Introduction
During recent decades, rapid developments have increased the relevance of genetics in medicine, and the discovery of more and more genes involved in disease aetiology will increase the number of genetic tests that are available in daily clinical practice.\textsuperscript{1-3} Disease definitions will be redefined by genomics, as new predisposing risk factors become apparent. Tests may be used for the prediction, diagnosis and optimisation of treatment for most common diseases.\textsuperscript{4} The introduction of these tests will result in an increase in the number of people who have the opportunity to make use of these new possibilities and wish to do so. Consequently, for all genetic and non-genetic health care providers this will result in an increasing demand for genetic testing. Moreover, non-geneticist physicians will often be the first in line to be confronted with questions from patients about genetics, and they will become important providers of genetic care in the future.\textsuperscript{5} As non-geneticists will play a major role in ordering these tests, interpreting the results, referring patients to genetic centres and communicating with patients about genetic issues, their knowledge of genetics will become an essential element of good clinical practice. The minimum requirement is, therefore, adequate knowledge of genetics and the genetic tests that are available.

**Genetic knowledge among non-genetic health care providers**

Although these recent rapid developments in the field of genetics make it important that health care providers are adequately educated in genetics and keep up to date with these developments, it is not clear what health care professionals learn about genetics during their basic, specialised and continued medical training, or whether their knowledge is, indeed, adequate for daily clinical practice. Several studies have shown that many non-genetic health care providers lack the necessary knowledge and skills to react appropriately to the increasing demands from patients.\textsuperscript{6-10} For example, Hunter et al.\textsuperscript{7} found that, although a majority of Canadian physicians (family physicians, obstetricians, paediatricians and internists) considered their knowledge of genetics to be adequate, only a minority were confident in their ability to provide genetic counselling for simple scenarios. According to Watson et al.\textsuperscript{8}, general practitioners in Great Britain currently feel that they lack the relevant knowledge and skills to manage patients who are concerned about their family history.

**Unfavourable effects of the lack of genetic knowledge**

Genetic disorders and congenital anomalies are a major cause of mortality and morbidity. Most couples want to know if they are at increased risk of having a child with a congenital disorder.\textsuperscript{11,12} Identifying these couples and providing them with information about their risk could help them make an informed reproductive choice. From this point of view, it is important to investigate how well general practitioners (GPs) perform their tasks with regard to identifying and informing couples who are at
increased risk of having a child with a congenital disorder. In 1989, De Smit et al.\textsuperscript{13} found that many pregnant women in the Netherlands, were not informed about their risk of giving birth to a child with a congenital disorder before they consulted a midwife or a gynaecologist, which is usually too late. Aalfs et al.\textsuperscript{14} found that, despite the fact that most GPs were in favour of genetic counselling before pregnancy instead of during pregnancy, a considerable number of women were already pregnant before they visited the genetic centre for the first time. In that study, one of the main reasons for referring a patient during pregnancy, instead of before pregnancy, was the GP’s unawareness of potential risk factors. In addition, Sikkens et al.\textsuperscript{15} reported that only a small number of the parents of children/foetus in a register of congenital anomalies had visited a genetic centre for counselling. Lack of knowledge about cystic fibrosis (CF) carrier testing has already been found to be a problem; people with a family history of CF do not always receive the correct information from their physicians, and children with CF have been born, whereas their parents would have opted for testing and prenatal diagnosis if it had been offered.\textsuperscript{16} Moreover, a study among schoolchildren with learning disability showed that basic genetic investigation in order to find the cause of their disability was infrequent.\textsuperscript{17}

Physicians also appear to have insufficient knowledge about diseases which generally occur in adulthood. Yong et al.\textsuperscript{18} reported that only a limited number of physicians were aware of the importance of paternal family history in determining the risk for hereditary breast cancer. Wideroff et al.\textsuperscript{19} found that very few primary care physicians used genetic testing to assess cancer susceptibility (ordered a test themselves or referred patients for testing).

\textit{Causes of the lack of genetic knowledge}

According to Hofman et al.\textsuperscript{6} despite the increase in knowledge of genetics among more recently graduated physicians in the USA, deficiencies still remain. One possible cause of this reported lack of knowledge and skills is lack of attention of genetics in the undergraduate or postgraduate medical curriculum. An additional cause of deficiency in knowledge of genetics could be that GPs and other specialists can choose whether or not they wish to include genetics in their continuing medical education. No specific subjects are obligatory and therefore only physicians with an interest in genetics are likely to attend the genetic courses.

\textit{Opinions of non-genetic health care providers concerning genetic screening and testing}

Due to the recent discovery of an increasing number of genes and disease-causing mutations, screening programmes may become available to identify couples who are at
risk of having a child with a congenital disorder. Prenatal carrier screening leaves only a limited number of reproductive options open for carrier couples, compared to preconceptional screening, and may impose time-constraints when decisions about a prenatal diagnosis have to be made. Screening couples who are planning a pregnancy would therefore make it possible to identify, for example, carrier couples, and to inform these couples about their risk and about the reproductive options that are available. An important barrier for the implementation of a genetic screening programme might be the negative opinion of physicians. In a study carried out by Suther and Goodson, almost half of the respondents said that they were not likely to order carrier testing or to order a predictive test to assess a disease risk (55.5% and 64.5%, respectively). If primary care providers are to offer carrier testing and predictive tests it is important to know about their beliefs with regard to genetic testing and their self-perceived skills.

**Professional competence**

In order to answer the question “What do physicians need to be able to do for effective practice”, the Royal College of Physicians and Surgeons of Canada (RCPSC) has produced a competency-based framework that describes the principal generic abilities of physicians oriented to optimal health and health care outcomes. In this document seven areas of competence are highlighted: medical expertise (including medical knowledge, clinical skills and professional attitude), communication skills (including doctor-patient communication and communication with other professionals and individuals), collaborative work (health care teamwork), management (individual, as member of a team, and as a participant in the health care system), health advocacy (to advance the health and well-being of individual patients, communities and populations), scholarship (ongoing learning and improvement) and professionalism (ethical practice, profession-led regulation, and high personal standards of behaviour). These areas of competence appear to share many aspects, and are not independent of each other. In the rapidly developing field of genetics the most important areas are medical expertise, communication skills, scholarship, and professionalism. For practical reasons, only a limited number of areas of competence that are relevant for genetics could be included in our investigations.

**Outline of the thesis:**

The main aim of this study was to investigate the genetic knowledge, and the beliefs towards genetic screening and testing, and the self-perceived genetic skills among non-genetic health care providers in the Netherlands. The following research questions were addressed:
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Q1  What is the existing policy and practice in the Netherlands with regard to genetics in undergraduate, post-graduate and continuing medical education?

Q2  Is the knowledge of genetics and awareness of genetic tests among GPs, gynaecologists (GYNs) and paediatricians (PEDs) adequate for the possible provision of genetic tests?

Q3  What are the beliefs of GPs, GYNs and PEDs towards genetic testing?

Q4  Are GPs, GYNs and PEDs willing to offer new genetic tests to their patients?

Q5  Are GPs, GYNs and PEDs prepared to deal with different genetic tests?

Q6  What are the opinions of physicians with regard to preconceptional genetic carrier screening, and which factors are associated with a positive opinion?

Q7  Have the attitudes of GPs and their activities concerning genetic counselling changed over a 10-year period (1989-1999)?

Q8  Do medical students nearing graduation have the required level of genetic knowledge that is relevant for daily practice?

Chapter 2 describes the current (2002) policy and practice in the Netherlands in with regard to genetics in undergraduate, post-graduate and continuing medical education, but excludes the training of individuals specialising in clinical or laboratory genetics and genetic associates (Q1). Chapters 3, 4 and 5 present the results of a questionnaire survey among GPs, GYNs and PEDs. Chapter 3 describes the knowledge of genetics and awareness of genetic tests among physicians, and factors associated with a higher level of knowledge (Q2). Chapter 4 describes the beliefs and perceived skills of GPs, GYNs and PEDs with regard to genetic tests (Q3, Q4, Q5), and chapter 5 describes the opinions of GPs, GYNs and PEDs with regard to preconceptional CF carrier screening and variables associated with their opinions (Q6). Chapter 6 presents the result of a questionnaire survey among GPs, the aim of which was to investigate whether the activities and the attitudes of GPs concerning genetic counselling changed between 1989 and 1999 (Q7). Chapter 7 presents the results of a study among medical students nearing graduation, to investigate whether they had the required level of genetic knowledge that is relevant for daily practice (Q8). Chapter 8 starts with a critical evaluation of the outcome of the studies focusing on genetic knowledge and the opinions on genetic testing among non-genetic health care providers. This is followed by some recommendations for further research and the overall conclusions. Chapter 9 includes a summary of the studies presented in this thesis, a summary of the evaluation of the medical curriculum and the recommended changes, and some suggestions for future research projects.
References


