Summary
Genetic knowledge, opinions and self-perceived competence of non-genetic health care providers

During recent decades, rapid developments have changed the relevance of genetics in medicine. Consequently, all genetic and non-genetic health care providers will be faced with an increase in the demand for genetic testing. In the future, non-geneticists will most likely play a major role in ordering and interpreting genetic tests, referring patients to genetic centres, and communicating with patients about genetic issues. Therefore, their knowledge of genetics will become an essential part of good clinical practice. Medical professional competence has been defined in seven areas of competence: medical expertise, communication skills, collaborative work, management, health advocacy, scholarship and professionalism. In this thesis several aspects of medical expertise, communication skills, health advocacy, and professionalism have been investigated. The main focus is on genetic-related knowledge, beliefs with regard to genetic testing, and self-perceived competence among non-genetic health care providers, i.e. general practitioners, gynaecologists and paediatricians.

The existing genetics policy and practice in the Netherlands in relation to undergraduate, post-graduate and continuing medical education

The 2002 curricula of the eight medical schools providing medical education and the curricula of all of all types of specialised medical training programmes were examined, to investigate whether medical health care providers in the Netherlands are adequately educated in genetics. In Chapter 2 the collection of data on the current level of genetic education of non-genetic health-care providers and the evaluation of these data are described in detail. In most medical schools very few hours are allocated to genetic education. This subject is relatively invisible, being integrated only in a minority of courses. If present at all, it comprises only a small fraction of the total course (a mean of 8%). Only three of the 27 training programmes for medical specialists and for MDs caring for mentally handicapped people included a formal genetic education programme. Continued genetic education courses on genetics are seldom offered. However, training in midwifery includes at least 3 weeks of genetic education, and courses on genetics are offered frequently to practicing midwives. In conclusion, we found that there was a lack of visibility of genetics in the current medical education, and there appear to be no general, nationally defined final goals concerning genetic education for non-genetic health care providers in the Netherlands.
The knowledge of genetics among general practitioners, gynaecologists and paediatricians, their beliefs with regard to genetic testing and their self-perceived competence

Chapters 3, 4 and 5 present the results of a questionnaire study among general practitioners (GPs), gynaecologists (GYNs) and paediatricians (PEDs) in the Netherlands. A random sample of 200 GPs, 300 GYNs and 200 PEDs received a questionnaire, and all 65 PEDs who were members of the Section on Congenital and Inherited Disorders (SCID) of the Paediatric Association of the Netherlands (SCID) also received a questionnaire. In addition, all registered clinical geneticists (CGs, n=58) received the knowledge section of the questionnaire for validation purposes (Chapter 3). Of the 200 GPs, 300 GYNs and 265 PEDs, physicians who were no longer working were excluded, as were GYNs who were no longer working in the field of obstetrics. The response rate was 64% for GPs (124/195), 69% for GYNs (198/285), and 72% for PEDs (177/247). No differences were found between the two groups of PEDs (SCID members vs. non-members), so they were combined for further analysis.

Only questionnaires from respondents who had answered more than 75% of all the questions were included in the analysis (Chapters 4 and 5). Further analyses were therefore based on the remaining 124 GPs, 197 GYNs and 176 PEDs.

The results for the knowledge section of the questionnaire (Chapter 3) included only those respondents who answered all the knowledge questions. Further analyses described in Chapter 3 were therefore restricted to the answers from 122 GPs, 187 GYNs and 164 PEDs. In the validation group, 90% of CGs (52/58) returned the questionnaire.

Chapter 3 presents results regarding genetic knowledge and awareness of genetic tests among the physicians, as well as the factors influencing their knowledge and awareness.

The knowledge scores of the GPs (mean 64% correct answers, 61-66% [95% CI]), GYNs (mean 75% correct answers, 73-76% [95% CI]) and PEDs (mean 81% correct answers, 79-82% [95% CI]) were lower than those of the CGs in the validation group (mean 95% correct answers, 94-96% [95% CI]). The variation in individual scores was highest among the GPs; the 5th percentile of GPs, GYNs and PEDs, respectively, was approximately 40%, 52% and 62% correct answers and the 95th percentile of GPs, GYNs and PEDs, respectively, was approximately 85%, 90% and 92% correct answers.

There was a specific lack of knowledge about DNA testing. For example, 74% of the GPs, 50% of the GYNs and 46% of the PEDs were unaware of the availability of DNA tests or biochemical tests for sickle-cell anaemia. Apart from specialty, important factors that were positively associated with the knowledge scores of non-geneticists were: more recent graduation from medical school, having taken an elective course in genetics, and providing genetic counselling in their own practice. In
conclusion, there are clear deficiencies in the overall genetic knowledge of many non-genetic health care providers.

Chapter 4 describes the results of that part of a questionnaire study, among GPs, GYNs and PEDs, which focused on their beliefs with regard to genetic testing and their self-perceived competence.

The innovativeness of the physicians (likelihood that they would offer new genetic tests) was measured. In total, 35% of GPs, 44% of GYNs and 54% of PEDs were (very) likely to routinely offer a predictive test for treatable common disorders, even when others in their specialty would not do so. If it was common practice, 94%, 96% and 96%, respectively, would do so.

Approximately 20% (17–27%) of the physicians thought that the use of prenatal testing by parents for hereditary breast cancer was appropriate, and between one third and one half (34–47%) thought that this was also appropriate for sickle cell anaemia. Almost all physicians (83–97%) thought that testing was appropriate for Duchenne muscular dystrophy and for cystic fibrosis.

The majority of physicians (67-92%) felt (moderately) confident that they could cope with all (future) test-related situations. Of the physicians, 53% would personally counsel a couple about prenatal diagnosis before referring them to a clinical genetic centre. However, 19% would express their own personal opinion about prenatal diagnosis during a consultation and 7% would express their own opinion about pregnancy termination. Approximately 10% of the physicians (8-11%) would disclose information about a predictive test for Huntington disease without permission to the patient’s children.

In conclusion, as many as 46-65% of these physicians had reservations about offering genetic testing if it was not standard practice. Overall, the PEDs were the most willing to offering genetic testing, and the GPs were the most reluctant. The beliefs of the physicians with regard to the use of prenatal testing by parents for different diseases varied widely. However, all the answers concerning the appropriateness of the use of prenatal diagnosis by parents ranged in the same way from high (Duchenne muscular dystrophy) to low acceptance (Alzheimer disease). Most physicians perceived their competence as adequate. However, in spite of their own perception of adequate competence, more training is important because some “gold standards” for professional clinical genetics, such as non-directiveness, are not adhered to by all GPs, GYNs and PEDs. Special attention should be paid to directive method of counselling (specifically for prenatal diagnosis and pregnancy termination) and the disclosure of information to third parties.

An important barrier for the implementation of a preconceptional genetic screening programme might be the negative opinion of physicians with regard to genetic carrier
screening. In Chapter 5 the knowledge and the opinions of GPs, GYNs and PEDs with regard to preconceptional CF carrier screening, and the possible factors that are associated with their opinions are described. In total, 63% of the GPs, 69% of the GYNs and 72% of the PEDs supported preconceptional CF carrier testing if a couple requested a test. Of the GPs, GYNs and PEDs, 16%, 19% and 25%, respectively, answered that they were in favour of actively offering a test with 95% test-sensitivity to all couples who were planning a pregnancy. Therefore, physicians are sympathetic towards preconceptional CF carrier screening if the couples themselves request a test. Physicians had reservations about routinely offering a CF carrier test. A positive opinion with regard to preconceptional CF carrier screening was associated with the following variables: “considering the test-sensitivity as less important” (GPs, GYNs), “high perceived risk of having a child with CF” (GYNs), “providing genetic counselling in their own practice” (PEDs) and “reassurance when both partners test negative” (PEDs).

**The change in activities and the attitudes of general practitioners concerning genetic counselling over a 10 year period (1989-1999)**

Chapter 6 describes the results of a questionnaire survey among 98 GPs, the aim of which was to investigate whether the activities and the attitudes of GPs towards genetic counselling have changed between 1989 and 1999. In 1989 a random sample of 124 GPs in a province of the Netherlands (Noord-Holland) received a questionnaire about this topic. Of these GPs, 98 were contacted again in 1999, and 71 completed the original questionnaire again (72% response rate). Comparison of the answers in 1999 to those in 1989 showed an increase in the percentage of GPs who provided genetic counseling when there was a definite risk factor for having a child with a congenital disorder, such as a previous child with a congenital disorder. In both 1989 and 1999 the GPs seldom followed a recommended combination of oral and written information, such as an appointment in combination with a brochure. Only data that was available in the databases on the risk indicator ‘use of medication’ increased over the years. In 1999 the GPs were still supporters of a directive method of counselling, and seemed to believe that the main goal of genetic counselling was to prevent hereditary and congenital disorders. Although an increased number of GPs provided genetic counselling when there was a definite risk indicator or referred to a clinical geneticist, only limited improvement was found in the attempts made by GPs to collect such data on risk indicators.
The level of genetic knowledge that is relevant for daily practice among medical students nearing graduation

To investigate whether the level of genetic knowledge that is relevant for daily practice among medical students nearing graduation was sufficient to enable them to react appropriately to the change in relevance of genetics in medicine, a computer examination was developed and validated in a group of clinical geneticists, medical students nearing graduation and non-medical students. A detailed description can be found in Chapter 7. The examination consisted of 215 genetic questions, classified into three categories of relevance: “essential” knowledge, “desirable” knowledge and “too specialised” knowledge. To set an independent standard, the questions were also judged by clinical geneticists and non-genetic health care providers in an Angoff procedure. A total of 291 medical students nearing graduation, from seven out of the eight medical medical schools in the Netherlands, participated. As expected, the mean score for “essential” knowledge (71.63%, 70.74–72.52 [95% CI]) was higher than for “desirable” knowledge (55.99%, 55.08–56.90 [95% CI]), and the mean score for “too specialised” knowledge (44.40%, 43.19–45.62 [95% CI]) was the lowest. According to passing scores set for “essential” knowledge, as defined by the designers, the clinical geneticists and the non-genetic health care providers, only, 0%, 26% and 3%, respectively, of the participants would have passed. These results suggest that medical students nearing graduation lack the level of genetic knowledge that is essential for daily practice. Changes should be made in the medical curricula in order to improve genetic knowledge among medical students, to enable them to react appropriately to the change in relevance of genetics in medicine.

Concluding remarks

The results of the studies presented in this thesis have provided better insight into the level of genetic knowledge and beliefs with regard to genetic testing among non-genetic health care providers. In general, GPs, GYNs and PEDs are willing to counsel a patient about genetic testing, would offer a predictive test if it is common practice, and perceive their competence as adequate. There seem to be few thresholds in the implementation of genetic testing by non-genetic health care providers. However, it has been demonstrated that many (future) non-genetic health care providers lack the necessary knowledge and counselling skills to react appropriately to the increasing demands of patients concerning genetic testing. In order to optimise the benefits of genetic testing for patients now and in the future, in medical education attention should be paid to the lack of genetic knowledge, the tendency towards counselling directly and (although in a minority) the intention to disclose genetic information to third parties.
These results indicate that the development of education programmes for non-genetic healthcare providers and changes in the medical curricula are needed in order to improve the provision of genetic-related health care for patients.