A Clinical Perspective on Ethical Issues in Genetic Testing

R. H. Sijmons, M.D., Ph.D., 1 I. M. Van Langen, M.D., Ph.D., 1 and J. G. Sijmons, MM.L., Ph.D., Ph.D. 2

1Department of Genetics, University Medical Centre Groningen, University of Groningen, Groningen, The Netherlands
2Molengraaff Institute for Private Law, Faculty of Law, Utrecht University, Utrecht, The Netherlands

Genetic testing is traditionally preceded by counselling to discuss its advantages and disadvantages with individuals so they can make informed decisions. The new technique of whole genome or exome sequencing, which is currently only used in research settings, can identify many gene mutations, including substantial numbers of mutations with unknown pathological effect; it may, therefore, threaten this balanced approach if it is used in a clinical setting. We discuss the ethical challenges of several approaches to pre- and postnatal DNA testing, individual privacy versus the interests of families and of scientists, and the clinician’s duty to re-contact if new information or options become available.

Keywords: ethics, exome sequencing, genetic counselling, genetic testing, privacy, whole genome sequencing

INTRODUCTION

To patients and their clinicians, the worldwide effort to unravel the genetic nature of health and disease holds the promise of more detailed and timely diagnoses, better prediction of health risks, and improved prevention and treatment of diseases. Knowledge of individual genomes is expected to lead to more personalized medicine (Ginsburg and Willard, 2009; Guttmacher et al., 2010). So far, however, the genetics revolution, and the field of pharmacogenomics, has had less impact on daily medical practice than some had predicted (Daly, 2010; Feero et al., 2010; Hamburg and Collins, 2010; Relling et al., 2010; Varmus, 2010). Patients and families with single gene disorders have probably...
gained most as genes and mutations associated with these types of disorders are increasingly being identified, facilitating diagnosis and sometimes treatment. In contrast, the study of the genetic contribution to common disorders has yet to lead to major improvements in their healthcare. Genetics research is, however, gaining momentum and is clearly changing from a small, isolated field of academic study into a topic relevant to many different medical disciplines. The genetics of several types of cancer and of cardiovascular disorders are among the most prominent examples today. With this increasing momentum, the ethical aspects of genetic testing as an integral part of health care are also gaining importance. Some of the ethical challenges that are now clinically relevant are relatively old in nature, but they now need to be dealt with on a wider scale. Other challenges are new, resulting from the application of recently developed, high-throughput techniques, for example whole genome sequencing. Here we review these clinically relevant ethical issues against the principles of good professional conduct in medicine: do good, do no harm, and respect the patient’s autonomy (Tabor and Cho, 2007). We acknowledge that differences in cultural, ethnic and religious backgrounds, and in economical resources are relevant to a discussion of “genethics.” In addition, legislation and medical professional guidelines have an impact on applying genetics to healthcare; they may differ widely between nations, but are not independent of the cultural, ethnic, and religious backgrounds. We do not aim to cover all the possible differences, but have instead chosen to review the practical ethical issues in genetic testing as seen from our own clinical perspective (see Appendix 1 for a summary of clinical genetic practice in the Netherlands).

THE TRADITIONAL CLINICAL GENETIC APPROACH TO GENETIC TESTING

Genetic testing is traditionally carried out in a clinical setting where a particular individual and/or family medical history has led to the suspicion of one or more genetic, single gene, disorders running in the family. In this setting, as in the case of genetic testing for certain congenital malformations or mental handicaps, it is used as a diagnostic tool. DNA test results may confirm a particular diagnosis or make it less likely. It is usually the desire for a diagnosis that makes patients or the parents of affected children decide to undergo such testing, in the hope that it will reveal the cause of their disease, help establish a prognosis, offer a guide for therapy, or support decisions on having children. Alternatively, it may decrease stress in patients and their families if it can be concluded that particular genetic disorders are unlikely given a negative DNA test result. In healthy individuals who may carry a mutation that has been identified in their families, presymptomatic DNA testing (also referred to as predictive testing) is possible. Such testing warrants special attention, as it may lead to the prediction of a relatively high risk for serious disease
in as yet healthy individuals. Identifying these genetic risks may lead to discrimination in work and taking out insurance, and may cause psychological stress. On the other hand, there may be effective preventive options available, as in hereditary colorectal cancer for example, and even in an untreatable disease, knowledge of carriership may guide an individual’s future choices. Unlike their symptomatic relatives, asymptomatic family members can choose to remain ignorant of their exact risk, although in time they, too, may manifest the disorder. Psychological studies in subjects undergoing predictive testing for treatable as well as untreatable monogenic disorders have revealed that no serious, long-term negative effects need to be feared, provided that state-of-the-art genetic counselling is given with the testing. However, predictive testing of children requires special consideration. Testing children for carriership of adult-onset disorders would violate their right to weigh the advantages and disadvantages of testing for themselves when they reach adulthood. The general consensus is, therefore, that testing of asymptomatic children should be restricted to testing for those disorders that manifest and require medical intervention during childhood, like certain cardiovascular disorders that are associated with sudden cardiac death at a young age if left untreated. The right of parents to make autonomous choices on behalf of their offspring is, therefore, sometimes overruled by the medical professionals involved with these families.

Because of the large impact that genetic testing may have on medical management, psychological well-being, family relationships, individual financial circumstances, and on the predicted health status of offspring and other blood relatives, DNA testing is not necessarily a good thing to do at a certain point in time for a certain individual. Therefore, to avoid maleficence and support autonomy, DNA testing is generally preceded by one or more genetic counselling sessions (Uhlmann et al., 2009) during which the medical, psychological, and economical advantages and disadvantages of diagnostic or predictive testing are discussed with the patients and/or with the parents, when appropriate. In this process, the testing for untreatable conditions associated with a high morbidity and mortality requires additional effort since the psychological burden may be greater. After testing, the results are discussed, and the medical management is then adapted when appropriate and psychosocial support offered when necessary. Together, this is a time-consuming process that can, however, be justified by the complex nature of the differential genetic diagnosis, DNA testing and the potential impact of the test results on the patients and their relatives. Deviations from this careful and evidence-based model of genetic testing and counselling may be practical for logistic or commercial reasons, but the benefits for the tested individuals should still be evident and outweigh the burden such an approach imposes on them. Alternative ways of genetic counselling, like group counselling and the use of decision aids, are being evaluated and may be useful in situations where resources for individual counselling are not available. In all cases, counselees
should be given enough information to reach a well-informed decision, as informed consent is a leading principle in both medical ethics and patient's right (e.g., Article 5 Biomedicine Convention (Council of Europe, 1997; Dute, 2005). There are several examples of these nontraditional types of genetic testing, and they are likely to become increasingly important.

**WHOLE GENOME OR EXOME SEQUENCING**

New technology and its ever decreasing costs now permit the analysis of the whole genome, or of all the protein coding regions, referred to as the “exome,” of selected individuals (Feero et al., 2010; Ormond et al., 2010). These individuals are selected because they have either been diagnosed on clinical grounds with a certain genetic disorder for which the molecular cause should be unrav-elled, or they are suspected of having an unknown genetic disorder: in both cases it has not been possible to identify a germline mutation through the so-called candidate gene approach. This latter, traditional, approach involves testing for mutations in genes known to be able to cause the same or very similar disorders. Using whole genome or exome sequencing in such cases raises several ethical issues that need to be addressed. Although this new and powerful technique may indeed increase the chances of identifying the gene mutation responsible for the patient’s disorder, knowledge of the exact function of that gene may still be limited and, therefore, the impact on the prognosis, prevention, and treatment of the disorder may still be rather small. In addition, the hunt for the causative gene mutation will involuntarily, and on an unprecedented scale, also identify many mutations that have nothing to do with the present disorder, but which may nevertheless have important health implications for the individuals as well as their relatives (Ashley et al., 2010). Genetic predispositions for treatable as well as untreatable diseases may thus be revealed, and the nature of these conditions, therefore, warrants discussion with the individual before testing. For the thousands of different conditions that may be discovered, this approach becomes unrealistic and thus threatens the individual’s informed decision-making and, therefore, his or her autonomy (McGuire and Lupski, 2010). Thus, whole genome or exome sequencing, if performed outside the research setting, challenges the presupposition of available essential information leading to an informed consent.

In addition to this ethically important drawback, thousands of DNA variants may be revealed in a single test with as yet unknown health risk associations (Ormond et al., 2010). Last but not least, the sequencing technique is not perfect and even near 100% accuracy will, because of the massive scale of sequencing to be implemented, lead to the false identification of thousands of mutations and variants (Ormond et al., 2010). Each of the findings that are potentially clinically relevant would, therefore, need to be confirmed by extra, traditional testing. We feel these considerations, at the present time, form
a strong argument against introducing whole genome or exome sequencing for general diagnostic work and reporting the findings outside special clinical research settings. Within research settings, a practical solution with respect to informed decision-making might be to sequence the whole genome or exome but to give individuals the choice of learning only part of the results. For example, only mutations in genes leading to preventable or treatable disorders, or, in the case of children, only to those conditions medically relevant at their particular age. Individuals might also choose to learn of different types of test results at different times in their life rather than all at once, although this requires special measures for storage and retrieval of the data. Other options would be to reveal only findings relevant to the particular disorder that was the indication for performing whole genome or exome sequencing screen. In a research setting, revealing no test results at all to the individuals would also be an option. We expect whole genome or exome sequencing techniques to be readily applicable to the clinical testing for particular groups of genes, by creating test subsets for genetically heterogeneous disorders, for example, mental retardation or congenital heart disease. This approach will primarily be used in a diagnostic setting to find the cause of disease in patients.

Historically, genetic disorders are diagnosed in families after one or more clinical manifestations in those families and only after there is a suspicion of high genetic risk. This may eventually lead to requests for prenatal detection of the particular disorder. Some couples, however, might choose to prevent all births of seriously affected children before anyone has been affected in their own, immediate family. Thus, a future application of modern sequencing techniques might be to offer screening for carriership of all autosomal recessive (AR) hereditary disorders, for example, to such couples. Carriers of a mutation for an AR disorder usually have a negative family history for that disorder until they have their first affected child. In most cases, a single AR mutation does not cause health effects in the carriers themselves, but there are exceptions, e.g., carrying a mutation in the ataxia telangiectasia (ATM) gene increases sensitivity to ionizing radiation. All individuals carry one or more AR diseases. If a couple both carry a mutation for the same AR disorder, their chance of having an affected child is 25% in each pregnancy, and counselling could focus on that particular disorder and prenatal testing or pre-implantation genetic diagnosis (PGD) (see section below) could be offered. If unrelated couples start a family, their chance of having a child with such a disorder is generally small. However, specific ethnic groups have been shown to have a high risk for certain AR disorders, e.g., the blood disorder thalassemia, and they are already being offered testing for it on a population basis. In addition to testing for mutations for AR disorders outside these particular ethnic groups, testing for carriership of basically all X-linked recessive (XLR) disorders in women who want to have children would be a comparable approach. These XLR diseases manifest in sons who carry the mutation.
PRENATAL DIAGNOSIS AND PGD

There are special ethical aspects to prenatal diagnosis and termination of pregnancy as these involve care for the unborn child, for the mother, and other direct relatives. Prenatal testing and abortion are, therefore, typically subject to legislation that protects the rights of the child and of the mother, as well as to guidelines issued by religious authorities. Most efforts in prenatal diagnostics are aimed at identifying chromosomal abnormalities that occur more frequently in the pregnancies of relatively older women, although they can occur at any age. Down syndrome, also referred to as trisomy 21, the presence of an additional chromosome 21, is the most typical example of such a congenital condition. The principles of screening, following the World Health Organization (WHO) principles as formulated by Wilson and Jungner (1968), have been widely discussed. The implementation of international recommendations, like those of the Council of Europe, R(92)3 (Council of Europe, 1992) and R(94)11 (Council of Europe, 1994), allow for different sorts of genetic and prenatal screening in European countries for conditions such as these. This population-based screening used to be performed through chromosomal analysis in chorionic villous biopsies or amniotic fluid taken from pregnant women over a certain age. Nowadays, there is a tendency to select women at risk of having a child with Down syndrome, or another numerical chromosomal abnormality, not just by their age but also by other risk parameters like specific biomarkers in maternal blood and ultrasound measurement of the amount of fluid behind the neck of the foetus, referred to as the nuchal translucency test. Typically, prenatal diagnosis is performed in the first trimester of pregnancy, which permits an easier abortion technique to be used if an abnormal foetus is detected and the couple decide to have the pregnancy terminated. The minority of prenatal testing is performed in pregnancies that have a high risk for a particular single-gene disorder. In these cases prior medical history, e.g., the previous birth or death of an affected child, has revealed such a risk and the wish for prenatal testing has been discussed during genetic counselling, preferably before the next pregnancy. PGD is a new technique of testing for single-gene disorders or chromosomal abnormalities and is performed on embryos that are just a few days old in order to select them for implantation based on the test outcome (Basille et al., 2009). Its main advantage is that it avoids selective abortion, which in our experience is what makes it attractive in the eyes of the prospective parents. Its main disadvantages are that it requires in vitro fertilization and the chances of becoming pregnant are much reduced compared to that of unassisted reproduction. As in postnatal genetic testing, prenatal testing and PGD should be performed only after a process of counselling and informed autonomous decision-making. Non-directiveness is one of the most important principles in genetic counselling and applies particularly to reproductive issues. It also applies to the termination of pregnancy.
With the increasing numbers of single-gene conditions that can be identified by DNA testing, although each is relatively rare, the numbers for prenatal DNA testing and PGD will increase (Robertson, 2003; Offit et al., 2006; Klitzman et al., 2008). Based on our experience and the literature, we have no particular concern that this type of expansion would lead to ethical challenges any different to those we face now, i.e., we would not be on the “slippery slope,” although we may one day be confronted with the problem of genetic selection, of choosing between the alternative lives for a series of pre-tested embryos. It is nowadays exceptional that parents request prenatal DNA testing for a condition that is regarded by our Western/European society as easily treatable or preventable. Because of this, at least in the Netherlands, there has never been a need to introduce a list of hereditary conditions that have sufficiently high associated degrees of morbidity and mortality to warrant prenatal testing and abortion, and instead the choice is left to the parents. In the Netherlands, there is such a list for PGD, partly because access to PGD is restricted for capacity reasons, and the procedure was first offered for conditions that were lethal or led to severe congenital disorders. Also, the introduction of PGD triggered a national political debate on the risk of reviving eugenics and the possible promotion of “designer babies.” In recent years, the most important question in this respect was whether diseases with incomplete penetrance and manifestation in adulthood, e.g., hereditary breast- and ovarian cancer, should also be eligible for PGD. As a result of this debate, the Dutch government has set up a national committee to formulate indications for PGD. In our opinion, it would be ethically sound to approach indications for PGD in the same way as for prenatal diagnosis, a procedure that now rightly puts much weight on the autonomy of the prospective parents (Handyside, 2010). Current legislation with regard to PGD varies greatly between countries, which encourages cross-border “medical shopping” (Soini, 2007). The reasons against applying whole genome or exome sequencing in a routine clinical setting have been discussed above. These reasons also apply to its use in prenatal diagnosis and PGD, but there are some additional ones. First, the time frame available to discuss and consider the findings, given the boundaries set by the Dutch law on abortion, is extremely limited. Second, many of the mutations found might not lead to a decision to terminate the pregnancy but would be relevant to adult-onset treatable diseases. The choice to know of such risk factors should preferably be made by the child on reaching adulthood, rather than disclosing this information to the parents at an earlier stage. An ethical dilemma arises when parents demand early disclosure as the legal representatives of their infant, in order to decide whether the child should exercise its right to know or right not to know.

We would like to emphasize that offering options for prenatal testing, PGD, and termination of pregnancy should never mean to imply that these options are in fact being advocated to the individual couple nor would we favor the idea of liability on account of parental negligence of genetic or prenatal
screening; see the French Perruche case and the Dutch Kelly case (Hondius, 2005). Autonomous choice can only be protected if optimal care for individuals born with congenital and/or genetic disorders is assured, even in economically difficult times.

NEWBORN SCREENING

Newborn population screening includes screening for a range of genetic conditions, most of them metabolic disorders, either through DNA analysis or analysis of metabolic products. These newborns are not screened because they have a prior high risk of developing these conditions based on family history, like an affected sibling. On the contrary, the risk for each of the screened conditions is usually relatively low. This screening is performed outside an individual genetic counselling setting where the advantages and disadvantages for testing for each of the conditions could be discussed with the parents before testing takes place. Instead, in newborn population screening, blood samples are collected during the first few days after birth while oral information on the testing is usually very limited, although supported by printed and online information. This process is justified because the tested conditions are serious and treatable, and such large-scale testing cannot include time-consuming individual counselling prior to testing. A thorough consideration of the advantages and disadvantages of testing is integral part of the process to obtain the state’s permission to conduct population-based screening, although not every aspect can be foreseen. Newborn screening can generate incidental results, notably on the carrier status of a newborn, i.e., the presence of a single mutation for an autosomal recessive disorder which would, in most cases, not result in an abnormal phenotype. Whether or not this information should be revealed to the parents rather than leaving the choice to the child on reaching adulthood is still under debate (Hayeems et al., 2008; Miller et al., 2009). With all this in mind, we consider that whole genome or exome sequencing should not be used in a newborn screening programme in the near future (see also the section on this technique) (Almond, 2006).

DIRECT-TO-CONSUMER TESTING

Some commercially available genetic tests, for example for some pharmacogenetic variants, or for particular highly penetrant disease-causing mutations, can be of practical medical value. However, the direct-to-consumer marketing of genetic tests, for single-gene disorders or complex diseases, has the obvious ethical disadvantage of limited or poorly informed autonomous decision-making. In a commercial setting, not undergoing testing is usually not a desirable outcome for a pre-test counselling procedure and, in many
cases, no counselling is offered at all. Still, for some individuals, the option to be tested outside a clinical setting, in anonymity, might have advantages, for example if they are concerned about genetic discrimination (Wolfberg, 2006). Not everyone trusts society’s adherence to the fundamental principles of non-discrimination on the grounds of genetic heritage (Article 11, Biomedicine Convention) (Council of Europe, 1997; Hendriks, 2005).

Personal genome testing is currently commercially offered to consumers, with screening for DNA polymorphisms that are associated with a range of health risks for complex disorders caused by multiple genetic factors in combination with lifestyle and environmental factors (Samuel et al., 2010). Each of these genetic risk factors is only weakly predictive of a particular disease, and science has yet to find a way to combine all the factors that predict an increase or decrease in risk into a clinically applicable model (Hunter et al., 2008; Janssens et al., 2008). From our clinical point of view we, therefore, do not support such testing yet. For the reasons discussed earlier, we also see no place for direct-to-consumer whole genome or exome sequencing in the near future.

PRIVACY AND GENETICS

The privacy of individuals undergoing genetic testing must be protected. The outcome of such testing, however, can have severe implications for health risk estimation and the medical management of the patient’s relatives who share the same DNA, and this alone suggests that privacy should not be maintained under all circumstances (Gilbar, 2007). In a way, an individual’s genetic diagnosis is also a family diagnosis. Fortunately, in our Western/European society, tested individuals are usually keen to share important information on the genetic diagnosis with their relatives; the importance of the genetic diagnosis for offspring and other relatives is usually one of the reasons for seeking testing in the first place. Patients might need help to spread information in their families in a meaningful way, and the clinic should offer support in this for example by providing family letters. The level of information given to relatives should take into account that some would rather not have known this information, the so-called “right not to know” (Andorno, 2004; Gilbar, 2007; Forrest et al., 2007). The “reasonable” individual, however, is expected to appreciate being informed on genetic health risks, particularly for treatable conditions. A major ethical dilemma develops especially in situations where patients refuse to share information on a diagnosis made for a serious genetic disorder that is highly preventable and where there is a high risk to relatives carrying the same disease mutation. In such cases, the clinician may suspect that the relatives would rather know than not know of the diagnosis and faces a conflict of interest with respect to patient–doctor confidentiality. Of course, all
A Clinical Perspective on Genetic Testing Ethics

these issues should have been discussed with patients before testing, but with respect to the legal and professional guidelines in cases where a problem nevertheless arises, no consensus has been reached yet. One can even argue that the clinician should actually check whether clients have in fact spread medically useful genetic information within their family, although in many cases it would be technically difficult to perform such a check. Again, there is little consensus on how to deal with these issues (Lucassen and Parker, 2010).

Another threat to privacy is the scientific need to put information on patients’ genotypes and the associated clinical details, phenotypes, into public scientific databases, the locus-specific databases (LSDBs). Only through such a global effort can most of a common class of mutations, the missense mutations, be clinically interpreted. Missense mutations are identified in many individuals, but their clinical importance remains unclear unless large datasets are examined. The outcome of such a scientific analysis is, therefore, in the direct interest of the patients tested. Although only anonymous patient data are submitted to the LSDBs, patients might still be identifiable by, for example, their relatives in the case of a rare disease, and/or a rare missense mutation, and/or a specific geographic occurrence of the disease. Putting less detailed and, therefore, less identifiable information into the LSDBs reduces the value of such databases, making it clear that resolving this issue is an ethical challenge. It is now under debate in the professional community (Povey et al., 2010). Interestingly, patients might actually want to be able to contribute data directly to these databases and thus be in direct control of the amount of detail visible. Equally, they would always be legally entitled to withdraw their data from the database.

DUTY TO RE-CONTACT?

The knowledge on all aspects of genetic disease is constantly evolving, and new information might become available that is important to individuals and their relatives who have been tested and counselled in the past. Although this might be true for all fields of medicine, a duty to re-contact—though not a well-defined and acknowledged obligation in general—is a special concern in genetics because individuals may not act on information for years after the genetic consultation, e.g., with respect to reproduction, genetic diagnostics have preventive and therapeutic implications in an increasing number of cases, preventive options change over time, and information may be relevant to an extended family (Hunter et al., 2001). On the other hand, the duty to re-contact patients when new medical information becomes available is still ethically controversial, if only because it is not clear that new information will always be welcomed at that moment. In addition, genetics clinics generally lack the manpower to handle re-contacting efforts and would often face
technical difficulties in retrieving from their files the patients with unclear diagnoses who might now have new diagnostic testing options. We, therefore, currently ask our patients to contact us again after a couple of years, or if there is a change in family or personal medical history, or whenever they feel new information would be important for them. Online databases and disease-based Web sites for patients to check whether any progress has been made for a particular genetic disorder and the continued education of other healthcare workers who may still be in direct contact with the patients are other options to ensure that new information reaches the patients and families involved.

CONCLUDING REMARKS

The increasing knowledge on the genetics of health and disease brings expanding pre- and postnatal genetic testing options. Testing is not necessarily a good thing under all circumstances, and clinical guidance to help patients and their families make autonomous, informed decisions and protect their privacy are ethical cornerstones in clinical genetics. Whole genome or exome sequencing is a major technical breakthrough, but its clinical application should, in our opinion, be introduced stepwise and in a controlled clinical research setting in order not to violate these ethical principles. We need to educate healthcare workers as well as the public about what is possible and impossible in the field of genetics, to avoid hypes and misinformation (Brower, 2004; Burke, 2004; Schmitz, 2010). There should be a continued public and professional debate on the ethical issues arising in the evolving field of applied genetics in individual and population screening settings, ranging from access to healthcare and the privacy of genetic data, to the options for prenatal testing and assisted reproduction (Tomasini, 2007; Landeweerd, 2009).

APPENDIX 1: SUMMARY OF CLINICAL GENETIC PRACTICE IN THE NETHERLANDS

DNA Diagnostics

DNA diagnostics for germline mutations may legally only be performed in, or in close cooperation with, the DNA laboratories connected with the Departments of Clinical Genetics of the university hospitals, and for cancer syndrome diagnostics only, in the laboratory of one specialist cancer hospital. Most DNA diagnostics and all presymptomatic DNA testing can only be requested by, or in close consultation with, clinical geneticists. The results are always discussed with the patient by the clinical geneticist or a genetic counsellor working under the supervision of a clinical geneticist. As a result, the genetic counselling around these diagnostic tests is guaranteed in the
Netherlands. The diagnostic DNA laboratories are organized in a national platform on DNA diagnostics where, for example, quality control issues are discussed (LOD, www.dnadiagnostiek.nl). The laboratory specialists performing diagnostic genome analysis have their own professional society (VKGL, www.nvhg-nav.nl).

**Genetic Counselling**

Complex genetic counselling is only offered in clinics organized by the departments of clinical genetics of the university medical centers, and in the familial cancer clinic of one specialist cancer hospital. Genetic counselling is provided by clinical geneticists and genetic counsellors. Clinical genetics is an officially recognized medical specialism with its own professional society (VKGN, www.vkgn.org). There is a formal training for genetic counsellors and a dedicated professional society (NVGC, www.nvgc.info). For psychosocial support, specialized staff members are available in the Departments of Clinical Genetics, such as social workers and psychologists. This group has its own professional society (GLOBE, www.globe-nl.org). There is a growing number of national protocols to support the genetic testing and counselling for genetic disorders.

**Genetic Diagnosis and Insurance Issues**

Genetic counselling, genetic testing, preventive surveillance, treatment, and follow-up for genetic disorders are covered by regular Dutch health insurance. This is a national system of obligatory health insurance with private health insurance companies. These companies are legally obliged to provide a package with at least a minimal set of insured treatment, as defined by the government, including all curative treatment. It is illegal for insurers to refuse an application for this basic insurance or to impose special conditions. Affordability is guaranteed through a system of income-related allowances and individuals and employers pay income-related premiums. With respect to life and disability insurance, insurance companies apply thresholds under which no information on genetic diagnoses is requested from clients. In 2010, these thresholds were €160,000 for life insurance and €32,000 for disability insurance.

**REFERENCES**


