global grading of the website on a ten-point scale (1: no value; 5: insufficient 6: sufficient; 8: good 10: excellent), and a question about the frequency of using the website. One question was on whether the participant would recommend the website to colleagues (yes/no). The demographic survey asked about participants' general characteristics (male/female; age; professional background (GP, midwife, medical specialist, medical student, patient/other).

Part III

Summarizing discussion and future perspectives



Chapter 7

Proposed roadmap to stepwise integration of genetics in family medicine and clinical research

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Abstract

We propose A step-by-step roadmap to integrate genetics in the Electronic Patient Record in Family Medicine and clinical research. This could make urgent operationalization of readily available genetic knowledge feasible in clinical research and consequently improved medical care. Improving genomic literacy by training and education is needed first. The second step is the improvement of the possibilities to register the family history in such a way that queries can identify patients at risk. Adding codes to the ICPC chapters “A21 Personal/family history of malignancy” and “A99 Disease carrier not described further” is proposed. Multidisciplinary guidelines for referral must be unambiguous. Electronical patient records need possibilities to add (new) family history information, including links between individuals who are family members. Automatic alerts should help general practitioners to recognize patients at risk who satisfy referral criteria. We present a familial breast cancer case with a BRCA1 mutation as an example.

Introduction

Public health benefits of advancements in understanding the human genome are still to be realized for common chronic diseases such as cardiovascular disease, diabetes mellitus, and cancer¹. International attempts to integrate and operationalize such knowledge into clinical practice are in the early stages, and as a result, many questions surround the current state of this translation.¹⁻³ Most physicians lack genetic knowledge and skills that might be relevant for decision support in daily practice.⁴ Family history taking and family tree drawing need to be introduced. Oversight of clinical utility of genetic testing should be supported by eHealth facilities to bypass unfamiliarity with facts on genetic testing. Shortcomings in registration systems and inadequate implementation of genetics in existing guidelines are reported and result in inability to register genetic information in Electronic Patient Records. Privacy and risk of discrimination cause concerns when registration is considered. Consequently, inadequacy to deliver genetic services is reported in literature.¹ We present a roadmap to integrate actual genetic knowledge into the Electronic Patient Record and into clinical research in Family medicine, which would enable urgent operationalization of readily available knowledge feasible in daily genetic medical care.

Evidence for necessary change

The clinical relevance of integrating genetics in clinical practice was demonstrated for several familial diseases such as colorectal cancer and breast cancer. Dove-Edwin et al. calculated mortality risk reduction up to 80% by identifying and subsequently screening individuals with an increased familial colorectal cancer (CRC) risk.⁵ Cancer risk management options through genetic testing for BRCA mutations and subsequent options for preventive surgery after testing positive can empower women and can also reduce morbidity and mortality.⁶ Currently, a large number of patients in whom screening would be beneficial, are out of sight or being missed by their physicians.^{7,8}

Barriers to change

Scheuner *et al.* identified deficiencies in primary care workers' basic genetic knowledge and ability to interpret familial patterns.¹ This is in line with our prioritised educational topics, including knowledge of basic genetic principles, the most common genetic disorders and family history communication skills.⁹ Taylor and Edwards stated primary care should be encouraged to invest more time and energy in questioning and registering family history data.¹⁰ However, they also stressed identified barriers such as time constraints should be encountered. They

identified the need to develop strategies to overcome difficulties preventing general practitioners (GPs) from routinely obtaining family history information as well as strategies to support accurate record keeping in the Electronic Patient Record.¹⁰

Another identified barrier is the presence of ambiguous referral guidelines to clinical genetics and other medical specialists for patients with a possible high risk at familial disease, such as cancer.⁹ Computerised decision support might be helpful in familial risk assessment for common cancers (e.g. breast, ovarian and colon cancers) and would render timely genetic risk assessments and consequently support referrals more consistent with guidelines. These results support the implementation of genetics education aimed at enhancing effective referral indications and options.

A roadmap for translation

In order to be able to truly turn useful genetic discoveries from the laboratory bench to daily clinical practice, a roadmap is crucial to make urgent translation feasible. First, advances in the genomic literacy of health care providers are indispensable. Secondly, innovative and practical ICT tools to apply these newly acquired knowledge and skills are needed, such as registration of family history and registry alerts supporting this.

We propose a step-by-step roadmap to effectively integrate genetics in daily family medicine to its full potential:

Step 1: Improve basic knowledge of genetics in clinicians and develop skills and attitude to obtain and interpret a family history through effective education; For example, training on oncogenetics for GPs was recently developed and evaluated in collaboration with The Dutch College of General practitioners. Also, a website on genetics targeted to GPs was developed to easily obtain information on, amongst other topics, genetic diseases, referral guidelines and family history taking (huisartsengenetica.nl, translated “GP and genetics”). Oncogenetic knowledge, skills and attitude were effectively transmitted through an accredited online and live interactive training and could internationally serve as an example for other common topics (i.e. reproductive medicine, familial coronary heart disease and diabetes) and possibly other medical specialties provided that they are *translated* to its medical systems.

Step 2: Add relevant International Classification of Primary Care (ICPC) codes and other coding strategies for simple registry of family history and develop and sup-

port coding skills;

In order to identify and track persons and/or families at risk for hereditary diseases adequate coding is a starting point. We propose to add a number of codes for simple registration of family history. This will enable and support adequate case-finding and decision strategies.⁸

Proposal for adding codes to ICPC-2 list in case of oncogenetics

We propose to add a number of codes in order to enable simple but structured registry of a family history. In ICPC-2, which is the most frequently used coding system for GPs in Western countries, these codes should be included in Chapter A (General and Unspecified), under A21 "Risk factor for malignancy". ICPC-2 was developed by the WHO and classifies patient data and clinical activity in the domains of General/Family Practice and primary care, taking into account the frequency distribution of problems seen in these domains. It allows classification of the patient's reason for encounter (RFE), the problems/diagnosis managed, interventions, and the ordering of these data in an episode of care structure. ICPC-2 has a biaxial structure and consists of 17 chapters, each divided into 7 components (comp.) dealing with symptoms and complaints (comp. 1), diagnostic, screening and preventive procedures (comp. 2), medication, treatment and procedures (comp. 3), test results (comp. 4), administrative (comp. 5), referrals and other reasons for encounter (comp. 6) and diseases (comp. 7). (see <http://www.who.int/classifications/icd/adaptations/icpc2/en/index.html>)

Mapping is available between ICPC and ICD-10, which was also developed by the WHO for broad application in healthcare registries. The codes suggested below should suit other coding systems such as SNOMED as well.

A21 Personal/family history of malignancy (Existing code)

- A21.1 One or more 1st degree family member(s) with breast cancer
- A21.2 One or more 2nd degree family member(s) with breast cancer
- A21.3 One or more family member(s) with bilateral or multifocal breast cancer
- A21.4 Breast cancer in the family in one or more men

A99 Disease carrier not described further (Existing code)

- A99.1 BRCA-1 mutation carrier
- A99.2 BRCA-2 mutation carrier
- A99.3 TP53 mutation carrier
- A99.99 Carrier of mutation in other specified gene

Step 3: Improve access to up-to-date and unambiguous referral guidelines; For example, in the Netherlands multiple referral guidelines for hereditary cancers were developed independently (OncoLine, Foundation for detection of hereditary

tumors (In Dutch STOET), clinical genetics centers in University hospitals and The Dutch College of General practitioners (NHG)). Limited usable information however is available for General Practitioners, i.e. only for Diagnostics of Breast Cancer and Rectal Bleeding. The guidelines are heterogeneous and difficult to interpret. We propose to improve this by agreeing on national multi-disciplinary referral guidelines and provide synchronized online access to up-to-date and easy to interpret versions.

Step 4: Provide service or online apps to (self) register family history including family relations, that can be coupled with routine healthcare registries and the EMR used in primary care too; The best way to re-use and expand previously recorded family history information and to view this history from the perspective of a different family member is by recording parent-child relations and diagnoses with the correct family member. This would require functionality to be added to the Electronic Patient Record. In order to overcome privacy issues and an online app or website to register family history is recommended (for example: myfamilyhistory.com or familyhealthware.com).

Step 5: Pro-active genetic services integrated in clinical practice facilitated by ICT (for example family history registry and registry alerts); For example, the GP or nurse practitioners should be able to (periodically) register or consult family history information directly into the Electronic Patient Record. Accurate and up-to-date treatment and referral guidelines and subsequent automatic alerts should pop up when certain combinations of symptoms and familial risk factors indicate referral to a clinical geneticist or other medical specialist.

Illustration of the proposed roadmap with a familial breast cancer case in clinical research and family medicine:

Step 1: Patient name: Angela B., Female, age 35.

Angela lives in the city with her husband and two daughters aged 13 and 10. She works as a hair dresser, has been happily married for a decade and the family just bought a new home in the suburbs. She consults the GP on a busy Monday morning with the following complaints: Lump in left breast which she noticed during the weekend. The 4 cm irregular swelling is not painful but rather sensitive. The skin on the swelling is a little red and dimpled. Angela has no medical history, but since you followed the oncogenetic training for GPs a few weeks ago you are aware of the possible familial risks of breast cancer and decide to take her family history. Angela's mother died of breast cancer when she was only 50 years of age 10 years ago. Her mother's father had an unknown cancer and died at age 55. Angela tells you, when you further ask her for her family history, her sister had bilateral breast

cancer at age 30 and died of ovarian cancer at age 33, two years ago. Her two other and younger sisters seem healthy.

On father's side of the family no one has been diagnosed with cancer yet.

Step 2: If proposed codes would be added the following could be registered:

Two first-degree family members with breast cancer at an early age: mother (died at age 50) and sister (age 30, died 33, bilateral breast cancer). : A21.1 and A21.3

One first-degree family member with ovarian cancer at an early age (sister age 30, died age 33).

Step 3: You are alarmed by the family history and the medical complaints of Angela. After checking the referral guidelines for cancer online, you talk with Angela about referral to the closest hospital as soon as possible for further diagnostics and possibly necessary surgical treatment. You also inform her of the chance that she might be a carrier of a DNA mutation which could be further analysed by a clinical geneticist. You promise to call the clinical geneticist and discuss the problem. The clinical geneticist agrees Angela needs further genetic DNA testing based on this positive family history and will invite her this week to quickly start DNA testing, which may inform further treatment. You call Angela afterwards and she is grateful for taking her case so seriously.

Step 4: Angela is alarmed by the fact that her positive family history for breast and ovary cancer could mean an added risk to her and her daughters to develop breast or ovarian cancer and decides to use the online tool to easily register her family history together with her family members during the upcoming family reunion. Although it was a little awkward at first to ask her family members for their medical history, they agreed to do so anonymously online and repeat this every 5 years. Angela shows her family tree online to her GP who registers relevant information in his Electronic Patient Record and uses this information to build a pdf with only initials and years of birth of family members and adds this to her record. Not only is she now able to take her family history to her GP, the other family members who used the online tool are also able to do so. The whole family is enabled to operationalize their family history through a snowball effect.

Step 5: Five years later Angela's daughter Stephany, then aged 18, visits the GP with gynaecological problems. She feels a painful swelling. She started to study law in a different city and her new GP uploaded her medical and family history into his Electronic Patient Record. The Electronic Patient Record has alarmed Stephany's new GP with a pop-up that Stephany is carrier of a *BRCA2* mutation since the clinical geneticist not only diagnosed Angela with a mutation, but unfortunately also her two daughters. Angela's daughter is frequently checked with a physical and

MRI by a surgeon familiar with familial breast- and ovarian cancer who follows the national guidelines for familial cancer. Now that she has these complaints you decide to call the surgeon and after careful deliberation you refer her the same day to the clinic for further diagnostics. Fortunately, no abnormalities are found through the gynaecological and vaginal ultrasound examination.

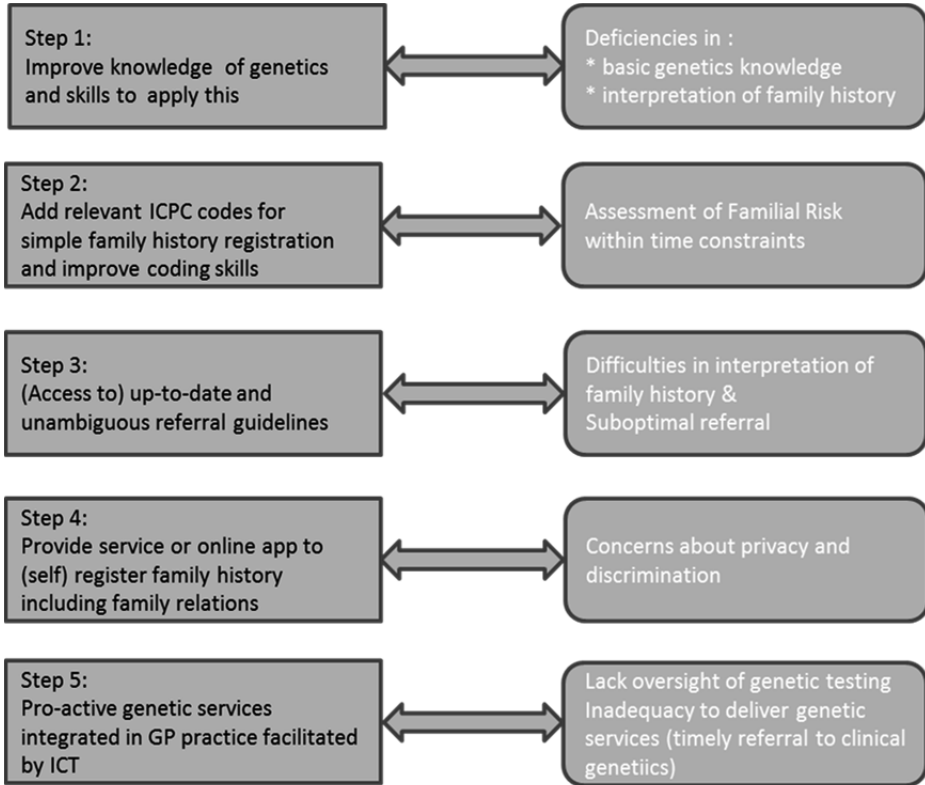


Figure 1: Proposed roadmap to stepwise integration of genetics in family medicine

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Chapter 8
General discussion



Mrs. A visited her GP because her mother died at the age of 35 of breast cancer. The GP referred her to a clinical genetic center, where testing for BRCA mutations was discussed. Mrs. A chose to undergo a test and turned out to be a carrier of a BRCA mutation. Once she had two children, she decided to go for prophylactic salpingo-oophorectomy and mastectomy. After some years she tells her GP that although it was not easy to decide on the surgical interventions and the physical and emotional consequences she has to deal with every day, she is happy cancer is precluded.

More and more patients and physicians will face questions on genetics in their own lives and professional career. Genetic core competences for non-genetic health care workers have been defined, as a prerequisite for implementing genetics education for general practice.¹⁻⁵ Continuing Professional Development (CPD) modules should be based on an educational needs assessment of general practitioners (GPs) referring to the three domains of educational activities: cognitive (*knowledge*), psychomotor (*skills*) and affective (*attitude*).⁶⁻⁸

This thesis focused on the exploration and consensus finding of genetic educational needs and priorities among primary care providers. Furthermore, we developed and evaluated three modules based on the priorities identified (an online and an interactive oncogenetic training for GPs as well as a website (*huisartsengenetica.nl*) with supporting genetics information applicable in daily practice for general practitioners). This final Chapter starts with a short outline of the background and objectives of our studies. Subsequently, the findings will be discussed in relation to the research questions. The Chapter concludes with future perspectives on genetic education in primary care, both nationally and internationally, including recommendations for changes in the medical curriculum and future research.

Background and objectives

Genomics holds great promises to improve human health, although also sometimes short term implications are overestimated.⁹ GPs are facing a daunting informational challenge to keep abreast of the expanding body of genomics knowledge and attain competencies for informed use of its potential for personalized patient care.¹⁰ Requests for DNA-based predictive testing could increase in case of a positive family history and GPs will thus be more involved in preventive check-ups. It is therefore important for GPs to be competent to take and interpret a family history and deal appropriately with patients' questions and concerns. Lack of genetic literacy in GPs is widely recognized; therefore we aimed to explore genetics educational needs and priorities, followed by determining the effectiveness of organized genetic CPD modules for GPs tailored to these needs, taking oncogenetics as an example.

PART I

Creating an agenda for effective genetic educational strategies:

Needs assessment and prioritization in primary care

In order to successfully implement genetics education in primary care, perspectives on the educational needs and role of genetics were explored (Chapter 2). The development of effective education requires a training program with a strong theoretical basis and rigorous evaluation which should tailor to the specific settings and needs of primary care professionals.^{8,11}

Educational needs assessment

The focus group study reported in Chapter 2, indicated that Dutch primary care professionals need, and would welcome, more extensive education in genetics.¹² Four major themes emerged in relation to the role of genetics in primary care and the related educational needs: (1) basic knowledge, (2) education on family history taking and the potential clinical consequences, (3) ethical dilemmas and psychosocial effects related to genetics and (4) insight into the organization of regional genetics services for possible referral. There was general agreement that increased genetics knowledge and family history taking results in a better understanding of the organization of genetics services in order to promote more appropriate and timely referrals. A similar need for genetics education in primary care was also found in other studies, in which genetics experts proposed learning outcomes and core competencies in genetics for non-genetic health care professionals.^{13,14} The results indicated a paucity of knowledge in primary care professionals would lead to poor recognition of and unresponsiveness to genetic problems in daily patient care.

The results presented in Chapter 2 differ from others with regard to the need for increased genetics knowledge among midwives.^{15,16} Midwives in the Netherlands, UK and Sweden reported a low confidence with genetic issues in clinical practice, and identified psychosocial, screening and risk assessment aspects of genetic education as being important to them, rather than technical aspects or genetic science. Midwives in our study however, seemed more confident of their basic genetic knowledge, did not perceive as strong a need to adapt existing educational modules for they could have been relatively well prepared on this specific topic. This difference could have also been due to differences between master programs in midwifery, recent post graduate training programmes for midwives on prenatal screening in the Netherlands or differences between health care systems (UK, Sweden and Netherlands).

The focus group study showed GPs recognized the important role of genetics in primary care (Chapter 2). Qureshi *et al* similarly reported that primary care

practitioners recognize relevance of genetics in daily practice, such as detecting and managing risk of multifactorial disorders and genetic reproductive risk and targeted drug therapy.¹⁷ This was in contrast to a study conducted by Fetters *et al.* in 1999.¹⁸ They found GPs were reluctant to invest in self-education in genetics, because genetic problems were believed not clinically relevant. Our study showed that today's primary care providers are aware of a progressive impact of genetics on primary care and therefore increasingly conscious of what they don't know. This difference in perspective could be due to time lapse, finishing up of the Human Genome Project or clinically relevant advances in next genome sequencing (NGS).¹⁹ The need for attention to genetics in educational modules and thus the potential to become an integral part of primary care was recognized.

Prioritization of educational needs

Chapter 3 reported the Delphi procedure used to prioritise GPs' educational needs, as identified in focus groups. The results aimed to inform the development of effective genetics education for GPs as reported in Chapters 4-6 describing *Development and Evaluation of Genetic Educational Modules for General Practitioners*. After three Delphi rounds, 29 topics were reduced to 10 priorities (Chapter 3). All eighteen expert participants completed all three rounds, with many comments per round, indicating strong involvement with the study aims. Our study generated consensus on a Top 10 of prioritised topics for GPs' genetics education.²⁰ The highest-ranking topics were concerned with *skill* and *knowledge* competences more so than *attitude* competence:

1. "Recognising signals that can indicate a hereditary component of a disease",
2. "Evaluating indications for referral to a clinical genetics centre", and
3. "Knowledge of the possibilities and limitations of genetic tests".

This was in contrast to previous research by others into the perspectives of GPs and midwives on the educational priorities and attitudes in relation to genetics, in contrast to our Delphi study which only involved GPs. In these studies, a need for genetics education was revealed in areas like psychosocial issues and screening, assessment of the risk for genetic malformations and basic genetics.^{15,16,18,21-26} More specific educational needs of primary care providers and their views on the role of genetics in daily practice and international efforts to translate these needs into education programs were in these previous studies in their early stages of investigation.²⁷⁻²⁹ Our Delphi study made it possible to operationalize the focus group results and thus enable further work towards integration of prioritized genetics educational topics in GP Continuing Professional Development (CPD) modules.

The results of the Delphi study differ from those of the Genetic-Educational Priorities (Gen-EP) scale study of 2004 by Calefato *et al.*, the previously discussed focus group study and from the American General practitioner Core Educational Guidelines, Core Competences in genetics for health professionals in Europe.^{5,30,31} Competences relating to attitudes received more attention in these studies, such as “ethical, legal and public health issues” and “psychosocial and counselling issues”. This difference may be attributed to the fact the Gen-EP study was not limited to the Netherlands but encompassed five European countries with differing health care systems. Against all odds, our Delphi study results showed genetics education should first focus on “knowledge” before moving on to “attitudes”. Some comments by experts on this issue were rather ambivalent: “Attitude is not specific to genetics” and “A good attitude should be an intrinsic component of the GP’s role”. Thus, although it may seem the “attitude” domain is considered essential for holistic GP genetics education (i.e. case based learning with medical ethical problems), effective implementation of genetics education may be jeopardised if too much attention would be paid to this area.

The results found in part 1 of the project provided the learning outcomes and core competences in genetics for non-genetic health care professionals as specified by genetic experts.³² This laid a firm foundation, supported by target group and expert opinions, for the development of genetics education modules for GPs presented in part 2 of this thesis.

PART II

Development and evaluation of genetic educational modules for general practitioners

The previous Chapters 1, 2 and 3 explained why it is urgent to develop and evaluate oncogenetics CPD modules. CPD seemed to be the obvious vehicle for remedying deficiencies in practicing physicians’ genetics knowledge and skills. Innovation of guidelines incorporating genetics advances and defining responsibilities of non-genetic healthcare professionals (e.g. when and how to refer or register) should follow if appropriate education and tools are in place.⁸ Two CPD modules were developed in collaboration with the The Dutch College of General practitioners (e.g. Nederlands Huisartsen Genootschap, NHG) fitting their conference theme “Oncology in primary care” for the year 2011- 2012: a Genetic online CPD module (G-eCPD) and a live CPD module taking oncogenetics as a model condition.^{33,34} We used the theme “oncogenetics” as a “coincidental” example and fit the adaptable multifaceted educational framework to hang on all prioritized topics found through the initial explorative Delphi studies building on genetic education core

competences and thus made case based genetics education available. An easily accessible website on genetics in general practice for daily use in practice was also developed in collaboration with the NHG and received a spot on their main website potentially recognizable for all Dutch GPs. The website means to support the G-eCPD and live CPD modules, which gives GPs on demand information such as information on how to do a family history, how to recognize hereditary diseases, basic knowledge of genetics information and how and where to refer.³⁵ The website is not limited to oncogenetics.

We aimed to measure the educational outcomes of the CPD modules overall, at all four levels of Kirkpatrick's framework for evaluating educational outcomes (satisfaction, knowledge and knowledge retention, behavior, and actual practice performance and results).^{36,37,38} The instruments and data collected are all described in table 1, as is the time frame.¹

Two randomized controlled trials (RCTs) for the first two modules (eCPD and live training) were conducted reported in the second part of this thesis in order to find out whether effective, adaptable and sustainable genetics education for GPs can be organized and developed.

Effectiveness of an online G-eCPD module

The online G-eCPD module aimed at improving knowledge about oncogenetics, resulted in positive outcomes at the first two levels of Kirkpatrick's framework (satisfaction and learning). Although evaluation of the educational outcomes of the G-eCPD module by questionnaire on the application of new knowledge in daily practice closely approached Kirkpatrick's third level, assessment by observation of actual change in clinical genetics behavior and improved patient genetic health outcome was currently absent. Studies have reported effectiveness of eCPD modules on other topics and suggested that course outcomes may benefit when a course is designed in accordance with a prior educational needs assessment.³⁹⁻⁴³ The advantages of online CPD have been broadly discussed and supported.⁴⁴

Table 1. Description of Instruments to assess genetic CPD outcomes

Instrument		G-eCPD ^{3,5}	Live Training ^{4,5}	Website ⁵	Description of instrument measurements
Demographics questionnaire	Time ¹	T0	T0	Not available (n.a.)	Gender, age, years of experience, type of practice, degree of urbanization of practice area
	KP level ²	n.a.	n.a.	n.a.	
Satisfaction questionnaire	Time	T1	T1	T3	Recommendation of activity, relevance to general practice, most appealing topics, grade, time spent
	KP level	I	I	I	
Knowledge test	Time	T0, T1 and T2 n.a.		n.a.	Multiple-choice Case based Knowledge test
	KP level	II	n.a.	n.a.	
Applicability questionnaire	Time	T3	T3	T3	Self-reported applicability of newly learned knowledge (genetic tests, basic genetic concepts, information sources) and skills (recognize and refer sooner) and referral behaviour as a result of CPD modules and website
	KP level	II	II	II	
Standardized Patients' checklist	Time	T0, T1 and T2 T0, T1 and T2 n.a.		n.a.	Standardized 28-item checklist assessing consultation behavior covering the full scope of the consultation
	KP level	n.a.	III	n.a.	
Referral data	Time	T3	T3	T3	Change in number of referrals by GPs
	KP level	IV	IV	IV	

¹)Time: The scheduled measurement times of CPD activities are as follows: T0 Pretest, T1 Posttest, T2 Retention test (G-eCPD 6 months post intervention, live training 3 months post intervention) and T3 one year post-intervention February 2013); ²) KP level: Kirkpatrick level; Effect evaluation of CPD modules is at four levels (I-IV) of Kirkpatrick; ³) Results reported in Chapter 4 of this thesis; ⁴) Results reported in Chapter 5 of this thesis; ⁵) Results reported in Chapter 6 of this thesis.^{33,34}

Effectiveness of a live oncogenetic CPD module

Standardized patients were used to measure effectiveness of live oncogenetics CPD module in terms of behavioral and organizational changes (Kirkpatrick level

3). The needs-based, interactive oncogenetics CPD module aimed at improving genetics practice behavior (Chapter 5, third level of Kirkpatrick). The oncogenetics CPD module resulted in improved genetic consultation skills. This effect was studied by using unannounced standardized patients and participants' satisfaction with the training and the reported applicability of what they have learned was studied by using online questionnaires. The four-hour training covered oncogenetic skills (family history, oncogenetic risk assessment, and efficient referral), attitude (medical ethical issues relevant to the identification of oncogenetic disease in family practice consultations), and clinical knowledge. One of the aims of including real patients and simulated consultations in our training was to promote a favorable attitude among GPs to the application of genetic competencies. A study by Carroll et al. measured practice intent to use clinical genetics scenarios and increase competence due to a multifaceted knowledge translation intervention, but used questionnaires and not actual ratings of observed practice behaviors.⁴⁵ Patient and societal perspectives on legal consequences of DNA-based testing results (for example being able to find a genetics information source or ability to obtain a mortgage or life insurance) however, demand that physicians' effective use of genetics CPD modules be demonstrated by actual performance in health care practice⁴⁶. We therefore deliberately deployed trained and blinded SPs to optimize the value of the measurement.

Future studies may investigate reproducibility of clinical performance in practice using incognito standardized patients (SP). Potential oncogenetic problems are considered very personal topics to discuss between a patient and their own GP.¹² This determined why it was not discussed in an incognito setting with a so-called "new" patient or unannounced, concealed SP.⁴⁷ Detecting the SPs by the participating GPs would become too easy in an incognito setting in the case of oncogenetics problems and would have been detrimental to the study.

In this study, increase in competences of implementing new consultation skills was demonstrated not performance in daily practice. Competency based assessments measure what doctors *can do* in controlled representations of professional practice; performance based assessments measure what doctors *actually do* in professional practice. Competency is therefore regarded a prerequisite for performance.⁴⁶ As the assessments took place in GPs' practices and consultation skills in potential oncogenetic situations are frequently encountered, performance may closely approach competences in this study. However, real life patients present in a wide range of ways which makes sustainably implementing acquired skills in different daily practice case scenarios a challenge and requires further studies.

Length of the project limited further research on continuous impact on applicability of the GP oncogenetics CPD modules in daily practice. This requires a parallel and ongoing process of exploration of educational needs and priorities. Various reviews have pointed to heightened effects on physician behavior of mul-

tiple interventions compared to single episodic interventions. Multifaceted interventions can tackle several common barriers to change and this combined operation may ultimately lead to improved practice performance. These aspects deserve further study.

A practice-based genetics website for GPs: www.huisartsengenetica.nl

The website huisartsengenetica.nl (*GP and genetics*) was developed in collaboration with Erfocentrum (Dutch information center on heredity and genetic disorders) and NHG, with on demand supportive information to be able to work on the learning tasks and apply competences in daily practice. From the beginning of the website, the website was used with visitor numbers (around 350 per week) expected from a website in a startup phase (personal communication with Erfocentrum erfelijkheid.nl). Users of the website reported to be satisfied and reported it tailored to their needs with supporting information applicable in daily practice. Although, limited number of self-reports showed the website was only visited once in a while, the website was also studied if referral was considered and would be commended to colleagues.

Opportunities for future research should focus on how to increase the website visitor numbers. Personal interviews or focus groups research could explore usability and user-friendliness of the website. It is also possible future genetics CPD modules should emphasize better the supportiveness of the website for the modules attended and potential in daily practice.

Self-reported change in practice performance and patient referral to clinical genetics centers as a result of the oncogenetic CPD modules and website

Chapter 6 reports sustained effects on professional practice after attending the CPD modules and the supportive website. GPs seemed to synthesize and apply the newly-learned consultation behaviors. Visitors to the website were highly satisfied with and reported the website to be applicable in daily practice. These results reflected an increased interest in content of the website and referral possibilities. The number of referrals to the clinical genetics centers however did not significantly change one year after launch of the modules and website. The reason could be because change in referral number is expected after about 10 years of medical education innovation and guideline changes (personal communication with Irina Stirbu, LINH, 2013). In theory, the same number or rather a decrease in referrals could also mean a reduction of unnecessary referrals while the number of appropriate referrals increases. The results did not show referral effectiveness and efficiency on a meta-level, for the live training reached only a small part of the total of Dutch GPs who attended the regional training. Knowledge acquired by those GPs

(in training) who attended the freely offered G-eCPD may not have been sufficient enough to have impact on referral. Still, the study results indicate an adaptable and effective framework for genetics education for health professionals.

Self-reported questionnaires showed us there was a subjective increase in applying genetics competences in daily practice as a result of the oncogenetic CPD modules. GPs reported they used the website to help them in the referral process, were very satisfied with the information applied on the website, especially the information on diseases, family history and referral. GPs considered referring patients possibly affected with oncogenetic diseases more frequently than beforehand. Self-reported questionnaires are subjective and further objective quantification of performance results in actual practice is required to substantiate evidence on referral efficacy and effectiveness. Once ICPC codes are in place in GP electronic patient records, it would be possible to compare referral numbers in a RCT by those who attended the CPD modules and those who did not and retrospectively check for efficiency of the referrals to clinical genetics centers involved.

Opportunities for improvement of genetic competences could mean applying advances in pharmacogenomics in daily medical care and therefore improvement of patient safety. As a recent example, aspirin chemoprevention showed to reduce cancer risk in Lynch syndrome carriers by $\pm 50\%$ and was recommended as standard of care.⁴⁸ Lynch syndrome is the most frequent monogenic subtype of colorectal cancer, on average occurring in 4 patients in every GP practice (of 2000 patients). What if this information were included in GP guidelines? Are GPs ready to recognize healthy family members at risk of Lynch syndrome, refer them for genetic testing, and inform them of this new possibility for prevention? Expanding quality of life by early detection in screening modules (e.g. heel prick screening in new-borns and timely referral to specialised paediatricians) and personalized treatment (e.g. timely referral in case of familial forms of breast cancer) will not materialize unless true translation into the primary care workplace and other non-genetic health care settings takes place.

PART III

Summarizing discussion and future perspectives

In order to create an agenda for effective genetic educational strategies in primary care, educational needs were assessed and prioritized. The focus groups identified needs in the following categories: genetics knowledge, family history, organization of clinical genetics services and ethical dilemmas and psychosocial effects.¹² These themes reflected a shift in the role of genetics in primary care with implications for

education. A Delphi study generated consensus on a top 10 of prioritised topics for GPs' genetics education. The highest-ranking topics were concerned with skill and knowledge competences: "Recognising signals that can indicate a hereditary component of a disease", "Evaluating indications for referral to a clinical genetics centre", and "Knowledge of the possibilities and limitations of genetic tests". The priorities resulting from the Delphi study informed the development of oncogenetic educational modules, including input for case-based education, to improve GP performance in genetic patient care. The GP-specific training proved to be a feasible, satisfactory and clinically applicable method to improve oncogenetics competences and could be used as an educational framework to inform future training activities with the ultimate aim of improving medical care.

Raising primary care providers' confidence through better organizational help and education would potentially enable them in busy practices to apply these approaches and makes fully implementing personalized genetic risk assessment possible. Future trials to identify barriers are necessary to effectively implement decision support systems in the future in real practice.

Potentially integrating genetics in daily primary care practice, through effective CPD modules and operationalization of previously found genetic educational priorities should be possible. Murray *et al* recently reported family health history data entered by patients in primary care Electronic Patient Record (EPR) is more efficient than standard of care data entered by GPs themselves.⁴⁹ Determining different data portal entries matching patient preference, influence of use on daily practice workflow and possible effects on screening and prevention need further study. It remains to be explored whether adapting the framework to a topic such as cardiogenetics will be as effective and consequently determine changes in referrals and registrations in the EPR. Tools to use to assess these changes in daily general practice such as (integration of genetics in existing) guidelines and ICPC codes for EPRs however need further development. Agreements on when and how to refer on a national basis among clinical geneticists and GPs needs further discussion.⁵⁰

Methodological reflections

Some methodological issues of the studies presented in this thesis should be noted. To assess the educational needs to adapt genetic CPD modules, a qualitative research design was used. This explorative study design made it possible to get a more complete picture than data obtained by, for example a questionnaire survey. Purposive sampling was used to recruit specific groups of professionals for focus group interviews in order to obtain rich, relevant and diverse data. The participants were expected to provide complete and possibly complementary perspec-

tives on genetics in primary care practice and education. Surveys alone cannot disentangle different contributing factors, for it is hard to know whether a survey is accurate or whether the terms used are unambiguous. Focus group research makes it possible to explore contributing factors and discuss possible defined terms. Although focus group studies may have introduced selection bias through purposive sampling, further investigation should show whether the results are generalizable to real practice and beyond the Dutch health care system.

The Delphi prioritization method should be an ongoing process, repeated every so many years. For learning needs and priorities may change as clinically relevant advances come forward such as preconceptional and prenatal carrier screening for inherited genetic disorders for example hemoglobinopathies or cystic fibrosis through NGS.⁵¹⁻⁵³ We believe the results of this study should be used in the near future to guide the implementation of genetics education in the Netherlands and perhaps even internationally. Whether the prioritized genetic educational topics cover genetics-related knowledge, skills and attitudes essential for every medical care provider, will have to be determined. Another limitation could be that the nature of the sample of selected experts for the Delphi study was drawn from the Dutch health care system. Relevance beyond this system remains to be investigated. Regression to the mean by adjustment of experts' opinions as a result of this consensus method could have also been a limitation. However, a high degree of agreement on the ten accepted topics contradicted this possibility. Awareness of previously found focus group results could have caused opinion bias limiting the Delphi study results. Validity of the results was checked however through triangulation, namely by comparing the focus group with the Delphi group procedure results.

To our knowledge, this was the first time a series of oncogenetic CPD modules was organized based on prioritized genetics topics evaluating educational effects.⁵⁴ Collaboration with the NHG meant recognition and therefore better reach of the educational modules by Dutch GPs. Experts in implementation and GP education expected if the genetics education module model would be presented as a fruit basket (multifaceted format with an online and live training and supporting website) usually presented by the NHG, meant better reach to the GP learners much more so than when the project team would have tried to have implemented the modules on their own. This project therefore aimed for support by a broad range of stakeholders, which was previously regarded to be one of important factors for successful implementations of innovations in patient care.⁸ Whether the results also apply to other possible target groups in primary and secondary care, those in training, medical school students or high school biology students needs further research. Although there were no significant differences in participant characteristics between intervention and control group, the physicians in the studies presented in chapters 4, 5 and 6 appeared to be more female, younger and less

experienced, compared to the general profile of Dutch GPs.⁵⁵ This possibly reflects extra interest in genetics and/or using online learning modules by young female GPs who recently graduated. Previously a postal survey among GPs in Northern Ireland, suggested that GPs are interested in training courses on genetics and genetic testing. The results indicated male GPs and GPs who have been qualified for longer should be specifically targeted.⁵⁶ Whether the results can be generalized within and beyond the Dutch healthcare system, needs further investigation.

A limitation of both the online and live CPD module evaluations is the fairly large number of non-responders in the RCTs. The long duration of the studies (i.e. 3- 6 months) could have been an explanation; the reasons given by physicians however were lack of time or illness. Nevertheless, our retention rate (55%) was comparable to those reported for postal surveys among GPs (60%) and much higher than the rate found in other RCTs involving GPs.^{57,58,59} The financial incentives for participants may have introduced selection bias. Larger-scale national and international RCTs with adequate power are nevertheless warranted to further assess how genetics education can improve health care, validity and increase generalizability. It is well known GPs are busy people with full agendas possibly resulting in low response rates in RCTs. RCTs among GPs should therefore try to study topics of interest in daily practice for GPs focusing on urgency of the problem. Reasons for low-response rate could reflect possible barriers of implementing changes, which should therefore be critically studied and possibly be acted on in future RCTs studying effectiveness of genetics CPD modules.

Conclusions

To organize effective genetics education for GPs, tailoring education to the needs and priorities of prospective users seems to be an important factor in order to effectively implement genetics in daily practice and potentially improve behavioural competences and performance. Evaluation of genetic training modules can be achieved at several levels of Kirkpatrick, including satisfaction, knowledge, attitude and behaviour (referral; changing the practice of registering family history).

A systematic policy of implementation revealed effective promotion and dissemination regionally and nationally (NHG) results in sustainable genetics education. Reform of existing guidelines and (ICT) tools for pre-symptomatic referral and genetic testing is necessary to further integrate pro-active genetic services in GP practice.

Recommendations for changes in genetic education

The integration and visibility of genetics in daily medical care and education is still very limited.^{27,60,61,62} Barriers to operationalization of effective genetics education in clinical research and daily primary care can essentially be overcome with constructive and indispensable collaboration. For truly turning useful genetics discoveries from the laboratory bench to daily clinical practice, a roadmap is crucial to make urgent translation feasible.⁵⁰ A few recommendations for further development of genetic education can now be suggested.

1. Involve primary care physicians in identifying their learning goals and priorities, and build effective training modules based on these.
2. Multifaceted genetics CPD modules should support one another. An online genetics CPD module can improve knowledge competences, and thus prepare for the live CPD module focussing on genetic consultation skills supported by a genetics website with on demand information.
3. Innovative and practical ICT tools should be developed to support advances in genomic literacy of health care providers. Their use in daily practice should be trained accordingly.
4. End-objectives for genetic education in the medical curriculum are not clearly defined and agreement upon this would be recommended, although it should be recognized that the field is dynamic. Integration of case based genetics problems in study methods and therefore increasing clinically relevant genetic knowledge and consultation skills, has now been shown to be efficient and should thus be an integral part of the medical curriculum.
5. The results could contribute to the development of genetics CPD modules in general and international. The online and live program and supportive website could potentially reach a large group of physicians in primary and secondary care provided that it is adapted to the specific target groups. Possibly also medical school students and high school biology classes could benefit from this framework for genetics education.
6. It would be challenging logistically to introduce SPs as a routine educational activity in daily outpatient practice. In some of the Dutch medical curricula however, SPs are already part of routine education to train physical examination and consultation skills. Also, the SPs are professional actors who were very enthusiastic about the oncogenetics training and expressed their motivation to train GPs on a frequent basis. The GPs themselves were also enthusiastic about SPs coming to their practice since it gave them an unexpected training situation in their own practice. With logistic organization and finances well in place, SPs could therefore with minor adjustments be introduced on a regular basis.

Recommendations for further research

Further research should focus on the following topics:

1. There is an urgent need for a clear description of responsibilities in daily practice and guidelines to enable effective use of developments in genetics in primary care. Especially descriptions of the genetic responsibilities of primary care providers and their specific role in this area will have to be addressed in the future, as was recommended by Baars *et al.*⁶³ Consequently, further investigation is necessary whether the framework designed to deliver genetics education for active GPs, is just as effective when it is delivered to GPs in training, midwives (in training), non clinical genetic medical specialists (such as paediatricians, gynaecologists and other medical specialists potentially confronted with genetic problems in daily practice).
2. Short term, decision support tools to enable integration of genetics in daily practice should be developed and implemented for example on oncogenetics, such as ICPC codes in GP electronic patient records.⁵⁰ This would enable timely awareness of possible familial diseases and referral. How to use these tools in daily practice should be part of future genetics education.
3. The number of adequate referrals to clinical genetics departments or timely identification of patients with a cancer predisposition syndrome (4th level of Kirkpatrick's framework for evaluation of educational outcomes) should be part of further studies. Previous studies have shown that as far as genetics is concerned non-genetic healthcare workers require not only education but also clear guidelines and definitions of their responsibilities.^{12,20,28} Improving possibilities to register the family history in such a way that queries can identify patients at risk as has been proposed, could promote impact.
4. Also, further research could be done on effective referral when ICPC codes for correct family history (self) registration are in place improving genetic consultation by GPs. Insight into the organization of regional genetics services and the referral system should sustainably be enhanced through education and online supportive information (huisartsengenetica.nl), to promote interdisciplinary collaboration.
5. Long term, further research should focus on effective genetics CPD module implementation to increase competences and performance among the rest of Dutch GPs and those outside the Netherlands. Possible effectiveness on genetic health care could then be investigated. International health systems may be organized very differently. Patients may go to the medical specialist without interference of a filter played by the GP for example. We assume different health systems may require adaptable content, but a uniform educational framework.

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Chapter 9

Summary



Training in genetics and genomics for primary health care workers

Medical professionals in primary care are increasingly expected to deliver genetic services in daily patient care and need to be prepared for patients asking for information and advice on genetics. This requires appropriate skills and knowledge of genetics needed for daily practice. However, postgraduate (physician training) and master (midwifery training) programmes in primary care and public health are currently failing to meet these perceived educational needs. Improvements in genetics education for primary care providers are thus needed to keep up with the rapid developments in genetics/genomics.

The main objectives of this study project were to reflect on current genetics/genomics developments with primary care workers, to help them identify their learning priorities and to evaluate three CPD modules in oncogenetics developed in collaboration with multidisciplinary team of general practitioners (GPs), educationalists and clinical geneticists familiar with genetics in primary care. Key factors for successful future training were identified and could make integrating genetics step by step in daily genetic primary care possible.

In the first part of this thesis an agenda for effective genetic educational strategies was created for which a needs assessment and prioritization of genetic education in primary care were studied.

Chapter 2 presents the results of a focus group study, which explored the role of genetics in primary care (i.e. family medicine and midwifery care) and the need for education in this area as perceived by primary care health workers, patient advocacy groups and clinical genetics professionals. Forty-four participants took part in three types of focus groups: mono-disciplinary groups of general practitioners and midwives, respectively and multidisciplinary groups composed of a diverse set of experts. Four themes emerged regarding the educational needs and the role of genetics in primary care: (1) the need for genetics knowledge, (2) taking a family

history, (3) ethical dilemmas and psychosocial effects in relation to genetics, and (4) insight into the organisation and role of clinical genetics services. The role of genetics in primary care was perceived to be unduly limited as a result of care providers' inadequate genetics knowledge and skills. Although all focus group participants acknowledged the importance of genetics education, general practitioners seemed to feel this need more urgently than midwives and more strongly emphasized their perceived knowledge deficiencies. The study results indicated that Dutch primary care providers need, and would welcome, more extensive education in genetics, for instance in postgraduate and master programmes.

Chapter 3 presents the results of a Delphi consensus procedure, which aimed to operationalize the focus group results and prioritise topics for genetics education for general practice. Topics mentioned as learning goals were rephrased in line with core competences. The study conducted, consisted of three rounds. A purposively selected heterogeneous panel (n=18) of experts, comprising six practising GPs also engaged in research, five GP trainers, four clinical genetics professionals and three representatives of patient organisations, participated. Educational needs regarding genetics in general practice in terms of knowledge, skills and attitudes, were rated and ranked in a top 10. The entire panel completed all three rounds. Kendall's coefficient of concordance indicated significant agreement regarding the top ten genetic educational needs ($P < 0.001$). "Recognising signals that are potentially indicative of a hereditary component of a disease" was rated highest, followed by "Evaluating indications for referral to a clinical genetics centre" and "Knowledge of the possibilities and limitations of genetic tests". It was concluded that the education priorities resulting from the study could be used to guide the development of genetics genetic education to improve GP performance in daily practice.

The results described in Chapters 2 and 3 informed the development of genetic Continuing Professional Development (CPD) modules, including input for case-based education, to improve GP performance in genetic patient care. More research into the educational priorities in genetics is needed to design courses that are suitable for master programmes for midwives.

The second part of this thesis describes the development and evaluation of three training modules: a Genetic online Continuing Professional Development (G-eCPD) and live genetic CPD module, taking oncogenetics as an example, supported by a website on genetics for GPs (*huisartsengenetica.nl* or "*GP and genetics*"). Identification of patients at risk for hereditary cancers is considered essential to inform decisions about early screening, genetic testing, and pre-symptomatic risk reducing options. To our knowledge these were the first randomized controlled trials

(RCTs) to investigate improvement of GPs' oncogenetic knowledge and professional behavior after participation in educational modules.

In Chapter 4 an RCT was conducted to evaluate the educational outcomes of a G-eCPD module at the first two levels of the Kirkpatrick framework (satisfaction and learning). The aim of this G-eCPD module was to provide physicians sufficient knowledge and skills through didactic presentations, interactive cases and enabling tools such as information about regional possibilities for referral and consultation. It meant to enable GPs to identify patients with an inherited predisposition to cancer; draw a family tree as a tool for identifying patients at risk for hereditary cancer; describe the most common types of hereditary cancer (i.e. breast, colon) and the likely genetic mutations involved; apply oncogenetics guidelines in identifying patients for whom referral is indicated or not and find relevant information online; explain the possibilities and limitations of oncogenetic testing; discuss with patients periodic examinations and risk-reducing surgical options that are available to patients with hereditary cancer.

The G-eCPD module aimed at improving GPs' knowledge about oncogenetics, and was conducted between September 2011 and March 2012. The study method was a parallel-group pre-post-retention (six-month follow-up) controlled group intervention trial, with repeated measurements. Of the total of 80 Dutch GP volunteers (40 intervention group and 40 control group randomly assigned), 44 participants (20 intervention, 24 control group) completed all the learning activities, knowledge tests, and questionnaires. For validity reasons, recruitment was limited to all GPs practicing outside the two Dutch provinces in the North (Noord Holland) and South (Limburg) where GPs could also participate in the live CPD module. The findings of the RCT showed that satisfaction with the module was high, with the three item's scores in the range 4.1-4.3 (five-point scale) and a global score of 7.9 (ten-point scale). Knowledge gains at post-test and retention test were 0.055 ($P < 0.05$) and 0.079 ($P < 0.01$), respectively, with moderate effect sizes (0.27 and 0.31). The participants appreciated the applicability in daily practice of knowledge aspects (item scores 3.3-3.8, five-point scale), but scores on self-reported identification of disease, referral to a specialist, and knowledge about the possibilities/limitations of genetic testing were near neutral (2.7-2.8, five-point scale). It was concluded that the educational effects reported in this study can inform further development of online G-eCPD aimed at improving physicians' genetics knowledge and could potentially improve patient care. The online CPD module with its framework could reach a large group of physicians with a wide variety of backgrounds, but adapted to its audience, possibly also medical school students and be used in high school biology classes.

Chapter 5 describes whether a live oncogenetics CPD module improves GP consultation skills. In this pragmatic, blinded RCT, the intervention consisted of a four-

hour training (December 2011 and April 2012), covering oncogenetic consultation skills (family history, familial risk assessment, and efficient referral), attitude (medical ethical issues), and clinical knowledge required in primary care consultations. Outcomes were measured using observation checklists by unannounced standardized patients and self-reported questionnaires. For logistic reasons, recruitment was limited to all GPs practicing two Dutch provinces in the North (Noord Holland) and South (Limburg) who did not participate in the evaluation of the G-eCPD. Of 88 randomized GPs who initially agreed to participate, 56 completed all measurements. Key consultation skills trained equipped the GP to recall clinically relevant information about types of hereditary cancer (breast, ovarian, colon, skin) including genes associated with oncogenetics syndromes most commonly tested for; recognize patients with features suggesting inherited predisposition to cancer; draw a family tree as a tool to identify patients at risk; discuss (possible) familial and hereditary cancer risks, management of potentially developing hereditary cancer (i.e. surveillance and risk-reducing surgical options) and related ethical issues; identifying patients for referral for risk assessment and find relevant information online using oncogenetics guidelines; explain the possibilities and limitations of oncogenetic testing; and know when to consult and/or refer to a genetics specialist. These key consultation skills significantly and substantially improved; regression coefficient post-intervention equal to .34 and .28 at 3-month-follow-up indicating moderate effect size. The results show sustained improvement three months after the training as well as high satisfaction with the training and positive perceptions of the practical applicability of training topics.

Chapter 6 aimed to determine long-term (self-reported) genetic consultation skills among GPs who participated in the G-eCPD and live CPD modules (i.e. increased genetics awareness and referrals to clinical genetics centers), and interests in and satisfaction with the website. The genetics CPD modules achieved sustained improvement of oncogenetic competencies. Participants reported to be more alert of genetic problems. 88% of those who attended the live training reported to more frequently refer patients to the Clinical Genetics centers, compared to 29% of those who attended the online oncogenetics training. No significant change in referral numbers however was reported by the Clinical Genetics centers before and one year after the training. Moreover, sustained interest in and satisfaction with the newly developed GP website were investigated among website visitors who completed the pop-up questionnaire. Only a small number (38 visitors, 22 of these were GPs) completed the questionnaire during the 1- month study period. Satisfaction with the website and perceived applicability of the website on appropriateness of referrals however were highly scored. Website visitor numbers are increasing; with the page most often consulted "family tree drawing".

Thus, self-perceived genetic consultation skills increase long-term and there is interest in and satisfaction with the supportive website.

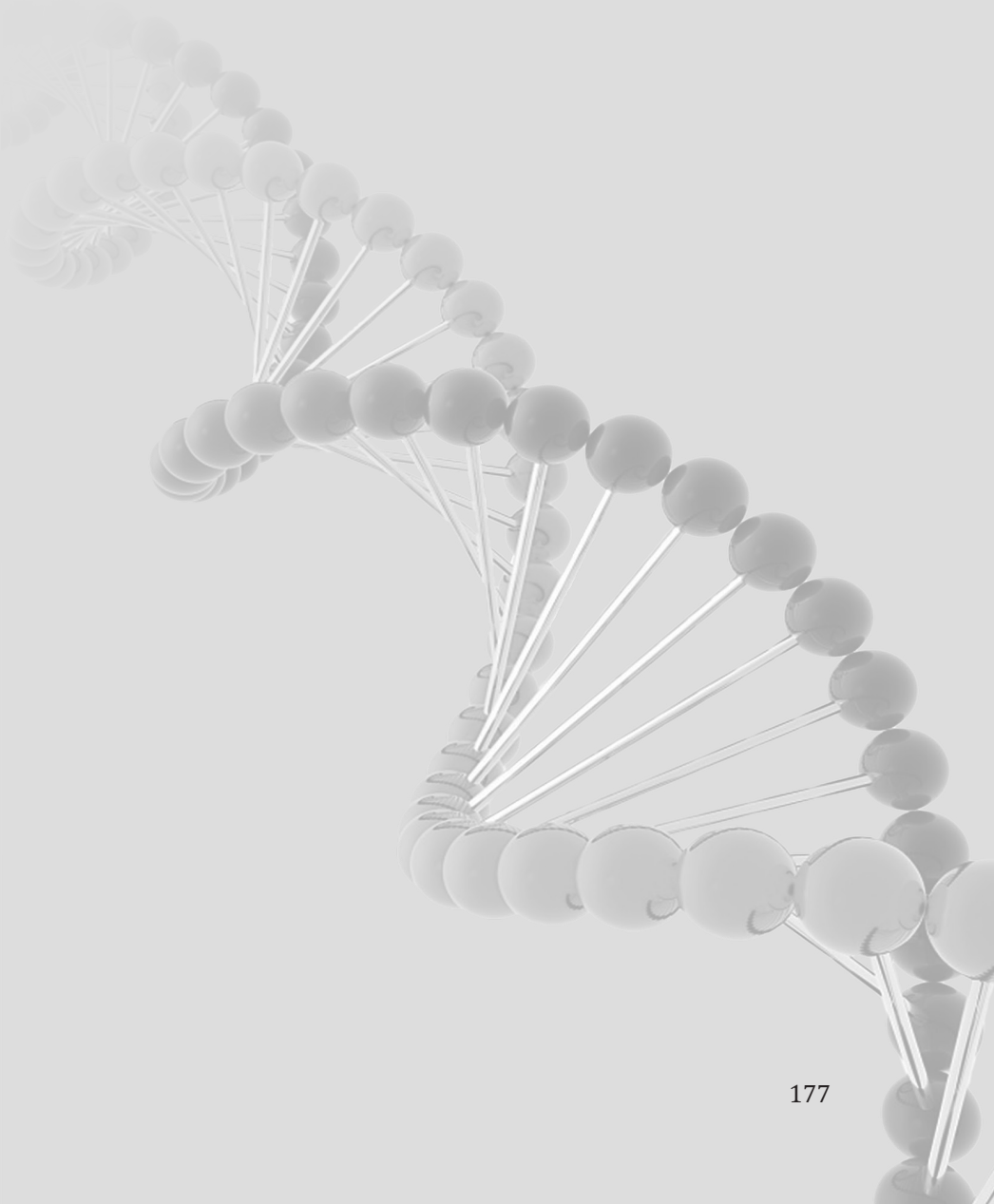
In Chapter 7 a step-by-step roadmap was proposed to integrate genetics in the Electronic Patient Record (EPR) in Family Medicine and clinical research. This could make urgent operationalization of readily available genetic knowledge feasible in clinical research and consequently improved medical care. Improving genomic literacy by training and education is needed first. The second step is the improvement of the possibilities to register the family history in such a way that queries can identify patients at risk. Adding codes to the EPR in the International Classification of Primary Care (ICPC) in the ICPC chapters “A21 Personal/family history of malignancy” and “A99 Disease carrier not described further” and other coding strategies for simple registry of family history and develop and support coding skills is proposed. EPRs need possibilities to add (new) family history information, including links between individuals who are family members. Multidisciplinary guidelines for referral must be unambiguous. Automatic alerts should help GPs to recognize patients at risk who satisfy referral criteria. A familial breast cancer case with a *BRCA1* mutation as an example was used for illustration.

Concluding remarks

The results of the studies presented in this thesis have provided better insight into how non-genetic specialists (e.g. GPs and midwives) perceive the increased role for genetics in primary care and consequently recognized the importance of genetics education. The responsibilities of primary care providers with regard to genetics require further study. The GP-specific oncogenetic CPD modules supported by the Dutch genetics website *huisartsengenetica.nl*, suggest to be a feasible, satisfactory and clinically applicable method to improve oncogenetics competences in daily practice and suggests to be an adaptable and effective educational framework to inform future training activities with the ultimate aim of improving genetic medical care.

Chapter 9

Samenvatting



Onderwijs over genetica en genomics voor eerstelijns zorgverleners

Van zorgverleners in de eerste lijn wordt in toenemende mate verwacht dat zij in de dagelijkse praktijk genetische zorg verlenen. Dit vereist een goede voorbereiding, vaardigheden en kennis die hen in staat stelt patiënten te helpen die vragen om informatie en advies op dit terrein. Er zijn aanwijzingen dat de specialistenoplossingen en de hogere beroepsopleidingen in de gezondheidszorg (verloskundeopleiding) en de sociale geneeskunde nog niet in staat zijn om aan deze wensen te voldoen. Verbeteringen in het geneticaonderwijs voor beroepen in de eerstelijnsgezondheidszorg zijn dan ook wenselijk, teneinde te kunnen garanderen dat zorgverleners op de hoogte zijn en blijven van de snelle ontwikkelingen op het gebied van de genetica.

Het eerste deel van dit proefschrift is gericht op het vinden van aanknopingspunten van effectief geneticaonderwijs. Hiertoe is onderzoek gedaan naar de onderwijsbehoeften van eerstelijnszorgverleners en is een prioritering opgesteld voor het verder ontwikkelen van geneticaonderwijs.

Hoofdstuk 2 beschrijft de resultaten van een focusgroeponderzoek over de rol van genetica in de eerste lijn (huisartsgeneeskunde en verloskundigenzorg) en de bestaande behoefte aan onderwijs op dit gebied. In totaal namen 44 deelnemers deel aan drie typen focusgroepen: twee monodisciplinaire groepen bestaande uit respectievelijk huisartsen en verloskundigen en een multidisciplinaire groep bestaande uit eerstelijnsopleiders, klinisch genetici en patiëntvertegenwoordigers. Uit de resultaten kwamen de volgende behoeften als thema's naar voren: (1) meer basiskennis genetica, (2) vaardigheden in het afnemen van een familieanamnese, (3) aandacht voor ethische dilemma's en psychosociale aspecten op het gebied van de genetica, en (4) inzicht in de organisatie en de rol van klinisch genetische diensten. De deelnemers waren van mening dat genetica ongelukkigerwijs een onnodig beperkte rol heeft in de eerstelijnsgezondheidszorg, hetgeen samenhangt met de

ontoereikende kennis en vaardigheden van zorgverleners. Hoewel alle deelnemers aan de focusgroepen het belang van geneticaonderwijs onderschreven, leken de huisartsen zich sterker bewust te zijn van de urgentie van onderwijs dan de verloskundigen. De huisartsen legden ook meer nadruk op de ervaren kennistekorten. De onderzoeksresultaten gaven aan dat zorgverleners in de eerste lijn in Nederland behoefte hebben aan en openstaan voor meer geneticaonderwijs, bijvoorbeeld in de specialisten- en masteropleidingen.

Hoofdstuk 3 beschrijft de resultaten van een Delphi-onderzoek met als doel consensus te bereiken over de prioritering van onderwerpen voor geneticaonderwijs voor huisartsen. Onderwerpen die voorgesteld werden als mogelijke leerdoelen werden geformuleerd volgens de kerncompetenties van het medisch onderwijs. Het onderzoek bestond uit drie rondes waaraan een heterogeen panel van deskundigen (n=18) deelnam, bestaande uit zes praktiserende huisartsen met onderzoekservaring, vijf huisartsenopleiders, vier klinisch genetici en drie vertegenwoordigers van patiëntenorganisaties. Het panel beoordeelde de genetica onderwijsbehoeften van huisartsen met betrekking tot kennis, vaardigheden en attitudes en stelde een top tien van onderwerpen samen. Alle panelleden namen deel aan alle rondes. De Kendall coëfficiënt van concordantie werd berekend om mate van overeenstemming over de top tien te bepalen ($P < 0.001$). “Het herkennen van signalen die mogelijk wijzen op een erfelijke component van ziekte” stond op nummer 1 en werd gevolgd door “het evalueren van verwijsindicaties naar een klinisch genetisch centrum” en “kennis van de mogelijkheden en beperkingen van genetische testen”. Geconcludeerd werd dat de onderwijsprioriteiten die uit dit onderzoek naar voren kwamen als richtlijn konden dienen voor het ontwikkelen van geneticaonderwijs gericht op verbetering van het functioneren van de huisarts in de dagelijkse praktijk.

De resultaten van hoofdstuk 2 en 3 vormden de basis voor het ontwikkelen van nascholingsmodules over genetica voor huisartsen en leverden informatie op voor te ontwikkelen onderwijscasuïstiek. [Verder onderzoek betreffende prioriteiten voor geneticaonderwijs is nodig voor het ontwikkelen van onderwijs voor de masteropleidingen voor verloskundigen.]

Het tweede deel van dit proefschrift beschrijft de ontwikkeling en evaluatie van drie onderwijsmodules: 1) een online nascholingsmodule (hoofdstuk 4), 2) een “live” nascholing (hoofdstuk 5), en 3) een website over genetica voor huisartsen (www.huisartsengenetica.nl). Voor beide nascholingsmodules diende de onco-genetica als voorbeeld. In de huisartspraktijk is het opsporen van patiënten met een erfelijke aanleg voor kanker van het grootste belang voor het voorkomen van ziekte en sterfte (bijvoorbeeld beslissingen over vroege screening, genetische testen en risicobeperkende pre-symptomatische maatregelen). In een geran-

domiseerd gecontroleerd onderzoek (RCT) zijn beide nascholingen geëvalueerd op effectiviteit in het verbeteren van de oncogenetische kennis en professioneel gedrag van huisartsen. Deze onderzoeken zijn voor zover wij weten de eerste gerandomiseerde gecontroleerde onderzoeken (RCT's) naar verbetering van de oncogenetische kennis en het professioneel gedrag van huisartsen na deelname aan onderwijsmodules.

In hoofdstuk 4 wordt een RCT beschreven betreffende de onderwijskundige uitkomsten van een online nascholingsmodule voor huisartsen. De uitkomsten zijn onderzocht op de eerste twee niveaus van het onderwijsevaluatiemodel van Kirkpatrick (tevredenheid en leerzaamheid). De module had tot doel artsen te voorzien van voldoende kennis en vaardigheden door middel van didactische presentaties, interactieve casuïstiek en ondersteunende instrumenten zoals informatie over regionale mogelijkheden voor verwijzing en consultatie. Doel van de module was dat huisartsen na de module in staat zouden zijn om: patiënten op te sporen met een erfelijke aanleg voor kanker; een stamboom op te stellen als instrument voor het opsporen van patiënten met een verhoogd risico op erfelijke vormen van kanker en dit te interpreteren; de meest voorkomende soorten erfelijke kanker (borst- en darmkanker) te beschrijven, evenals de genetische afwijkingen die daarbij een rol kunnen spelen; oncogenetische richtlijnen toe te passen bij het bepalen voor welke patiënten verwijzing al of niet zinvol is en te zoeken naar informatie op het internet; de mogelijkheden en beperkingen van oncogenetische testen uit te leggen; met patiënten te overleggen over periodiek onderzoek en risicobeperkende chirurgische opties voor patiënten met een erfelijke vorm van kanker.

Tussen september 2011 en maart 2012 werd de online module gericht op het verbeteren van oncogenetische kennis uitgezet en geëvalueerd onder huisartsen. Een interventiestudie met een controlegroep werd uitgevoerd met parallele groepen en dataverzameling door middel van herhaalde metingen met kennistoetsen en vragenlijsten voorafgaande (pretest), direct na (posttest) en zes maanden (retentietest) na de interventie. Van de tachtig huisartsen die werden gerandomiseerd naar interventiegroep en controlegroep namen 44 (20 in de interventiegroep en 24 in de controlegroep) deel aan alle metingen. Om de validiteit te vergroten werden alleen huisartsen geselecteerd die gevestigd waren buiten de provincies waarin huisartsen ook konden deelnemen aan de live nascholingsmodule (Noord-Holland en Limburg) (zie hoofdstuk 5). De resultaten gaven aan dat de huisartsen zeer tevreden waren over de nascholingsmodule. Drie tevredenheidsitems hadden scores van 4.1-4.3 op een vijf-puntsschaal en de totaalscore bedroeg 7.9 op een tien-puntsschaal. De kennisverbetering gemeten met de post-test en de retentietest bedroeg respectievelijk 0,055 ($P < 0,05$) en 0,079 ($P < 0,01$) met medium effectgroottes (0,27 en 0,31). De deelnemers waardeerden de toepasbaarheid van de kennisaspecten in de praktijk (itemscores 3.3-3.8 op een vijf-puntsschaal). De

scores betreffende het zelfoordeel ten aanzien van herkenning van ziekte, verwijzing naar een specialist en kennis over de mogelijkheden/beperkingen van genetische testen waren min of meer neutraal (2.7-2.8 op een vijf-puntsschaal). De conclusie was dat de evaluatie van de nascholing informatie opleverde voor de verdere ontwikkeling van online modules voor genetische nascholing van huisartsen en kunnen zo mogelijk een bijdrage leveren aan het verbeteren van de patiëntenzorg. De online nascholingsmodule en het daarvoor ontwikkelde raamwerk kan dienen als voorbeeld voor andere specialismen, de basisartsopleiding en het biologieonderwijs op middelbare scholen.

Hoofdstuk 5 beschrijft een RCT waarin onderzocht werd of een “live” ongenetische nascholingsmodule leidde tot een verbetering van de consultvaardigheden van huisartsen. De interventie in dit pragmatische geblindeerde, gerandomiseerde, gecontroleerde onderzoek bestond uit een vier uur durende training in: oncogenetische consultvaardigheden (familieanamnese, bepaling van familiair/erfelijk risico en doelmatig verwijzen), attitude (medisch ethische kwesties) en klinische kennis voor eerstelijns consulten. De interventie liep tussen december 2011 en april 2012. De uitkomsten werden gemeten met behulp van scoringslijsten voor geobserveerde contacten die door onaangekondigde gestandaardiseerde patiënten werden ingevuld en uit zelfbeoordelingvragenlijsten. Om logistieke redenen werden uitsluitend huisartsen geselecteerd die gevestigd waren in twee Nederlandse provincies (Noord Holland en Limburg) die niet deelnamen aan het evaluatieonderzoek van de online module. Van de 88 gerandomiseerde huisartsen die zich in eerste instantie bereid toonden deel te nemen aan het onderzoek, vulden 56 alle vragenlijsten in (pre- posttest, en een retentiemeting op drie maanden na de interventie). De belangrijkste consultvaardigheden die in de training aan de orde kwamen, betroffen het herkennen van klinisch relevante informatie over verschillende soorten erfelijke kanker (borst/eierstok-, darm- en huidkanker) inclusief de genen betrokken bij oncogenetische syndromen en de gerelateerde erfelijkheidstesten; het herkennen van patiënten met verschijnselen die wijzen op een erfelijke vorm van kanker; het opstellen van een stamboom als instrument voor het opsporen van patiënten met een verhoogd risico; het bespreken van een mogelijk risico op familiale en erfelijke vormen van kanker (b.v. regelmatige controles en risicobepalende chirurgische ingrepen) en daarmee samenhangende ethische kwesties; het opsporen van patiënten die in aanmerking komen voor verwijzing voor risicobepaling en het zoeken naar relevante online verwijsinformatie online op basis van oncogenetische richtlijnen; het toelichten van de mogelijkheden en beperkingen van oncogenetische testen; en weten wanneer het zinvol is een klinisch geneticus te consulteren en/of daarnaar te verwijzen. Er werd een significante verbetering gevonden in deze consultvaardigheden direct na de interventie en na drie maanden (regressiecoëfficiënt respectievelijk .34 en .28 met een medium effectgrootte)

lieten de resultaten een blijvende verbetering zien. Huisartsen waren tevreden over de module en gaven een positief oordeel over de praktische toepasbaarheid van de trainingsonderwerpen.

Hoofdstuk 6 betrof een onderzoek waarin met behulp van zelfrapportage werd onderzocht wat de lange termijneffecten (een jaar later) waren bij huisartsen die hadden deelgenomen aan de online en live nascholingsmodules. Daarnaast werden verwijscijfers opgevraagd bij de klinisch genetische centra en werd belangstelling voor en tevredenheid met de website huisartsengenetica.nl bepaald. Deelname aan de modules leidde tot een blijvende verbetering van oncogenetische competenties. De deelnemers gaven aan dat zij alerter waren op genetische problemen; 88% van de deelnemers aan de live training gaven aan dat zij patiënten vaker verwezen naar een klinisch genetisch centrum, terwijl dit gerapporteerd werd door 29% van de deelnemers aan de online oncogenetische training. De klinisch genetische centra rapporteerden echter dat er een jaar na disseminatie van de onderwijsmodules geen significante verandering was in het aantal verwijzingen vergeleken met voor de training. Blijvende belangstelling voor en tevredenheid met de nieuwe huisartsenwebsite werden onderzocht door middel van een “pop-up” vragenlijst voor bezoekers aan de website. Slechts een klein aantal van de bezoekers (38 waaronder 22 huisartsen) vulde de vragenlijst in gedurende de maand waarin het onderzoek plaatsvond. Er werden echter hoge scores gegeven voor tevredenheid met de website en de relevantie van de website voor beslissingen over juiste verwijzingen. Het aantal bezoekers van de website nam toe waarbij de pagina over het opstellen van een stamboom het meest geraadpleegd werd.

In Hoofdstuk 7 wordt een stapsgewijze route voorgesteld om te komen tot integratie van genetica in het elektronisch patiëntendossier (EPD) voor de huisartsenpraktijk en klinisch onderzoek. Dit zou de dringend noodzakelijke operationalisatie mogelijk kunnen maken van goed toegankelijke genetische kennis voor klinisch onderzoek en daarmee voor verbetering van de gezondheidszorg. De grootste urgentie geldt echter voor het bevorderen van genetisch alfabetisme door middel van training en onderwijs. De tweede stap betreft een verbetering van de mogelijkheden om een familieanamnese zodanig te registreren dat zoekstrategieën gebruikt kunnen worden om patiënten met een verhoogd risico op te sporen. Voorgesteld wordt om codes aan het EPD toe te voegen uit de hoofdstukken “A21 Familiegeschiedenis van maligniteiten” en “A99 Ziektedrager niet verder beschreven” in de International Classification of Primary Care (ICPC) en om eenvoudige methodes te bieden voor registratie van de familiegeschiedenis evenals het bevorderen en ondersteunen van codeervaardigheden. Voor EPD's zijn mogelijkheden nodig om (nieuwe) gegevens over de familiegeschiedenis toe te voegen, zoals relaties tussen personen binnen dezelfde familie. Multidisciplinaire richtlijnen voor

verwijzing dienen helder te zijn en geen ruimte te laten voor verschillende interpretaties. Automatische berichten kunnen behulpzaam zijn voor huisartsen om patiënten te herkennen met een verhoogd risico die voldoen aan criteria voor verwijzing. Ter illustratie wordt een patiëntencasus beschreven met familiale borstkanker met een *BRCA1*-mutatie.

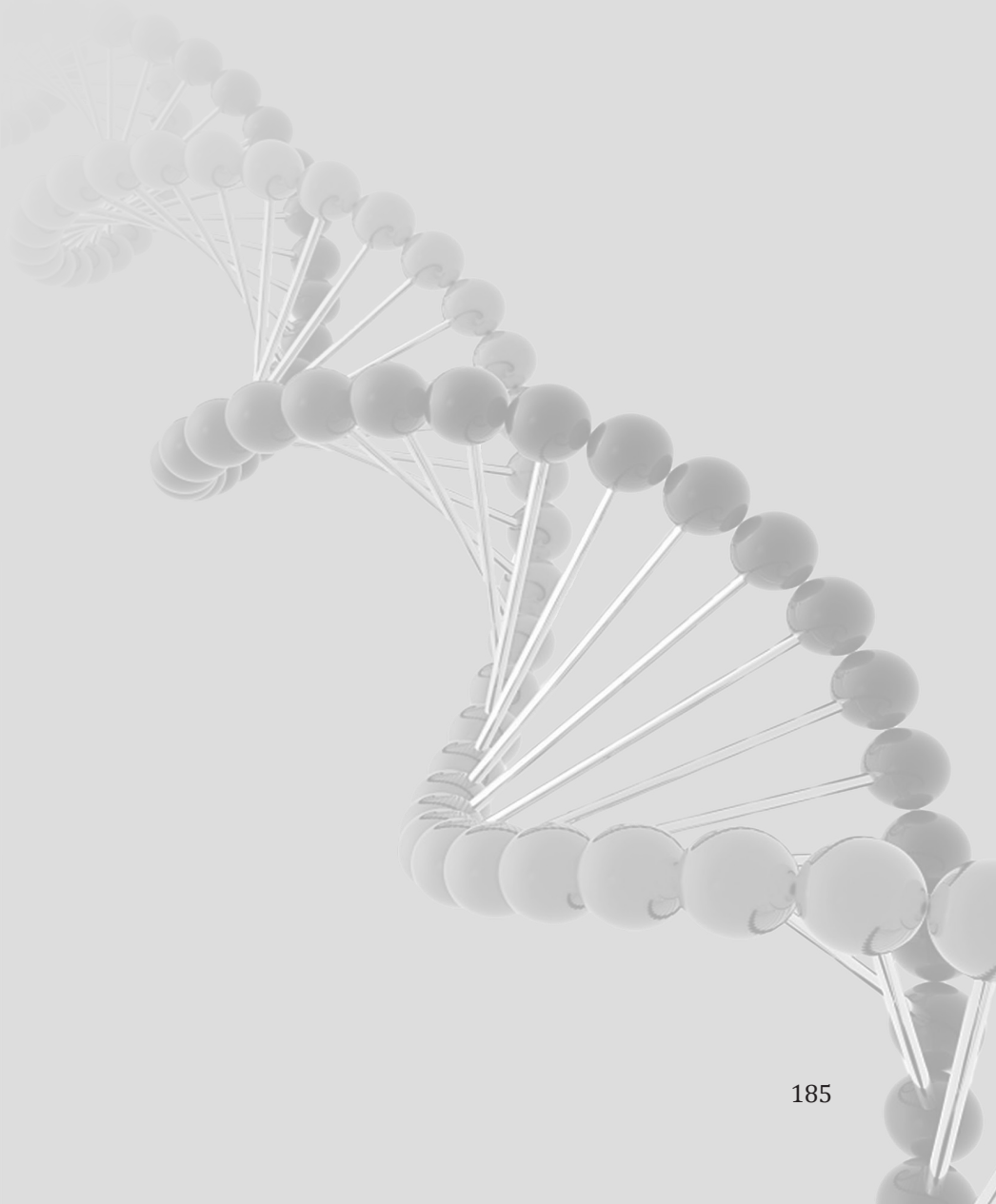
Tot besluit

De resultaten van de onderzoeken die in dit proefschrift zijn beschreven, hebben geleid tot een beter inzicht in de inzichten van niet-genetische specialisten, zoals huisartsen en verloskundigen, omtrent de toegenomen rol van de genetica in de eerstelijnsgezondheidszorg. De bevindingen onderstrepen het belang van genetisch onderwijs. Verder onderzoek is nodig om vast te stellen welke verantwoordelijkheden zorgverleners in de eerste lijn zouden moeten krijgen ten aanzien van de genetica. De speciaal voor huisartsen ontworpen oncogenetische nascholingsmodules ondersteund door de Nederlandse genetica website huisartengenetica.nl lijken een goede en klinisch toepasbare methode om de oncogenetische competenties voor de dagelijkse huisartspraktijk te versterken en tevens een flexibel en doelmatig raamwerk te bieden ter ondersteuning van de ontwikkeling en uitvoering van trainingsactiviteiten die uiteindelijk kunnen leiden tot een verbetering van de genetische zorgverlening.



www.huisartsengenetica.nl

Dankwoord



“Success.

I am fascinated by people who make work look seamless. Take Usain Bolt for example, you look at him run and you see greatness. You see that he looks nothing like the other sprinters. His face, his muscles are relaxed, whereas other sprinters are cringing - you can feel their tension just by watching. But even for Bolt, being talented alone was not enough to win. To win he had to train hard, despite his self-admitted distaste for hard work. The similar concept applies in professional world, as it is impossible to succeed on talent alone anymore – 20 years ago, maybe, but not anymore. Nowadays, success lies at the crossroads of talent and work ethic. And sure, everyone has different amount of talent, but few actually work hard enough to realize the talent they do possess. Therefore, I am inspired by the prospect of realizing my full potential and achieving “seamlessness” at whatever I do.”

*Artour Samsonov on inspiration, London Business School, MBA2011
interview on Impact*

Wat is een Dream team? Samen roeien met de riemen die je hebt, met de neuzen dezelfde kant uit en met een hart vol passie voor waar je mee bezig bent. Ik mag mijzelf gelukkig prijzen dat ik met zoveel zeer talentvolle, gepassioneerde, sportieve, lieve, inspirerende, ambitieuze mensen, collega's, vrienden, ouders, man en kinderen nu zover ben dat ik mijn proefschrift mag verdedigen. Ik had dit NOOIT alleen kunnen doen! Dit betekent voor mij absoluut geen einde maar een continuering van waar ik al jaren mee bezig ben: doen wat ik leuk vind en er met de volle 100% voor gaan! There is so much more to come!

Als je als huisarts wilt promoveren, twee kleine kinderen hebt, een man hebt die ook graag en goed academisch werkt als Cardioloog, graag wilt blijven rennen en roeien, ook soms nog eens wilt afspreken met vrienden en familie, dan moet je prioriteren en dus grenzen stellen. Dit wordt je niet altijd in dank afgenomen. Er wordt aan je getrokken en geloof in jezelf en wat je doet wordt dan wel eens op de proef gesteld. Maar ik ben iemand die sterker wordt door tegenwind en doorgaat als het nodig is. Dit project kon en wilde ik dus ook niet alleen doen en met alle hulp en steun is het gelukt! Een dankwoord is dus ook in mijn proefschrift onmisbaar, ook al bestaat deze uit vele pagina's.

“We tillen de boot uit het water. We trekken een doek over de cederhouten huid om hem af te drogen, brengen ‘m naar binnen en gaan de riemen halen. En dan, terwijl we samen naar de loods lopen, ieder een riem in de hand, slaat David heel even een arm over mijn schouder. Niet vriendelijk, maar vriendschappelijk. Niet spottend, maar gemeend. Ik ben zo moe. Ik knijp in het hard hout van de riem en voel mijn spieren nog een keer spannen. Er golft een diep geluksgevoel door mijn handen, mijn armen, mijn schouders, mijn borst en mijn benen. Ik ben moe en gelukkig.”

H.M. van den Brink, Over het water. 1998. Uitgeverij Meulenhof.

Ten eerste zou ik het Centre for Society and the Life Sciences (CSG) willen danken voor hun steun. Onze CSG dagen zijn na de CSG Academy ook na de promotie doorgegaan. De financiële bijdragen en jullie steun, ook vanuit het Center for Medical Systems Biology (CMSB) en Netherlands Genomics Initiative (NGI), maakten het mogelijk om te doen wat we wilden doen: goed onderwijs opzetten, implementeren en vervolgens gedegen evalueren. De eerstelijns zorg (maar hopelijk ook tweedelijns en medisch/biologie/middelbare school studenten) kan zo weer een hele stap vooruit zetten en samenwerking binnen de eerste en met de tweedelijns verbeteren op het gebied van genetica en genomics, maar belangrijker nog de gezondheidszorg vooruit helpen. Daarnaast natuurlijk ook al mijn collegae bij de afdeling klinische genetica van het VU medisch centrum en sectie Community Genetics en de vakgroep Huisartsgeneeskunde in Maastricht. Als flex-er en parttime dokter/onderzoeker heb je niet altijd mogelijkheid om meer tijd in de sociale relaties met collegae te steken. Dat heb ik wel erg jammer gevonden en ik hoop dan ook na mijn promotie nog lang met jullie samen te mogen werken. Ik heb de tijd die we wel samen hebben gehad erg gezellig gevonden. De NHG congressen, NACGG dagen en ESHG congres in Parijs waren een stuk minder gezellig geweest zonder jullie!

Ik zou ook graag al mijn collegae huisartsen, praktijkondersteuners en assistenten willen danken. In Medisch Contact stond (23 mei 2013) een stuk over gebrek aan wetenschappelijk onderzoekers onder huisartsen. Collegae zoals jullie maken het mogelijk te doen waar onze passie ligt: combinatie praktijk met onderzoek en zo de huisartsgeneeskunde op een hoger niveau brengen. Ook de patiënten wil ik graag bedanken: ook jullie hebben me op dagen gezien dat ik nachten had moeten doorwerken aan mijn onderzoek en met wallen op het werk met koffie in de hand er stond. Ik heb menigmaal een hart onder de riem gekregen door opmerkingen hoe belangrijk het is dat je als arts leert praten over genetica (een Klinefelter pati-

DANKWOORD

ent), drie generaties aan dames die met elkaar in gesprek gingen over borstkanker in hun familie en hun DNA lieten onderzoeken. Heel veel dank!

Ik wil het Nederlands Huisartsen Genootschap bedanken voor hun ondersteuning bij de duurzame ontwikkeling van het genetica onderwijs en de samenwerking tijdens het onderzoeksproject. Martina en ik gingen in de zomer van 2010 enthousiast praten met Rob Dijkstra (toen nog directeur implementatie bij het NHG, nu bestuursvoorzitter en directeur NHG) over het ontwikkelen van onderwijs over genetica. Kanker bleek het thema voor 2011. Jullie omarmden met jullie team enthousiast het onderwerp, hoewel het ook jullie veel tijd kostte en kanker bleek natuurlijk een goede kapstok te zijn om genetica aan op te hangen. Ik wil met name noemen: Rob Dijkstra, Dorry Cox, Shyama Sewpersad, Florijn Jonkers, Vroon Pigmans, Liesbeth van der Jagt, Mireille Ballieux en Els Bakker. Maar er zullen waarschijnlijk velen zijn die ik nu niet noem, maar op de achtergrond een grote rol hebben gespeeld. Jullie vertrouwen en inzet heb ik erg kunnen waarderen. Wij hopen dan ook dat het niet alleen bij "Kanker en genetica" blijft, maar dat we ook verder kunnen gaan met samenwerken, andere onderwerpen, buiten onze landsgrenzen, de huisartsen in de rest van Nederland en mogelijk ook andere zorgverleners in eerste en tweede lijn. We zijn al begonnen om verdere uitbreiding van het onderwijs op te zetten zodat niet tientallen maar honderden huisartsen bereikt kunnen worden.

Annemieke van Dijk, Petra Elders, Bestuursleden Huisartsenkring Amsterdam, 1e Lijn Amsterdam, WDH HOZL, ZIO (RHZ Heuveland) en WDH Meditta (Guy Schulpen, Jan van Rooij, Merijn Verburg, Roger Eurelings, Esther van Engelshoven, Frank Soomers, Safira Quick, Mischa van der Graaff, Irmgard Rietbroek, Guyonne Boskamp, Brigitte Paulissen): als collega huisartsen actief betrokken bij opzetten nascholingen en als medewerkers betrokken bij de regionale organisaties voor huisartsen in Limburg en Noord Holland hebben jullie mijn verzoek om ons onderwijs te ondersteunen met enorm veel enthousiasme opgepakt. Jullie hebben ons blijvend ondersteund waardoor huisartsen in staat waren geïnformeerd te worden over de nascholingen via jullie bij hen bekende websites en locaties. Maar ook de manier van communiceren heb ik erg prettig gevonden en hoop dan ook dat jullie bereid zijn dit in de toekomst te blijven doen: want we gaan door met regionaal genetica onderwijs!

Onze website www.huisartsengenetica wordt goed bezocht, met de samen opgezette webredactie (Lidewij Henneman, Marloes Brouns, Klaas Dolsma, Kristel van Asselt, Martijn Sijbom, Cecile Janssens, Petra van Overveld, Martina Cornel, Florijn Jonkers) willen we de kwaliteit hoog houden zodat huisartsen ook in de toekomst weten waar zij hun informatie over genetica kunnen blijven vinden. Marloes: heeft veel dank voor onze gezellige samenwerking en onze goede gesprekken.

“Persistence. It is the greatest lesson I learnt, and I learnt it from my college rowing coach (Harry Parker, Harvard University). Before every race, he would tell us to be patient. The thought was that you might not be able to break your competition right away but wear them down over the course of the race. The danger of trying to break your competition in a race is that if you are not able to achieve it quickly, you end up breaking yourself. By focusing on being patient, the focus remained on the process of preparing for a race and racing rather than the desired result (winning). After 4 years of racing in college, I started to appreciate the point that results often take time, and if you focus on the process rather than result, the results will always come out much better than the other way around.”

Artour Samsonov (Harvard alumnus, Olympic rower and married to one of my best friends Sue) on his greatest lesson learnt

Ik weet niet of ik een goede volgorde aanhoudt met iedereen bedanken, maar ja dan grijp ik weer terug op roeien: iedereen in de boot is belangrijk, je steunt elkaar en je moet samenwerken, de cirkel moet als het ware rond zijn en de roei haal moet als een machine lopen als ware het de ketting van een fiets. Sommige roei team leden zullen beter zijn in de eindsprint, anderen in het schoonhouden en verzorgen van de boot en de etentjes, anderen zullen de stuur-tjes vragen om te komen naar de trainingen. Maar iedereen is onmisbaar. Van boeg tot slag en stuur-vrouw of man. De Dames 8+ van MWC: Ik kon de afgelopen jaren niet veel mee-doen, meedrinken, meelachen, etc etc, maar ik hoop dat ik na het afronden van de promotie weer vaker kan instappen en het team weer compleet kan maken!;))) Heb jullie gemist en onze borrels op het terras aan de Maas zijn heerlijk in de zon. De BBQ in onze tuin zit hier net ver vandaan! Hans onze roeisessies om 6u 's och-tends gaan ook door!

Dé VNVA kadertrainingen van Ina Vader heb ik ook als een bevrijding ervaren. Ongelooflijk de teamspirit en dat we nog steeds bij elkaar komen op de mooiste plekken in Nederland, lekker decadent! Ik had meer contact willen houden en hoop dat dat nu weer wat makkelijker gaat. Jullie zijn zulke sterke dames en heb-ben me er echt doorheen getrokken, een voorbeeld voor elkaar. Ina, ik hoop dat je nog lang door zult gaan met de trainingen, vrouwelijk artsen ontberen steun om zichzelf verder te ontplooiën en jij doet dat op zo'n respectvolle manier waar je je veilig bij voelt om nooit te vergeten en direkt te gaan toepassen in de praktijk. Heel veel dank! Prof. dr. Henriette van der Horst attendeerde mij op deze training en werd vanuit de VNVA mijn coach de afgelopen jaren om werk en prive op heel persoonlijk niveau te bespreken. Heel veel dank daarvoor Henriette!

DANKWOORD

Ook wil ik graag de Working-Ladies (annex borrelclub) en de Oudervereniging van OBS Binnenstad bedanken voor jullie steun. Je brengt als ouder relatief veel tijd door op school en wat is er dan leuker als je dan ook nog leuke, interessante, lieve, ondersteunende dames/ouders/vrienden en vriendinnen tegenkomt die je bestaan als ouder combi huisarts combi promovenda kunnen ondersteunen? Marieke deed mee aan de avond 4 daagse, twee avonden werd ze opgehaald door Ghislaine, mama Jip. Je merkt dan hoe je elkaar steunt. Ja ja, je bent de avond 4-daagse service ☺. Het is een hectisch jaar geweest, van zieke vader ver weg in Amsterdam, verkoop oude huis met heen en weer fietsen en dan verhuizen naar ons prachtige huis aan de Aylvalaan, Marieke die tussendoor ook nog moest leren lezen en mijn hoofd dat bepaald niet naar fantasievol lezen stond en naar woordjes samen puzzelen om 19u 's avonds.... (Oh ja even voor de duidelijkheid, ik ben super trots: ze leest nu super!). Samen koffie op de woensdagmiddag bij de ene moeder om de een weg te brengen en dan hups naar de andere moeder op de bank om de ander op te halen, maken het leven soms wat lichter als het even niet zo licht is. Het zijn vaak die parels die het leven mooi maken.

Promoveren is net als wonen in Maastricht,
je moet het voortdurend aan iedereen uitleggen.
Amy Jonk, Maastricht in NRC, 04-01-2012

Voordat ik schrijf over mijn onderzoeksteam/groep, eerst alle aandacht voor de assistentes/secretarissen/secretaris: Marianne Hardonk en Wilma IJzerman in Amsterdam, Frits Ruijters en Ine Siegelaar in Maastricht: Jullie hulp en luisterend oor maakt het daadwerkelijk mogelijk om te doen wat ik doe en wat ik wil blijven doen: combineren van huisartsgeneeskundige zorg met onderzoek. Heel veel dank!

Ook wil ik Dr Irina Stirbu-Wagner van het LINH- NIVEL instituut danken voor onze samenwerking. We hebben de afgelopen jaren een aantal keren uitgebreid overlegd over verwijzingen van de huisartsen naar specialisten. Jouw ervaringen en enthousiasme hielpen mij erg te leren begrijpen hoe ingewikkeld dit proces is en hoeveel tijd het kost voordat er veranderingen in verwijsgedrag in de vorm van cijfers kunnen worden opgemerkt. Ik hoop dat we elkaar nog vaak zullen vinden!

I would also like acknowledge my inspirational research bosses in the USA: prof Carol Warner (Northeastern University, Boston, MA: you started the spark), prof McDonough (Medical College of Georgia, Augusta, GA: you made it possible for me to combine rowing in the Olympics team on the Savannah river with working in the laboratory a few hours a day) and dr Carolyn Schanen (UCLA, Los Angeles, CA: for making me understand how inspirational it can be to work on translating genetics into the clinic as a physician). Thanks!

Tja, maar waar zou ik zijn zonder mijn dream team OVOGG (Onderzoek van Onderwijs over Genetica en Genomics om volledig te zijn) vergaderingen (in Maastricht) en OVO (Onderzoek van Onderwijs) team vergaderingen (in Amsterdam)? Deze vergaderingen werden altijd enthousiast bezocht met altijd een heerlijke lunch. Ik wil jullie toch allemaal noemen: prof Connie Stumpel en prof Albert Scherpbier: heel veel dank voor jullie hulp met name bij de opzet van het project en me blijven aansporen om door te gaan, ook na het afronden van mijn promotie. Alle begin is moeilijk, jullie ervaring, expertise en enthousiasme heeft veel betekent. Heel veel. Dr Denhard de Smit, jouw aanwezigheid maakte dat we als een groot team konden samenwerken. Als ik aan jou denk, denk ik aan toekomst en aan samen met het NHG duurzaam onderwijs opzetten voor huisartsen en huisartsen in opleiding. Dr Scheltus van Luijk, Amsterdam blijft trekken, maar Maastricht geeft ook veel ruimte! Ik ben blij dat ook jij inziet dat Amsterdam-Maastricht daadwerkelijk bereisd kan worden met de trein! Jouw hulp als voorzitter tijdens de focusgroepen gaf een goed beeld van de inventarisatie en duidelijkheid en maakte dat we in staat waren binnen afzienbare tijd een mooi artikel te publiceren. Maar ook later bleef je met je steun en aanwezigheid erg belangrijk. Jouw expertise in het onderwijs gaf mij vaak helderheid over hoe je met veel plezier goed onderwijs kunt opzetten. Mereke Gorsira, wat geweldig dat je ondanks je drukke werkzaamheden ook nog de tijd hebt gevonden om mijn teksten na te kijken. Acceptatie van de artikelen is ook dankzij jouw hulp voorspoedig gelopen. Dank!

Overige leden van de promotie- en leescommissie:

Prof Gerda Croiset, ik leerde jou kennen tijdens de cursus van prof Albert Scherpbier in Utrecht: Onderzoek van Onderwijs. Jouw bevoegenheid is erg aantekelijk en je bent als vrouwelijk hoogleraar een groot voorbeeld voor mij maar ook voor anderen. Bedankt voor je enthousiasme en onder jouw leiding heerlijk gegeten en zeer interessante discussies met de arts assistentes in Maastricht!

Prof Joep Geraedts, wij kennen elkaar al jaaaaaren! Vlak nadat ik terug was uit Amerika van roeien en onderzoek mocht ik al een presentatie houden over mijn Rett onderzoek en resultaten. Ik heb deze aandacht zeer positief ervaren en uw aanhoudende enthousiasme werkt lang door. Nog steeds zijn we regelmatig in gesprek over hoe we het genetica onderwijs binnen de geneeskunde opleiding en opleiding voor medisch specialisten/huisartsen/aios kunnen implementeren.

Prof Hanne Meijers-Heijboer, beste Hanne, jouw enthousiasme tijdens de nascholingen voor huisartsen is zo aantekelijk geweest. Met jouw ervaring als klinisch geneticus, opleider en contacten met huisartsen over de jaren weet jij als geen ander de zaal mee te krijgen. We hebben elkaar ook erg persoonlijk leren kennen en hoop dan ook dat we na mijn promotie nog contact houden. Maastricht is een heerlijke Bourgondische stad waar je altijd welkom bent!

DANKWOORD

Prof Mattijs Numans, ik heb jou via Annet Sollie leren kennen en als bevlogen collega huisarts en hoogleraar op het gebied van Vernieuwen en verbeteren kwaliteit eerstelijns zorg natuurlijk degene waarvan ik hoopte dat hij in de promotiecommissie wilde plaatsnemen. Ik voel me dan ook echt vereerd dat je dit wilde doen en hoop nog lange tijd met je te kunnen samenwerken.

Prof Onno van Schayck, dank dat ook jij in mijn promotiecommissie wil deelnemen. Als hoogleraar preventieve geneeskunde en befaamd wetenschapper kon ik u natuurlijk niet missen, maar ja dat had ik natuurlijk al een paar jaar daarvoor met uw vrouw Cornel van Schayck besproken! Zij sprak tijdens de simulatiepatiënt trainingen in Maastricht hartstikke trots over u en nadat ik u een paar keer had gesproken en uw werk had gelezen hoopte ik dat u niet zou ontbreken. Heel veel dank!

Prof Connie Stumpel, ook jou ken ik natuurlijk al erg lang, zolang als prof Joep Geraedts in feite. Ja, het proefschrift is er eindelijk, na al mijn jaren werk aan Rett syndroom, maar goed, het leven is dan ook een weg die je mag bewandelen en meestal is de weg interessanter dan het doel op zich. Nu hoop ik op meer en verdere verbetering en vernieuwing van het onderwijs. Ik hoop dan ook dat we nog lange tijd zullen samenwerken. In Parijs tijdens het ESHG kwam je na mijn praatje erg enthousiast naar mij toe. Jouw steun aan het begin, maar dus ook aan het eind, van het project was onmisbaar, een goede basis betekent alles (maar goed daar weet je als klinisch geneticus en kinderarts natuurlijk alles van). Heel veel dank.

Dr Alan Guttmacher, we also met a long time ago and when we met I was very inspired by your enthusiasm to truly implement genetics in main stream medicine and making it possible to register family diseases. We have been in touch over the years and you have definitely been able through our mails, publications and interviews to re-spark my enthusiasm to proceed with genetics education in primary care. Thanks!

Deelnemers aan de focusgroepen, Delphi studie, PIN, nascholingen, ECWO studenten, simulatiepatiënten jullie steun is onmisbaar geweest. Woorden kunnen niet uitdrukken hoe ontzettend dankbaar ik ben voor jullie steun en deelname aan dit onderzoek. Jullie hebben het mogelijk gemaakt om genetica onderwijs vorm te geven.

Sarah, Jouke en Steven: jullie hulp is hard nodig geweest! Ik weet niet hoe ik jullie kan bedanken. Mijn dagen lijken te kort, zou zo een paar uur extra willen. Maar misschien zijn ze juist zo kort zodat je leert om met zulke geweldig getalenteerde studenten en (toekomstig) collegae samen te werken en te begeleiden. Ik hoop dat ik jullie ook wat heb kunnen leren en in de toekomst kan blijven spreken.

En dan mijn OVO *Dream* team:

Prof Cees van der Vleuten, Cees, als copromotor en als wereldberoemd expert op het gebied van onderzoek van onderwijs was jij mijn Maastrichtse wereldbegeleider. Wat een dream team lid. Jouw positiviteit gleed als een warme deken over het project. Je was altijd zo snel in je antwoorden en altijd positief. Mocht ik ooit een psycholoog nodig hebben dan hoop ik bij jou langs te kunnen komen ;-))

Dr Lidewij Henneman, Lidewij, wij kennen elkaar eigenlijk ook al langer dan vandaag. ☺ Ik zal niet zeggen hoe, maar toen ik dat wist dacht ik: dat komt wel goed. Jouw steun is super geweest. Bloed zweet en tranen, tja daar weet jij nu met mij alles van. Ik denk altijd: no limits! Jij denkt altijd: stay in the moment! Een super combi lijkt me zo. Ik hoop dat ik nog lang op je mag rekenen en jouw ervaring als klankbord mag gebruiken.

Prof Geert Jan Dinant, is het nu Geert-Jan, Geert Jan, GeertJan of GJ? ;-))), heel veel dank dat je mijn promotor wilde zijn in Maastricht. Als collega huisarts en hoogleraar huisartsgeneeskunde, kwam jij met al je ervaring vaak met zeer innoverende en enthousiaste ideeën naar voren. Als gedetacheerd huisarts-promotie-onderzoeker moest ik mijn draai vinden op de derde verdieping in Maastricht. Heel veel dank met je hulp daarbij. Jouw mooiste en beste advies aan mij was dat ik bij mijn kern moet blijven: "Jouw ideeën zijn goed, probeer niet nu al te veel te differentiëren!". Ik hoop dat we nog lang samen kunnen blijven werken!

Prof Martina Cornel, Martina jij bent meer dan alleen een promotor. We hebben de afgelopen jaren al heel wat meegemaakt samen. Je bent integer en hebt je hart op de goede plek zitten. Ik weet nog toen we begonnen aan dit project, ik je al bij voorbaat heb gezegd dat eerlijk zijn bij mij een voorwaarde is om goed samen te werken. Helaas heb ik door schade en schande ook andere ervaringen. Martina heeft mij daarin absoluut niet teleurgesteld en ik heb dan ook zoveel van je geleerd. Dank dat je mij het vertrouwen in de wetenschap weer hebt teruggegeven. Ik hoop dat ik je naast mijn promotor ook dierbare vriendin mag blijven noemen na alles wat we hebben besproken en meegemaakt. Het is echt een eer om binnen jouw sectie mijn promotieproject te hebben mogen afronden. Ik hoop dat we nog lang zo doorgaan. Je bent met de sectie en je familie altijd welkom in Maastricht, het lekkere eten, de wijn en de logeerverdieping staan klaar!

LEF

Breek de regels.

Volg nooit de uitgestippelde paden.

Probeer wat anders te doen, ook al vertellen mensen dat je gek bent.

Alles wat je kunt dromen is mogelijk, het enige wat je nodig hebt is lef.

Vrij naar Paulo Coelho, Dagblad De Pers, Maandag 26 mei, 2008

DANKWOORD

Vrienden (Angele, Michiel, Carina, Vincent, Carijn, Rob, Bjorn, Marian, Hans, Lana, Buckley, Sue (can't wait to hold the BLOB in my arms, Artour (thanks for your quotes! Rowing is truly inspirational and taught me so many lessons as well. It is great to have both of you as our friends and find we are on the same wavelength. Thanks for reminding me :))), Esther, Annet, Pieter, Rachel, Paul, Ghislaine, Stephan, Annemieke, Joel, Ingrid, Marije, Michiel, Simone, Leon, Lonneke, Pieter, Peter, Anke, Bea, Esther, Hafida, Swee, Blandine et Hakim, Susan en Hans, Kim, Robin, Renate, Narendar, Ineke, Bram, Jola, Kathleen, Bianca, Iris, Jolanda, Thea en Yuri, To en die vrienden die belangrijk zijn en zijn geweest op alle leuke en minder leuke momenten in ons leven. Vrienden zijn voor mij en voor ons echt onmisbaar gebleken in de afgelopen jaren. Sommigen zie ik vaker dan anderen, anderen zie ik door de afstand bijna nooit. Maar altijd weet ik dat we op jullie kunnen rekenen. Can't wait to see you again and have some great laughs!

I would really like to stop here and also thank my sisters in Long Beach California for being there for me in good times and in bad. If it was not for you, this thesis would not have been here. You know why. I hope we can soon meet again. Your warm hugs, smiles, prayers, emails, calls and welcome back party at Ann's house are unforgettable: I love you!

Maar ook onze nieuwe burens op de Aylvalaan. In een enerverend jaar als dit jaar, voelt het als een warm bad waar we in terecht zijn gekomen. Het klinkt misschien gek om ook de burens hier te noemen. Maar met ouders en soms ook vrienden ver weg is het voor ons echt bijzonder om jullie dichtbij altijd klaar te hebben staan als we de babyfoon even willen afgeven voor een spoeddienst, rennen op de berg, wielrennen sorry mountainbiken met buurvrouw en eten in restaurantje in de buurt. Jullie zijn altijd welkom en wij bij jullie. Ondanks onze drukke levens staan we altijd klaar voor elkaar en is het drinken van een wijntje samen dan net wat gezelliger (lekker snel thuiskomen ;).

Mijn paranimfen: Annet en Angele. Beiden gepassioneerde moeders, vriendinnen, artsen en onderzoekers (volgorde nog nader te bepalen;). Jullie blijven mij inspireren en ik hoop ik jullie. Dank voor al jullie hulp en ondersteunende gesprekken. De toekomst roept en de logeerverdieping in Maastricht staat natuurlijk ook voor jullie klaar! Gezelligheid kent geen tijd...

TO "LET GO"

Is not to cut myself off,
It is the realization
I can't control another

Pa en ma. Jullie zijn mijn hele leven heel belangrijk geweest bij het ondersteunen van alles waar ik mee bezig ben geweest. Maar ik denk ook dat jullie vaak hebben gedacht: waar is ze mee bezig? Ik herinner me hoe mijn vader bij de USA Nationaal roei team trainingen in Augusta, Georgia, met zijn cowboy hoed op stond met zijn lange regenjas aan, handen in de zakken, waarschijnlijk met krullende tenen en ons zag trekken aan die ergometers als bezetenen. Je zag hem denken: is dit nu leuk? Ja dat was leuk, en we waren trots, net als nu! Het afgelopen jaar heb ik met heel mijn hart en ziel zitten huilen aan je bed, angst dat ik je zou verliezen. De angst dat wij zonder jouw passie voor het leven door moesten was zo groot. Wij hebben als gezin ons best gedaan om vanuit Maastricht zo vaak mogelijk in Amsterdam jou en mama te ondersteunen. Wij zijn zo gelukkig dat je nu weer kunt genieten van het leven en met mama en vrienden naar Noordwijkerhout, camper, bootje op de Amsterdamse grachten, Ibiza, Frankrijk en Spanje kunt en daar kunt genieten van zon, zee, drank en spijzen. Ga hier nog lang mee door! Mama en papa: jullie liefde voor onderwijs, het huisartsen vak en jullie praktijkteam zullen me blijven inspireren.

Lieve Bas, Marieke en Joppe: jullie zijn mijn Dreamteam thuis en zijn onmisbaar bij alles wat mij bezighoudt! Bas onze gezamenlijke passie voor elkaar, de wetenschap, rennen, skiën en lekker eten waren onze eerste aanzet voor onze relatie. We kenden elkaar natuurlijk al 10 jaar toen we elkaar (weer) tegenkwamen in de Perroen tijdens de carnaval in Maastricht, maar wetenschap is wat onze liefde urenlang op de bank (ja echt) heeft aangewakkerd en dit nog steeds doet. Wij houden samen alle ballen in de lucht en mogen er trots op zijn dat we dit (meestal) met plezier blijven doen. Werk is ook maar werk, sport is ook maar sport, vrienden en familie zijn... nou ja. Jullie zijn meer dan de icing on the cake, zonder jullie heb ik geen zin in die cakejes. Een glimlach, zoen, knuffel zorgen voor het relativeren van alle ups en downs.

Ik hou van jullie, met heel mijn hart en ziel hou ik van jullie!

Liefde is...

... onze eigen koers naar het geluk varen.

Curriculum vitae



CURRICULUM VITAE

Isa Houwink was born on December the 13th 1973 in Haarlem, the Netherlands. After she received secondary education at the Barlaeus Gymnasium in Amsterdam, the Netherlands, she successfully passed the Propadeutics in Health Sciences. In 1994 she started studying medicine at Maastricht University and graduated March 2003. Her interest in science started during her science rotation in Boston with Prof. Dr. Carol Warner, Northeastern University, Massachusetts, USA, for which she won the Student Science Award in 1998 (“Aspects of preimplantation embryology and sex- related growth rate differences in mouse preimplantation embryos in the Ped fast and Ped slow strain *in vivo*.”). During her internship she rowed in Boston early mornings and early evenings at the Union Boat Club on the Charles River, where she was scouted and asked to row in the National team in Augusta, Georgia. She then decided to postpone the rest of her rotations and work on her dream to be part of the Olympic team for Sydney in 2000. Her interest in genetics and science led her to the Department of Human Genetics, UCLA, Los Angeles, California, where she was able to work with Dr. Carolyn Schanen on Rett syndrome research and methyl CpG binding protein MECP2 mutations.



Maastricht University was kind enough to let her fulfill her combined sports and scientific dreams and helped her to come back in 2001 and she successfully finished her internships in 2003. After obtaining her medical degree she started working as a resident at the departments of Obstetrics and Gynecology both in Amsterdam and in Heerlen. She then decided to become a General Practitioner to be able to be in a more personal relationship with her patients and started her residency in Maastricht in 2005. Since March 2008, Isa is a part-time practicing General Practitioner, where she is still able to show she is a family doctor with special interests in gynecology. In 2006 she published an accredited written Accredidact CPD module on pre- and postnatal screening together with prof. Martina Cornel. In 2010, together with prof. Martina Cornel, she also published a book on “Genetics in general practice” published by Elsevier in the series of Praktikum Huisartsgeneeskunde. From 2008 until 2013 she participated in a PhD project at the section Community Genetics, Department of Clinical genetics (VU University Medical Center Amsterdam) under the supervision of prof. Martina Cornel and prof. Geert Jan Dinant (Department of Family Medicine in Maastricht). Her project, funded by CSG Centre for Society and the Life Sciences /Centre for Medical Sys-

tems Biology, aimed to reflect on current genomics developments with primary care workers, and to help them identify their learning priorities.

In her free time she loves to row, yoga, run and ski with her husband Bas and is a mother to their children Marieke and Joppe, travel, read, enjoy her hobby photography and spend as much time in her house and garden with friends and family as possible.

List of publications



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E.J.F. Houwink European Society of Human Genetics Conference, Paris France, Effects of multifaceted oncogenetics training for general practitioners. Session: C10 Genetic Counselling, Education and Public Service. Nominated for Young investigators Award. **Oral presentation** *June 10th 2013.*

E.J.F. Houwink Erfocentrum & CSG Conference Utrecht, the Netherlands. Wat worden we wijzer van genetische testen? Erfelijkheid in de spreekkamer (voor huisartsen en andere medisch professionals). **Workshop** *May 24th 2013.*

E.J.F. Houwink. Utrecht University, Soesterberg, the Netherlands. Effects of multifaceted oncogenetics training for family physicians on satisfaction, learning and behavior: A report of two randomized controlled trials. Genetics education for the 21st century. Design Criteria and Good Practices. **Poster and International Expert Workshop**, *March 14th-16th 2013.*

Elisa J. F. Houwink, Lidewij Henneman, Myrte Westerneng, Scheltus J. van Luijk, Geert Jan Dinant, Cees van der Vleuten, Martina C. Cornel. Nürnberg, Germany. Creating an Agenda for Effective Genetic Educational Strategies: Needs Assessment and Prioritization in Primary Care. **Poster**. *June 23rd-26th 2012.*

E.J.F. Houwink De Landelijke Organisatie Van Aspirant Huisartsen (LOVAH) Conference 2012.. Werkgroep: Genetica in de huisartsenpraktijk. **Presentation and Workshop**. *January 27th 2012.*

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- E.J.F. Houwink**, S.C.M. van Esch, C.G. van El, F.J. Snoek and M.C. Cornel. Therapeutic Patient Education (TPE), Budapest, Hungaria. The challenge of diabetes genetic risk testing for the primary care setting: what about clinical utility? **Poster.** *November 5th- 8th 2008*
- E.J.F. Houwink**, C. Schanen. Maastricht Medical Students Research Conference, Maastricht, The Netherlands. Phenotypic manifestations in MECP2 mutations in classical and atypical Rett syndrome.. **Poster.** *April 2003.*
- U. Francke, M. Wan, S. Lee, X. Zhang, **E.J.F. Houwink-Manville**, H. Song, R. Amir, S. Budden, S. Naidu, J.L. Pereira, I. Lo, H. Zoghbi, C. Schanen. Biological Psychiatry. Rett syndrome and beyond: Recurrent spontaneous and familial mutations at CpG hotspots in the methyl-CpG binding protein-2 gene. **Poster.** *2000.*
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Weekboek Stille engel
- E.J.F. Houwink**, Published on: 20-05-2011 , *Ned Tijdschr Geneeskd.*
2011;155:B706 Weekboek Topsport
- E.J.F. Houwink**, Published on: 30-05-2011 *Ned Tijdschr Geneeskd.* 2011;155:B708
Weekboek Een passend puzzelstukje
- E.J.F. Houwink**, Published on: 03-06-2011 *Ned Tijdschr Geneeskd.* 2011;155:B709
Weekboek Perfectie als maatschappelijk fenomeen
- E.J.F. Houwink**, M.C. Cornel, MC. Online publication Kankerrisicotest: Gemiste kans: <http://www.ntvg.nl/node/652619>. 2012. Reactie op artikel: *Ned Tijdschr Geneeskd.* 2012;156:A4888

Book publication

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Prizes

- 2010 Putting plans into practice prijs CSG (Center for Society and The Life Sciences):
Title project: Developing successfully implemented guidelines for cardiovascular risk management in genetics/genomics for general practitioners.
E.J.F. Houwink, Prof. M.C. Cornel, in collaboration with Prof. Irene van Langen
- 1999 **E.J.F. Houwink**. Studenten Wetenschapsprijs, Student Science Award; Universiteit van Maastricht, Faculteit der Geneeskunde for her science internship with Prof. C. Warner at Northeastern University, Boston, MA, USA. Aspects of preimplantation embryology and sex-related growth rate differences in mouse preimplantation embryos in the Ped fast and Ped slow strain in vivo.

Appendices



1. Website www.huisartsengenetica.nl

CONTACT **ZOEKEN**

Huisarts en genetica

HOME | AANDOENINGEN | OVERERVING | FAMILIEANAMNESE | COMMUNICATIE | VERWIJZEN | DIAGNOSTIEK | ETHIEK

STOET / VKGN

Welkom

Deze website over genetica is ontwikkeld voor en door huisartsen en maakt het mogelijk snel en gemakkelijk een antwoord te vinden op vragen over erfelijkheid.

U vindt hier:

- Algemene informatie over erfelijkheid en erfelijke aandoeningen
- Casuïstiek
- Links naar andere websites met informatie over genetica

NIEUWS

Nascholingscursus erfelijke kanker voor huisartsen

22 juli 2013. Mediciara Projects organiseert in samenwerking met het NHG [in](#) het najaar in plaatsen verspreid over het hele land cursussen "erfelijke kanker in de spreekkamer".

De nascholing is bestemd voor huisartsen en is speciaal gericht op het omgaan met de verschillende aspecten en vragen rondom de erfelijkheid van kanker in de spreekkamer.

Inschrijving en informatie over het programma, de data, locaties en opzet van deze Standaard in de Praktijk (StP) cursus, [klik hier](#).

Erfelijke hartstoesnissen in Limburg

Op zaterdag 15 juni verschijnt in de weekly van Dagblad De Limburger een artikel over erfelijke hartritmesoosnissen in deze provincie. Het artikel gaat in op recente bevindingen bij patiënten met plotselinge hartstilstand of ritmesoosnissen, waarbij tevens dragerschap is vastgesteld van de SCN5A-mutatie delPhe1617. SCN5A is het gen dat codeert voor het cardiale natriumkanal.

De dragers van SCN5A delPhe1617 blijken familie van elkaar te zijn en wonen overwegend in de oostelijke helft van Zuid-Limburg (Vijlen, Mechelen, Gulpen, Parkstad-regio). Stamboomonderzoek leert dat er waarschijnlijk honderden dragers van deze zogenaamde *foundermutatie* zijn. Dragerschap is enkel via DNA-diagnostiek vast te stellen.

Op het ECG van patiënten worden of tekenen van lange-QT syndroom of geleidingsstoosnissen gevonden, maar bijna nooit de kenmerken van allebei tegelijk. Dit wijst er op dat ook andere factoren de relatie tussen genotype en fenotype beïnvloeden. Momenteel loopt er in Maastricht, in samenwerking met de Universiteit Münster een studie naar *genetic modifiers*.

Hoewel het ECG bij aangedane patiënten met SCN5A delPhe1617 fors afwijkend kan zijn, gaat het bij de meeste mutatie dragers om subtiele kenmerken of zelfs normale ECG-bevindingen. Aanvullende cardiologische onderzoeken, te weten Holter, signal-averaged ECG, inspanningsECG en echocardiogram, zijn vaak geïndiceerd om betere karakterisering te kunnen doen. In bepaalde gevallen wordt een klasse-I-provocatietest en cardiale MRI gedaan.

HANDIGE LINKS

www.erfelijkheid.nl
[Website over erfelijke aandoeningen](#)

www.oncoline.nl
[Richtlijnen oncologische zorg](#)

[NHG-Standaarden](#)
[Thuisarts.nl](#)

OMIM
[Database genetische ziektes](#)

Orphanet
[Database zeldzame ziektes](#)

MEEST GELEZEN

- Stamboom tekenen
- Stamboom en symbolen
- Erfelijke aandoeningen van A tot Z

ZOEK IN PUBMED

 ZOEKEN

2. Link to Genetic online Continuing Professional Development (G-eCPD) module on oncogenetics, in Dutch: "PIN 15/01 Kanker en erfelijkheid"

Link: <http://leren.nhg.org/mod/resource/view.php?id=4460>


U bent ingelogd als E.J.F. Houwink | Wijzig account | Contact | Uitloggen

Digitale leer- en werkomgeving

nederlands huisartsen
genootschap
Home
Producten
Winkel
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Menu

- > Kanker en erfelijkheid ✓
- > Over dit programma ✓
- > 1. Pretoets ✓
- > 2. Mamma- en ovariumcarcinoom ✓
- > 3. Darmkanker ✓
- > 4. Andere vormen van erfelijke kanker (facultatief) ✓
- > 5. Communiceren over risico's en erfelijkheid ✓
- > 6. Ethische dilemma's ✓
- > 7. Uw handelen nu ✓
- > 8. Posttoets ✓
- > NHG-Producten, links en literatuur ✓
- > Evaluatie ✓
- > Certificaat ✓
- > Colofon ✓

Kanker en erfelijkheid

Wees alert op erfelijke kanker: het komt niet vaak voor, maar alle huisartsen krijgen er vroeg of laat mee te maken.

De internet-PIN in vogelvlucht

Na het doornemen van deze PIN:

- kunt u op basis van de anamnese patiënten met (mogelijk) een erfelijke aanleg voor kanker herkennen;
- kunt u voor een patiënte met borstkanker in de familie een stamboom tekenen en het risico berekenen;
- bent u op de hoogte van de verschillende soorten erfelijke darmkanker;
- weet u welke periodieke onderzoeken en risicoreducerende operaties mogelijk zijn voor mensen uit een familie met erfelijke kanker;
- weet u waar u wetenschappelijk onderbouwde informatie over erfelijkheid kunt vinden op internet.



[Volgende >](#)

Interessant

www.huisartsengenetica.nl
Website voor huisartsen met informatie over kanker en erfelijkheid

Stoet / VKGN

ERFELIJKE TUMOREN

RICHTLIJNEN VOOR

DIAGNOSTIEK EN PREVENTIE

2010

Vragen over deze PIN?

Voor vragen of opmerkingen van technische aard, mail naar pin@nhg.org.

Voor opmerkingen van inhoudelijke aard, mail naar implementatie@nhg.org.

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3. Nederlands Tijdschrift voor Geneeskunde, Weekboeken

Gepubliceerd op: 13-05-2011

Citeer dit artikel als:

Ned Tijdschr Geneesk. 2011;155:B705

Weekboek : <http://www.ntvg.nl/publicatie/Stille-engel>

Stille engel

Isa Houwink

Huisartsen moeten zich voorbereiden op snelle ontwikkelingen in de genetica van complexe ziekten. Collega's zeggen wel te weten dat diabetes, depressie, kanker en acute hartdood binnen families voorkomen, maar helaas vergeten ze vaak deze kennis toe te passen in de dagelijkse praktijk. Zeker 1 op de 10 patiënten komt met een klacht waarbij erfelijke belasting een rol speelt.

Nadat ik 3 jaar genetisch onderzoek in Amerikaanse laboratoria had gedaan, kwam ik in 2001 terug om mijn coschappen af te maken. In Californië werkte ik mee aan genetisch onderzoek naar een stukje erfelijkheidsmateriaal dat het syndroom van Rett veroorzaakt. Meisjes met dit syndroom worden na twee jaar autistisch, verliezen het vermogen om woordjes te zeggen, zoals 'mama'. Dat is toch het woord dat je moederhart doet smelten als je dat de eerste keer hoort. Ouders die ik zag, konden niet geloven dat hun kind dat opeens niet meer kon en zo'n zeldzame aandoening had. In het laboratorium gingen wij op zoek naar dat foutje dat ervoor zorgde dat hun kind een 'stil engeltje' werd. Ik werd direct gebiologeerd door het feit dat zo'n kleine afwijking zulke desastreuze gevolgen kan hebben. Van heinde en ver werden wij gevraagd het DNA te onderzoeken van kinderen die mogelijk het rettsyndroom hadden.

Genetica werd een levenslange verslaving, want mijn kennis en kunde blijft niet onbenut. Ik werd mij bewust van het feit dat er een brug geslagen moet worden tussen de laboratoriumkennis en de toepassing hiervan in de kliniek. Tijdens mijn opleiding huisartsgeneeskunde in Maastricht stapte ik af op prof.dr. Geert Jan Dinant (vakgroep Huisartsgeneeskunde Maastricht), die mij in contact bracht met prof.dr. Martina Cornel (sectie Community genetics, VUmc, Amsterdam). Samen werkten we verder aan een promotieproject om deze brug mogelijk te gaan maken. Het Center for Society and Genomics (CSG) was bereid om het project in te bedden in haar programma en te steunen met alle multidisciplinaire contacten. Experts uit verschillende geledingen, waaronder patiëntenverenigingen, onderwijskundigen, collega's uit de eerste en tweede lijn zijn onmisbaar voor een succesvolle en duurzame implementatie van het onderwijs. Huisartsen blijken behoefte te hebben aan onderwijs over genetica, maar dan wel gericht op de praktijk. Geen taaie stof, maar problematiek aansluitend bij de dagelijkse praktijk.

Zo is de patiënt van 42 jaar met darmklachten voor mij nu niet meer zomaar iemand die in de tropen iets verkeerd heeft gegeten, maar blijkt iemand die zich zorgen maakt omdat zijn vader met dezelfde klachten op dezelfde leeftijd darmkanker bleek te hebben. Dit laatste is het puzzelstukje dat ik nodig heb om de waarschijnlijkheidsdiagnose te stellen en na een coloscopie blijkt hij op 10 cm van het rectum (verder dan mijn wijsvinger kan reiken) een ingroeierende tumor te hebben. Gelukkig kan hij snel worden geopereerd en daarna nog volop van het leven genieten. Genetica is niet ver weg en ook niet ingewikkeld.

Gepubliceerd op: 20-05-2011

Citeer dit artikel als:

Ned Tijdschr Geneeskd. 2011;155:B706

Weekboek : <http://www.ntvg.nl/publicatie/Topsport>
Topsport

Isa Houwink

Willen wij als niet-genetici meegaan met de snelle ontwikkelingen in de genetica, dan is het noodzakelijk ons te laten onderwijzen om onze patiënten beter te kunnen informeren. Alleen op die manier zijn alle mogelijkheden en beperkingen van de genetica anno 2011 in dagelijkse praktijk te brengen. Voor succesvol geneticaonderwijs zijn experts en 'stakeholders' nodig, zoals huisartsen en andere eerste-lijnszorgmedewerkers die betrokken zijn bij onderwijs, klinisch genetici, vertegenwoordigers van patiëntenverenigingen, onderwijsontwikkelaars en beroepsverenigingen. Onderwijs ontwikkelen is niet topsport, je moet er voor kunnen samenwerken en gecoacht kunnen worden.

Dat samenwerken stimuleert om tot goede prestaties te komen, werd mij duidelijk tijdens het meetrainen in het Amerikaans nationaal roeiteam in 1998-2000. Het doel was immers een wedstrijd te winnen door samen te werken in een optimale sfeer waarbij de roeislag werd gevolgd. Je staat als roeier nooit alleen, je vult elkaar aan en staat voor elkaar klaar. Er zijn dagen dat je beter presteert dan andere, maar altijd ga je voor het beste.

Bij het voorbereiden van het geneticaonderwijs voor de eerste lijn heb ik precies hetzelfde gevoel. In de focusgroepen die wij hielden als voorbereiding, zaten niet alleen huisartsen of verloskundigen die hun behoeften over geneticaonderwijs uitten, maar ook experts die vanuit patiënten- of onderwijsperspectief vertelden wat zij belangrijk vonden. De focusgroepen waren geen afzonderlijke discussiegroepen, nee, zij vulden elkaar aan en completeerden elkaar. Het werd ons als onderzoekers en ontwikkelaars van onderwijs duidelijk dat er vanuit de verschillende geledingen behoefte is aan geneticaonderwijs en dat wij elkaar hierbij vooruit willen helpen. Nu moeten we nog in hetzelfde tempo leren roeien. Eerder onderzoek liet zien dat huisartsen te weinig kennis over genetica hebben. Dat is hetzelfde als te zeggen dat die ene roeier misschien wel competent is om mee te peddelen, maar dan geen opbouwende feedback te geven om er samen voor te gaan. Voor dat laatste blijkt veel meer nodig te zijn.

De sfeer van elkaar op een stimulerende wijze feedback geven om zo tot prestaties te komen, heb ik ook ervaren in de kadertraining van de Vereniging Nederlandse Vrouwelijke Artsen, die werd gecoacht door Ina Vader. Ons team van 12 vrouwelijk artsen deed aan 'topsport'. Gedurende 3 keer 2 dagen kwamen wij als team van vrouwen bijeen om te leren hoe elkaar feedback te geven en deze te krijgen, vergaderingen te leiden, te leren waar je grenzen liggen en deze duidelijk te maken. Mij werd feedback gegeven over mijn kwaliteiten en minder goede kanten. Ik hoorde dat ik daadkrachtig ben, maar dat ik daarin kan doorschieten en wat flexibeler zou moeten zijn. Niet helemaal onherkenbaar... Maar goed, als promotieonderzoeker en huisarts in een medisch centrum, ben ik blij met mijn kennis over mijn kwaliteiten, allergieën, uitdagingen en valkuilen. Zo heb ik handvaten gekregen hier beter mee om te gaan. Of, zoals Loesje zegt: 'Waarom moeilijk doen, als het samen kan?'

APPENDICES

Gepubliceerd op: 30-05-2011

Citeer dit artikel als:

Ned Tijdschr Geneeskd. 2011;155:B708

Weekboek : <http://www.ntvg.nl/publicatie/Een-passend-puzzelstukje>

Een passend puzzelstukje

Isa Houwink

Isa Houwink is huisarts en promotieonderzoeker bij de afdeling Klinische Genetica, sectie Community genetics VUmc, Amsterdam, en bij vakgroep Huisartsgeneeskunde in Maastricht. Ze schrijft 4 weekboeken over het belang van genetica voor niet-genetici (e.houwink@vumc.nl).

Sommige huisartsen worden steeds meer subspecialisten binnen hun vakgebied. Er worden kadertrainingen georganiseerd bijvoorbeeld op het gebied van hart- en vaatziekten, ouderenzorg en diabetes. Experthuisartsen of kaderartsen zagezegd. Of ik nu ook opper om een kadertraining 'Genetica' op te zetten? Nee, daar heb ik geen behoefte aan, want genetica is generalistisch. De gehele eerstelijnszorg zou zich bewust moeten zijn van het belang daarvan. Bij de veelvoorkomende multifactoriële, ook wel complexe ziekten is kennis over genetische aspecten belangrijk. Denk maar aan kanker, alzheimerdementie, diabetes mellitus type 2 of familiale hartproblemen. Genetica is relevant, omdat in de complexe etiologie soms één gen een grote rol speelt, en het soms de multifactoriële etiologie handvaten voor (be)handeling geeft. Al deze ziekten vallen binnen de bestaande kadertrainingen voor huisartsen; nóg een nieuwe kaderopleiding is dus overbodig. Met het voorbereiden van onderwijs over genetica voor huisartsen zouden we onze huisarts subspecialisten onderwijs kunnen bieden binnen hun kaderopleiding. Genetica zou daarmee echter een kennisdomein worden voor alleen die huisartsen die deze trainingen volgen op dat ene specifieke vakgebied, en ik denk niet dat dat de goede weg is. Genen zitten in alle organen en nieuwe kennis speelt in alle levensfasen, daarom kan kennis over genetica de gezondheidszorg voor iedereen verbeteren.

De huisarts in Amsterdam Zuid bekommert zich om sommige van zijn Joodse patiënten die binnen de gemeenschap met elkaar trouwen. De plattelandsdokter wordt door het boerengezin met een kind met downsyndroom gevraagd of het iets is waar de broer van vader van het kind zich ook zorgen over moet maken. Genetica komt heel dichtbij. Voor iedere arts, ook die zonder kadertraining. Daar is bewustwording voor nodig en onderwijs voor iedere arts.

Er wordt gezegd: hebben al die ontwikkelingen op het gebied van het genoom wel zin gehad? Heel veel, maar ook heel weinig. Met name op het gebied van zeldzame aandoeningen zie ik veel directe toepassingen voor de klinisch geneticus. Voor de huisarts zijn de puzzelstukjes bij complexe aandoeningen van belang voor het maken van de ingewikkelde puzzel voor gevorderden. Dat zijn namelijk de artsen die bij uitstek vaker met het bijltje hakken als het gaat om het diagnosticeren en behandelen van de complexe aandoeningen zoals diabetes of astma.

Een patiënte vertelt dat zij iets gek in haar borst voelt, en vertelt over haar angst voor kanker, wellicht iets engs in de familie waar eigenlijk nooit over gepraat mag worden, ja waarschijnlijk bij vader, maar ook bij een zus. In de familieanamnese vind ik als het ware een toverstokje: de puzzelstukjes vallen als een spannend verhaal naadloos in elkaar. In haar rechter borst voel ik een nauwelijks palpabele massa. Ik verwijst haar voor een mammografie en echo van de mammae. Helaas blijkt dat er sprake is van borstkanker, waarschijnlijk één van de erfelijke varianten. Patiënte is mij erkentelijk voor het feit dat ik snel heb gehandeld. Zo heeft de ontdekking van het genoom toch zin gehad!

Gepubliceerd op: 03-06-2011

Citeer dit artikel als:

Ned Tijdschr Geneeskd. 2011;155:B709

Weekboek : <http://www.ntvg.nl/publicatie/Perfectie-als-maatschappelijk-fenomeen>

Perfectie als maatschappelijk fenomeen

Isa Houwink

Isa Houwink is huisarts en promotieonderzoeker bij de afdeling Klinische Genetica, sectie Community genetics VUmc, Amsterdam, en bij vakgroep Huisartsgeneeskunde in Maastricht. Dit is haar laatste weekboek over het belang van genetica voor niet-genetici (e.houwink@vumc.nl).

Mijnheer Jansen (50 jaar) is al enige tijd niet fit. In zijn familie krijgen mensen kanker als ze rond de 50 jaar zijn. Hij komt mijn spreekkamer binnen en eist: 'Ik wil genetisch onderzoek!' Heeft hij ook een verhoogde kans op kanker of is alles goed? Mij bekruipt het gevoel dat wij als maatschappij bezig zijn een droom te verwezenlijken van perfectie, alsof we alles kunnen voorspellen. Een genetische test zou kunnen vertellen of alles goed is, zelfs blijft, en de angst voor imperfectie kunnen wegnemen.

Het Centre for Society and Genomics (CSG), waarbinnen ons project 'Onderwijs over genetica voor huisartsen wordt uitgevoerd, '...beschrijft, analyseert en verbetert de relatie tussen de samenleving en genomics-onderzoek. Daarmee draagt het CSG bij aan de aansluiting van genomics-onderzoek op de verwachtingen en vragen van de samenleving.' De samenleving heeft dus verwachtingen van de genetica. Het CSG sluit hierbij aan.


De huisarts houdt zich volop bezig met preventie. Onlangs lanceerden NHG en LHV het preventieconsult. Daarin is de rol van genetica helaas gering. Terwijl de maatschappij wel van ons als arts verwacht dat we de genetica betrekken bij het voorkómen van aandoeningen zoals acute hartdood, kanker en diabetes mellitus, liefst al in een presymptomatisch stadium. Uit onderzoek bleek dat 9 van de 10 ziekten waar Amerikanen het vaakst aan overlijden een genetische component hebben. Zaaïen wij dan angst en wekken wij verwachtingen die wij niet kunnen waarmaken? Via internet kan de individuele patiënt een DNA-test bestellen. Al voor enkele honderden dollars kun je je genoom laten screenen op allerlei aandoeningen. Maar wat doe je ermee, als patiënt en – niet onbelangrijk – als huisarts? Wat is de klinische bruikbaarheid als de patiënt daar met zijn formuliertje zit? Hoe nu verder?



De ogen zijn op mij gericht en ik wil de patiënt niet teleurstellen of nog erger: bang maken. Ik weeg mijn woorden, gelukkig heb ik even tijd. Genen hebben geen haast, geen ambulance die afkomt op een ziekmakend gen. Nee, ik kan een familieanamnese afnemen, vragen waarom de patiënt de test wil laten doen en wat de verwachtingen zijn, en zeker zo belangrijk: overleggen met een klinisch geneticus.

Als huisarts vormen wij de brug tussen de kliniek (ofwel het genomics-onderzoek) en de samenleving. Er wordt van ons verwacht deze brug te slaan. Kortgeleden zijn door Eurogentest competenties voor niet-klinisch genetici geformuleerd, waaruit blijkt waaraan huisartsen zouden moeten voldoen. Een uitdaging, maar zeker niet onmogelijk. Zoals ik eerder schreef in de vorige 3 weekboeken: genetica toepassen in de dagelijkse praktijk is niet moeilijk, als we genetica als puzzelstukje gaan zien bij het oplossen van de diagnostiek en behandelingen van de vaker voorkomende aandoeningen. Zo wordt genetica wellicht nog eens 'perfect' toegepast!

4. Reacties Medisch Contact

Link: <http://medischcontact.artsennet.nl/archief-6/tijdschriftartikel/18834/-schrik-maar-ook-begrip-1.htm>




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Schrik, maar ook begrip

Publicatie	Nr. 08 - 20 februari 2007
Jaargang	2007
Rubriek	Reacties
Auteur	I. Houwink en M.C. Cornel
Pagina's	349

In het artikel Schrik, maar ook begrip (MC 2/2007: 58), over de hieprikscreening bij pasgeborenen, geven Pennings c.s. kritiek op de gebrekkige kennis van huisartsen, en daarmee de gebrekkige informatievoorziening aan ouders, over het MCADD-onderzoek (medium-chain-acyl-CoA-dehydrogenase deficiëntie). Dit geeft ouders een gevoel van onzekerheid. Pre- en postnatale zorg behoren toe aan de huisarts en dus moet ook de pas geïntroduceerde hieprikmethode tot het kennispakket van de huisarts behoren.

Wat is de oorzaak van dit gevonden gebrek? Misschien moet de huisarts op te veel verschillende gebieden zijn kennis bijhouden? Is het wellicht de gebrekkige informatievoorziening in de vorm van goede nascholing voor huisartsen? Is de keuze in nascholingsprogrammas te vrijblijvend? Of is het misschien de tendens dat de pre- en postnatale screening in de eerste lijn enkel richting de verloskundige dreigt te gaan? Is het misschien wel beter zo?

Wij zijn van mening dat de huisarts mee moet gaan met deze tijd en we nemen de kritiek dus zeer serieus. Genetica is geen moeilijk vak, maar wel een vak dat praktisch en toepasbaar moet zijn voor de huisarts. Helaas is er tot nu toe weinig steun voor onderzoek naar genetica in de huisartsenpraktijk en naar de vraag hoe huisartsen zo efficiënt mogelijk zijn na te scholen op dit vlak. Terwijl het huisartsenvak preventie hoog in het vaandel heeft staan, en genomiconderzoekers bezig zijn de genetische basis van ongeveer alle frequent voorkomende ziekten bloot te leggen.

Wij pleiten voor meer scholing van huisartsen op dit gebied en onderzoek ter evaluatie van de effecten van scholing, zodat de patiënt laagdrempelig naar de huisarts blijft gaan.

Maastricht, Januari 2007
I. Houwink, huisarts i.o. en bestuurslid NACG (Nederlandse Associatie voor Community Genetics)
M.C. Cornel, hoogleraar community genetics VUmc en voorzitter bestuur NACG

Afdrukken
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Er zijn nog geen reacties bij dit bericht. Ziet u geen reactieformulier? Log in.

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Genetica - E.J.F. Houwink en M.C. Cornel

Publicatie	Nr. 50 - 16 december 2010
Jaargang	2010
Rubriek	Media & Cultuur
Auteur	Sophie Broersen
Pagina's	2715

Opfrissertje voor de genetikakennis

Huisarts Isa Houwink en geneticus Martina Cornel schreven het boekje Genetica, uit de reeks 'Practicum huisartsgeneeskunde, een serie voor opleiding en nascholing'. Het dunne boekje is in een avond doorgewerkt en na afloop is – als het goed is – de basale genetikakennis van de huisarts weer opgefrist. Van hoe je ook alweer een stamboom tekent en wie eerstegraads familieleden zijn tot hoe overerving werkt volgens Mendel, of juist niet helemaal volgens Mendel.

Maar ook onderwerpen als een afwijkende uitslag bij de hiepriek, preconceptieozorg en erfelijke vormen van kanker komen aan bod. Aan de hand van realistische casuïstiek en vragen die in de praktijk worden gesteld, voeren Houwink en Cornel de lezer langs relevante onderwerpen. En dat in zeer leesbare bewoordingen en met handige verwijzingen naar verschillende websites.

De huisarts zal geen eerstelijns geneticus zijn na lezing, en de aanbeveling om te overleggen met of te verwijzen naar de klinisch geneticus komt vaak terug. Voor de huisarts die toch al geïnteresseerd was in de genetica, zal het soms wat té basaal zijn. Maar om gewoon wat beter beslagen ten ijs te komen, is het boekje een aanrader. SB

E.J.F. Houwink en M.C. Cornel, Genetica, in de reeks 'Practicum huisartsgeneeskunde, een serie voor opleiding en nascholing', Elsevier Gezondheidszorg, 91 blz., 25,75 euro.

• **Rubriek Scoop**

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