General Discussion
Discussion and Conclusions

This thesis focuses on the knowledge of genetics, the opinions on genetic testing and the self-perceived skills among non-genetic health care providers such as general practitioners (GPs), gynaecologists (GYNs) and paediatricians (PEDs). Medical professional competence has been defined in seven areas of competence: medical expertise, communication skills, collaborative work, management, health advocacy, scholarship and professionalism, several areas of which have been addressed in the previous chapters. This chapter starts with a short recapitulation of the background and objectives of our studies. Subsequently, the findings will be discussed in relation to the research questions, and compared with the results of other studies. The chapter concludes with future perspectives of genetic counselling and testing provided by non-genetic health care providers, including recommendations for changes in the medical curricula and further research.

Background and Objectives

During recent decades, rapid developments have increased the relevance of genetics in medicine. Consequently, for all genetic and non-genetic health care providers there will be an increase in the demand for genetic testing. They will more often be the first in line to be confronted with questions related to genetics and genetic testing. In the Netherlands, like in many other (European) countries, general practitioners (GPs) act as “gatekeepers”, regulating the entry of all individuals into the rest of the medical system. In order to provide each individual with adequate genetics-related care, the genetics-related medical competence of GPs is of particular interest. As has been found in many studies, a considerable number of parents who are at risk of having a child with a congenital disorder are not adequately informed or referred timely for genetic counselling. For that reason, the genetics-related medical competence of gynaecologists (GYNs) and paediatricians (PEDs) is therefore also of interest. In addition, many people who are, themselves, at risk of developing a (future) disorder with a genetic component are unaware of this potential risk, and remain uninformed or inadequately informed by their GP or specialist. In order to explore the knowledge of genetics, self-perceived genetic skills and the opinions on genetic screening and testing among non-genetic health care providers in the Netherlands eight research questions were addressed.
The eight research questions addressed in this thesis

**Question 1: What is the existing policy and practice in the Netherlands with regard to genetics in undergraduate, post-graduate and continuing medical education?**

In order to answer this question the curricula of the eight Dutch medical schools providing medical education and the curricula of all types of specialised medical training programmes were examined for the year 2002. In the Netherlands, medical undergraduate education takes a minimum of six years of theoretical and practical examinations. Although the curricula vary, the medical schools share a general framework of final goals. At most medical schools, the number of hours spent on genetic education is minimal, and genetics is relatively obscure, because it is integrated in several courses, comprising only a small proportion of the total course (a mean of 8%) (Chapter 2).

Only three of the medical specialist training programmes (other than clinical genetics), namely obstetrics and gynaecology, neurology, and paediatrics, and also the training of MDs for mentally handicapped people included a formal genetic education programme. Several of the general practice institutions at the eight medical schools allocated some time for genetic education in their GP training programme, but there are apparently no formal, national regulations. For non-genetic health care professionals there are no continuing education courses other than occasional optional courses, and there is little organized continuing medical education to encourage them to keep up to date with developments in genetics (Chapter 2).

In the Netherlands there appear to be no general, nationally defined final goals regarding genetic education for non-genetic health-care professionals, and without such goals it is hard to provide comparable education in different institutions. More specifically, this lack of visibility of genetics may lead students to overlook its importance (Chapter 2). The findings presented in this thesis are generally in line with the findings of studies carried out in other European countries, where, also, many different organisations are responsible for the regulation, assessment and provision of medical education. Therefore, changing these curricula may become a challenge for the future.

**Question 2: Is the knowledge of genetics and awareness of genetic tests among general practitioners (GPs), gynaecologists (GYNs) and paediatricians (PEDs) adequate for the possible provision of genetic tests?**

There are clear deficiencies in the overall knowledge of genetics among many non-genetic health care providers (Chapter 3), and there is a specific lack of knowledge about DNA testing. The mean knowledge scores of the GPs were lower than those of
the GYNs, which were lower than those of the PEDs. Furthermore, the variation in individual scores was highest among the GPs and lowest among the PEDs. Apart from specialty, important factors that are positively associated with the knowledge scores of non-geneticists are: more recently graduated, having completed an elective course in genetics, and providing genetic counselling in their own practice (Chapter 3). The deficiencies in knowledge of genetics that were found in this study among a large percentage of physicians in the Netherlands are in line with the findings in other countries.\textsuperscript{10-14} A possible cause of deficiency in genetic knowledge could be that GPs and 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. In addition, there is very little organised continuing medical education to encourage them to keep up to date with developments in genetics (Chapter 2). These deficiencies in genetic knowledge have been recognized by physicians themselves in several studies, and they have expressed a need for more genetic education.\textsuperscript{14-18}

**Question 3: What are the beliefs of general practitioners (GPs), gynaecologists (GYNs) and paediatricians (PEDs) towards genetic testing?**

It is well known that clinical geneticists are trying to provide information without issuing a directive.\textsuperscript{19} In contrast with this attitude, the GPs in our study were more supportive of a directive method of counselling (Chapters 4 and 6). Nineteen percent of the respondents in our study would make their own opinion about prenatal diagnosis known to their patients. This is still much a lower percentage than that found in a study in the USA, in which 44% of the respondents indicated that they would make their opinion known.\textsuperscript{20} As in the USA, GYNs were more reluctant than GPs to give an opinion (Chapter 4).\textsuperscript{20}

**Question 4: Are general practitioners (GPs), gynaecologists (GYNs) and paediatricians (PEDs) willing to offer new genetic tests to their patients?**

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 no others in their specialty would do so. This is in line with findings in the USA, where 51% of the physicians answered that they would do so.\textsuperscript{10} In our study, once common practice 94%, 96% and 96%, respectively would do so (Chapter 4). More than half of the respondents in our study (53%) would counsel before referral to a genetic centre (Chapter 4). This is in line with the findings of a study in the USA, where 49% of the respondents said that they would counsel patients about prenatal diagnosis.\textsuperscript{20} Several studies have reported that primary care providers are willing to incorporate more genetics in their practice in future \textsuperscript{21}, but they also expressed a need for more genetic education.\textsuperscript{14-15 17-18 22} The respondents in several studies would more readily adopt genetic testing if the patients expressed an interest in testing (Chapter 5).\textsuperscript{23}
**Question 5: Are general practitioners (GPs), gynaecologists (GYNs) and paediatricians (PEDs) prepared to deal with different genetic tests?**

Most of the physicians in our study felt (moderately) confident about their ability to cope in all genetic test-related situations mentioned in this study. This is not surprising, because a number of these abilities apply to every day health care practice (Chapter 4). Very few physicians would disclose the results of tests for Huntington disease to non-family members (0-3%) (Chapter 4). This is in line with findings in the USA, where 1 – 5% of the respondents stated that they were willing to disclose this information to non-family members. However, a remarkable percentage (2-11%) of the physicians in our study said that they would disclose information about this predictive genetic test to family members, and especially to the patient’s children, without permission from the patient (Chapter 4).

As almost all the physicians said that they would inform a patient and ask for consent before ordering a predictive test, and that they would report both favourable and unfavourable results to their patients, no problems should be expected on these aspects of providing genetic tests by these non-genetic health care providers (Chapter 4). The question that remains is whether the competence of the physicians is adequate for them to act in all kind of genetic test-related situations, because of their tendency towards directive counselling and (even in a minority) the disclosure of genetic information to third parties (Chapters 4 and 6).

**Question 6: What are the opinions of physicians with regard to preconceptional genetic carrier screening, and which factors are associated with a positive opinion?**

In our study, as well as in other studies, there is more support from the physicians if couples who are planning to have children request a cystic fibrosis (CF) carrier test, compared to actively offering screening (Chapter 5). The opinions about preconceptional CF carrier screening are apparently influenced by the way in which the test is offered (actively/passively) and the characteristics of the test (test-sensitivity) (Chapter 5). A disadvantage of such a passive approach is that people with a higher level of education might have easier access to testing than others. Despite the introduction of routine CF carrier screening in some states of the USA, research in Maryland, Virginia and Washington DC has shown that none of the family medicine physicians, and only 8% of the internal medicine physicians, 19% of the obstetricians and 13% of the paediatricians stated that they had ever ordered a CF carrier test or referred a patient for testing. Although, in a study carried out by Loader et al. the primary care providers (obstetricians-gynaecologists) found it inconvenient to offer CF carrier screening to non-pregnant women, at the end of the trial the participating providers had a significantly more favourable attitude towards screening for all women of reproductive age than at the beginning of the trial.
**Question 7: Have the attitudes of GPs and their activities concerning genetic counselling changed over a 10-year period (1989-1999)?**

GPs' attitudes towards genetic counselling did not change between 1989 and 1999. During this period there was limited improvement in their attempt to collect data on risk factors for having a child with a congenital disorder (Chapter 6). It has been suggested by Watson et al. that GPs register only a limited family history, and that it is often out of date and not adequate enough to assess genetic risks. Nevertheless, Rose et al. found that family history-taking and genetic counselling was feasible in primary care. According to Aalfs et al., limited alertness and awareness among GPs about genetic risk factors in their patients played a major role in the less appropriate timing of referral of patients for genetic counselling (during pregnancy instead of before pregnancy). Therefore, the implementation of routine family history-taking in general practice is strongly recommended.

**Question 8: Do medical students nearing graduation have the required level of genetic knowledge that is relevant for daily practice?**

Among medical students nearing graduation there are clear deficiencies in the level of genetic knowledge that is relevant for daily practice. Their knowledge is also too specialised (Chapter 7). Only a very small minority of the students in our study chose to take either an elective course (3%) or an elective clerkship in genetics (1%) (Chapter 7). Furthermore, as demonstrated by Challen et al., the integration and visibility of genetics in the medical curricula in several countries in Europe is very limited.

**Limitations**

Several limitations of the studies presented in this thesis should be taken into account. First of all, most of the data that are presented were collected by means of a questionnaire. Information about opinions and competence related to the possible provision of genetic tests may therefore differ from actual behaviour and opinions. However, these opinions and this behaviour were measured in large samples of physicians and with sufficient response rates, and therefore their answers should reflect their intended behaviour. In some countries the GPs do not have the role of “gatekeeper”, due to differences in the health care systems, and therefore the results of this study should be extrapolated with caution to other countries. In the meantime, the educational system in the Netherlands has been changed to the Bachelors/Masters structure. Therefore, there could have been a change in the visibility of genetics in the medical curricula. Furthermore, little is known about the effectiveness of various interventions recommended in this thesis, such as changes in the medical curriculum,
so the interventions may therefore not turn out to be effective. Careful effect evaluation is a prerequisite.

Overall Conclusions

In the future non-geneticists will play a major role in ordering and interpreting the results of genetic tests, referring patients to genetic centres, and communicating with patients about genetic issues, so their knowledge of genetics will become an essential aspect of good clinical practice. At present, however, the knowledge of genetics among physicians is insufficient to fulfil this role (Chapters 3 and 7). In addition, although the physicians perceive their skills as adequate, concerns exist about their support for a more directive way of counselling than which is recommended for genetic counselling (Chapters 4 and 6). Education programmes for non-genetic health care providers should therefore not only focus on their level of genetic knowledge, but integrate their skills as well. Special attention should be paid to a non-directive way of counselling, and also to the fact that the disclosure of information about a predictive genetic test to family members, especially the patient’s children, without the permission form the patient is undesirable (Chapters 4 and 6). Because of the positive opinions of physicians towards genetic tests for patients who request these themselves, one could argue that education programmes for the public could help to reach all people who are interested in genetic testing.

Recommendations for changes in the medical curricula

The integration and visibility of genetics in the medical curricula is very limited (Chapter 7), and it is therefore important that end-objectives for genetic education in the medical curriculum are defined and agreed upon. The time spent on genetics should be spent more efficiently, and there should be more focus on knowledge that is relevant for daily practice. Another solution could be to make the study methods more efficient, i.e. acquire knowledge on a more problem-orientated basis, integrating genetics more with other subjects. In that way, not only medical students, but also other specialist could benefit from the integration of genetics in these subjects. Furthermore, the potentials of applying genetics in medical care could also make medical students more alert for the possible application of new genetic tests in other disciplines. As argued by Korf, there should be a clearer connection between the basic genetics and the clinical approaches. In comparison, Boreham concluded that the lack of knowledge of pharmacotherapeutics among medical students gave rise to the same argument to promote the trend to integrate basic medical science into clinical cases, as in a problem-oriented curriculum. A possible solution to enhance the visibility of genetics in medicine is to introduce genetics not only as a more visible element of one
of the compulsory clerkships, such as gynaecology, paediatrics or internal medicine, but also to integrate genetics in the basic teaching of these subjects.

**Recommendations for further research**

Further research should focus on the following topics:

1. Research is needed in order to investigate what kind of genetic education physicians would like to have and what kind of genetic tests they are willing to implement in their own practice. A questionnaire study is already being carried out among GPs, GYNs, PEDs and midwives to investigate this subject (AMC Plass, VU University Medical Center, GenED-project part 2, unpublished data).

2. More research is needed to investigate the knowledge of genetics and beliefs concerning genetic testing among other specialists, e.g. neurologists, to assess whether these other groups of specialists also need training in specific genetic subjects.

3. In the long term, when changes have been made in the education programmes, the level of genetic knowledge among medical students nearing graduation and among non-genetic health care providers should be re-assessed in order to evaluate the effectiveness of the education programmes.

**Concluding Remarks**

The results of the studies presented in this thesis have provided better insight into the level of knowledge of genetics, beliefs, and self-perceived competence concerning genetic testing among non-genetic health care providers. 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 from patients for genetic testing. 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 genetics-related healthcare to patients. Furthermore, many non-genetic health care providers have a positive opinion on offering genetic testing if the patients themselves ask for these tests, and if these tests are common practice. Therefore, education programmes for the public are recommended, together with clear guidelines for referral and genetic testing.
References


