Recommendations for genetic counselling related to genetic testing

1. Introduction:

The main goal of the EuroGentest Network of Excellence (www.eurogentest.org) is to improve the quality of genetic testing. As patients' understanding of the results and consequences of the test is an integral part of genetic testing, EuroGentest also aims at improving the quality of genetic counselling services associated with genetic testing, across Europe. One of the goals is to establish recommendations for genetic counselling in connection with different testing situations.

2. Method:

To achieve this, the group assigned by EuroGentest for this task has collected and analysed international and European non-national guidelines and policies related to genetic counselling, as well as some relevant national recommendations and other documents. In addition, legislation related to genetic counselling in EU countries has been collected. Three workshops attended by experts on genetic counselling have been organized (May 2005, September 2006, December 2007). In addition, data has been collected with the help of surveys. The draft of the recommendations was distributed to European clinical geneticists and genetic counsellors and European National Human Genetic Societies for comments. The final version was written taking these comments into consideration.

3. Background:

3.1. Analysis of European legislation related to genetic counselling

Article 12 of the European Convention on Human Rights and Biomedicine (Council of Europe, 1997) requires appropriate genetic counselling prior to predictive or carrier testing (including tests to detect a genetic predisposition or susceptibility), but it has not yet been ratified by all Member States (http://conventions.coe.int/Treaty/Commun/ListeTraites.asp?MA=9&CM=7&CL=ENG). According to our study, most Member States as well as other European countries do not have specific legal provisions on genetic counselling; exceptions include Austria, France, Germany, Norway, Portugal and Switzerland.
(http://www.eurogentest.org/web/info/public/unit3/regulations.xhtml)

3.2. Analysis of international recommendations

Analysis of the international guidelines and policies related to genetic counselling identified several issues that were consistently cited as important. The most commonly mentioned were (1) appropriate training for the counsellors, (2) content of the information, (3) counsellees understanding of the information, (4) psychological support, (5) problems related to disclosure to the relatives, (6) need for consent, (7) autonomy, (8) confidentiality, and (9) fear of discrimination. (http://en.eurogentest.org/files/public/unit3/summaryofguidelinesMay06.pdf)

3.3. Analysis of data collected from European Human Genetic Societies

A survey on legislation, guidelines and generally applied practices in genetic counselling performed among the human genetic societies or contact persons in 38 European countries, in 2005-2006, found that there is no legislation directly related to genetic counselling in the great majority of these countries. There are, however, some professional guidelines related to counselling in some of the countries that do not have that legislation. According to the respondent, 13 countries have neither legislation nor guidelines. About half of the respondents considered that more regulation would be needed, but 10 respondents considered the existing national or international guidelines to be sufficient. (http://en.eurogentest.org/files/public/unit3/Results_of_survey_1_WP_3-1_Dec06.pdf)

4. Definitions:

This paper defines genetic counselling as follows:

Genetic counselling is a communication process that deals with the occurrence, or risk of occurrence, of a (possibly) genetic disorder in the family. The process involves an attempt by appropriately trained person(s) to help the individual or the family to (1) understand the medical facts of the disorder; (2) appreciate how heredity contributes to the disorder and the risk of recurrence in specified relatives; (3) understand the options for dealing with the risk of recurrence; (4) use this genetic information in a personally meaningful way that promotes health, minimizes psychological distress and increases personal control; (5) choose the course of action which seems appropriate to them in the view of their risk and their family goals, and act in accordance with that decision; and (6) make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder (modified from Frazer FC: Genetic counselling. Am J Hum Genet 1974:26:636-661, Biesecker and Peters: Process Studies in Genetic counseling: peering into the black box. Am J Med Genetics 2001:106:191-198).

The “appropriately trained person” that gives the genetic counselling is usually a genetic health care professional (clinical/medical geneticist, genetic counsellor or genetic nurse). In some situations, he or she can be another professional trained for a specific counselling task, such as an obstetrician in the case of pre-test counselling for risk of aneuploidy due to increased maternal age.

The recommendations apply to genetic counselling related to genetic testing, whereby the term genetic test is used mainly for tests performed in genetic testing laboratories (cytogenetics, molecular genetics and biochemical genetics) as part of genetic services. It is acknowledged that the same need for genetic counselling may exist when analysing other elements that may disclose equivalent genetic information (histological, imaging, family history, etc).

5. Different types of genetic testing situations and need for genetic counselling

The following chapters (5.1.-5.8.) briefly present the need for pre- and post-test genetic counselling in different testing situations. Depending on the context, the disease being tested and the implications for the individual and his or her relatives, there may be different needs. It is assumed that clinical utility of the tests concerned is adequate. This paper focuses on the average situations, but the authors realise that there may be exceptions in each category.

5.1. Diagnostic testing means a genetic test performed in a symptomatic individual to confirm or exclude a genetic condition. This is usually not very different from other medical tests performed in order to achieve a diagnosis, except for the possible involvement of relatives or implications concerning them. Pre-test genetic counselling may not always be necessary. As in case of any medical test, there should be free and informed consent which includes pre-test information, minimally what the test is for and what its implications are for the tested and for the family. If the test result is positive, the patient and the relatives should be offered genetic counselling. Even when the test result is negative, genetic counselling may be indicated.

5.2. Predictive testing refers to genetic testing in a healthy high-risk relative for a specific later-onset monogenic disorder. The mutation in the family leads to the disease or a considerably high risk for the disease (like in high risk familial cancers). There is not a complete consensus on the terminology: Some use "presymptomatic testing" as a synonym for "predictive testing" - and even prefer this term - while others restrict the terminology "presymptomatic testing" to mutations with full penetrance. Those in favour of "predictive testing" use the term in the context of mutations with incomplete as well as complete penetrance. Even if the family has previously been counselled, further pre- and post-test genetic counselling has to be offered, often accompanied by psychosocial evaluation and support.

5.3. Susceptibility testing (sometimes referred to as risk profiling) means a genetic test of a marker or simultaneous testing of several genetic markers with the aim to detect an increased or decreased risk for a multifactorial condition in a healthy individual. The clinical validity and utility of risk profiling for diseases of complex aetiology needs to be proven before clinical use. If the test is or is claimed to be capable of detecting high relative risk for a serious condition and thus has significant implications for risk assessment, treatment or prevention in a person or his/her near relatives, then pre- and post-test genetic counselling is needed. At present, this is rarely the case in multifactorial diseases when testing healthy individuals with non-specific family history.

5.4. Pharmacogenetic testing means testing for a genetic susceptibility for adverse drug reactions or for the efficacy of a drug treatment in an individual with a given genotype. They are ordered mainly by specialists other than clinical geneticists; and the need for proper genetic counselling by a genetic specialist will depend on whether the results have other implications than the decisions about the drug treatment for the person tested and his/her near relatives.

5.5. Carrier testing means a genetic test that detects a gene mutation that will generally have limited or no consequence to the health of that individual. However, it may confer a high risk of disease in the offspring, if inherited, from one parent (in case of X-linked inheritance, autosomal dominant premutation or chromosomal translocation) or in combination with the same or another mutation in the same gene from the other parent (in case of autosomal recessive inheritance). Pre- and post-test genetic counselling needs to be offered.

5.6. Prenatal testing refers to a genetic test (either to detect a mutation, linked haplotype or chromosomal change) performed during a pregnancy, where there is increased risk for a certain

condition in the foetus. Pre- and post-test genetic counselling for the prospective parents needs to be offered.

5.7. Preimplantation genetic diagnosis (PGD) means testing the presence of a mutation, linked haplotype or chromosomal change in one or two cells of an embryo in a family with a previously known risk for a Mendelian or chromosomal disorder, in order to select the unaffected embryos to be implanted. Pre- and post-test genetic counselling for the prospective parents has to be offered. This should be differentiated from preimplantation genetic screening (PGS), which aims at improved results of infertility treatment in families with no known genetic risks. In case of PGS, reproductive counselling by assisted reproduction professionals is usually appropriate.

5.8. Genetic screening means testing where the target is not high risk individuals or families, but where the test is systematically offered to the general population or a part of it (e.g. newborns, young adults, an ethnic group, etc.). All of the previously mentioned testing types can, in principle, be performed either in families at risk or as screening programs in different parts of the population. In screening programmes, pre-test information and post-test information has to be an integral part of the program, though the extent and content of information in these lower risk situations, and the professionals involved, may vary. In addition to this information, those who are found to be in a high-risk group, as a result of screening, should be offered genetic counselling.

6. General recommendations for genetic counselling

- Genetic counselling should be regarded as an integral part of the genetic testing process. Genetic counselling cannot be compulsory; medical acts are very exceptionally compulsory. It should, however, be offered and strongly recommended in most testing situations as explained above. If an individual insists on having a test without genetic counselling, the medical facts and possible consequences should be discussed by the clinician ordering the test. In these situations, non-genetics health care professionals have a responsibility to recognize their abilities and limitations with regard to provision of genetic services. Furthermore, both genetics and non-genetics health care professionals should not agree to testing without pre-test counselling in circumstances where doing so would go against their professional judgement. According to good clinical practice, predictive tests for future severe illnesses with no options for treatment or prevention should not be performed without pre- and post-test genetic counselling, psychosocial evaluation and follow-up.

- Genetic counselling has to be provided or supervised by a health-care professional appropriately trained for genetic counselling.

- Genetic counselling should be given in a language well understood by the individual. When this is not possible, options such as using an interpreter should be offered.

- Before actual testing takes place, there should be free and informed consent. In situations where testing children or other persons who are not able to give informed consent is considered, those individuals should be involved in genetic counselling and in the decision-making process, according to their capacities. Adequate authorisation for genetic testing of children or persons who are not able to give informed consent is required from their parents or legal representatives. Testing for adult onset conditions in children should only be considered when treatment or surveillance would begin in childhood.

-If the counsellee decides to proceed for the test, a description of the circumstances associated with the test should be sent, together with the sample, to the testing laboratory as the interpretation of the results depends on the context.

- The resources needed to perform genetic tests and to provide the appropriate pre- and post-test counselling should be developed and put in place simultaneously.

6.1. Pre-test genetic counselling

a) In pre-test genetic counselling individuals are informed about the purpose of the test, including up-to-date, reliable description about symptoms and natural history of the disease, prospects of prevention or early diagnosis and treatment, inheritance pattern, the risk of disease in the counsellee's situation, available reproductive choices, reliability and limitations of the test concerned, and possible psychological impact and other consequences of the test result to the counsellee and his/her family/relatives. Privacy and confidentiality of the results, as well as possible consequences related to its disclosure to third parties, such as insurance companies and employers, are discussed, when appropriate. The counsellor is not coercive in any way; this principle is also explained to the counsellee.

b) Pre-test counselling includes discussion about the rights to know and to decide including the right not-to-know.

c) Possible uncertainties due to present lack of knowledge are declared

d) The discussion about the need to inform relatives about the test result, as well as the best ways to do this, are initiated, especially in conditions where early diagnosis may improve the prognosis.

e) Depending on the resources available, as well as the context and the disease being tested, further genetic counselling sessions or consultation with a psychologist should be offered. The possibility of contact with a social worker and/or patient /lay support organisations should also be offered, where applicable.

f) Written materials and/or reliable Internet addresses related to the subject should be offered when available.

g) The counsellor should offer assistance in decision making, and encourage the counsellee to take ample time for it, whenever possible.

h) A written summary of the discussion should be prepared, if the counsellee so wishes.

6.2. Post-test genetic counselling

a) After disclosure of test results, the first focus is on the emotional impact on counsellee and others involved. Depending on the resources available, as well as the context and the disease being tested, follow-up contacts with the genetic counselling unit should be offered, and/or a consultation with a psychologist. The possibility to contact a social worker and patient/lay support organisations should also be offered. A written summary of the test result and issues discussed during the counselling should be, as a rule, given to the counsellee.

b) Points a) and b) from pre-test genetic counselling may have to be repeated.

c) Implications to the individual (including a follow-up plan, when relevant) and his/her near relatives should be discussed

d) A strategy to inform relatives has to be discussed with the counsellee (or, if necessary, a decision to discuss this further, after time for reflection).

e) Written material to help the counsellee to spread the information in the family may also be offered.

Depending on the emotional impact of the test result, it may be appropriate to discuss these issues in the disclosure session or in one or more follow-up sessions.

6.3. Information/counselling in screening programs

When a genetic test is offered within a genetic screening program to the general population, the situation is very different from that of genetic testing in at-risk families. Most importantly, the individuals in a screening program have not personally requested the test, and they may not know anything about the condition being tested.

For these reasons, it is extremely important to inform the public properly about the condition to be screened for, and the issues related to the screening program, including testing methods and their reliability, the implications of both “positive” and “negative” test results to the individual and his/her near relatives, the need for a confirmatory test, as well as the freedom to choose to participate. This may be achieved using different methods, including media, leaflets and programs in schools. Individual pre-test genetic counselling is, as a rule, not possible to organize, but should be made available for those who request it.

Communication with the population or population groups and individuals to whom screening is offered may be performed by health care professionals other than clinical geneticists/genetic counsellors/genetic nurses, provided that the professionals involved are appropriately trained and educated for the screening programme. Genetics specialists should be consulted when educational programs are planned,

If the resources do not allow for such comprehensive and well planned pre-screening information, the screening programme should not be implemented at all (with rare exceptions confining unquestioned health benefits to the individuals being screened, e.g. newborn screening for PKU).

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