Summary of the guidelines for genetic counselling

Within the WP 3.1, a list of international and European non-national guidelines and policies related to genetic counselling has been collected. In addition, some relevant national documents as well as documents from other continents have been taken into account. We welcome further information on guidelines that deal with genetic counselling.

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Background
Progress in life sciences has raised concerns about the social, cultural, legal and ethical implications of the ongoing development. Since the 1970s, the field of bioethics has grown considerably. The necessity of setting universal ethical guidelines for genetic services has been expressed in the past few years by scientists and practitioners themselves and by lawmakers and citizens. In addition, health professionals in the field find many practical situations so complicated that they have recognised the need for professional best-practice guidelines. Genetic counselling is the main topic of only a few policy papers, but it is included in the various recommendations concerning genetic services. Even if counselling is not mentioned explicitly, it is pointed out when talking e.g. about information, decision-making and patient’s rights.

Policy papers
Bodies that have produced policies on genetic services can be divided into four categories: political institutions, professional organisations, ethical boards and patient associations. The policy documents can be broadly divided into two groups: there are official ratified policy papers and reports that serve as background material. There are altogether 56 documents listed on this website that deal with genetic counselling. The documents are produced by these bodies:

- International institutions (UNESCO, WHO, OECD)
- European institutions (Council of Europe, European Commission, European Parliament)
- International professional organisations (WMA, HUGO, FIGO, ISONG)
- European professional organisations (ESHG, ESHRE, EuropaBio)
- Professional organisations from other continents (ASHG, NSGC, AGS, SSO, ASCO, AAP, HGSA)
- Ethical Boards (Nuffield, Conference of European Churches, President’s Commission)
- Patient associations (EURORDIS, GIG, IHA, DPI)

* Abbreviations

The list of the documents is at the end of the website.
Summary of the guidelines for genetic counselling

Even though the documents differ from each other, there are certain issues that are mentioned in most of them. These formulate the mainstream discourse of the counselling guidelines. In the guidelines genetic counselling is seen as a unique process of providing very special information. The conception of the speciality of genetic information defines the practices of ideal counselling: it is expected to give people the best possible capabilities to live with and act upon their genetic information. The key concepts play particularly important role in the documents that deal explicitly with the counselling. There are also papers where these issues are not covered, as they concentrate more on the particular issues of counselling. The topics and their content are summarized in the table below.

<table>
<thead>
<tr>
<th>Subject</th>
<th>Summary of content</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>PERSONS INVOLVED IN COUNSELLING</strong></td>
<td></td>
</tr>
<tr>
<td><strong>Counsellor</strong></td>
<td></td>
</tr>
<tr>
<td><strong>Profession</strong></td>
<td>Person who gives genetic counselling should be a professional specialist, as he needs to convey complex information. In common problems there is a growing need for non-physician healthcare providers and non-genetic specialist MD’s to give genetic counselling. More education in genetics is needed.</td>
</tr>
<tr>
<td><strong>Training</strong></td>
<td>Genetic counsellors should receive appropriate and on-going training in genetic conditions, risk assessment, psychosocial issues, bioethics, service system, communication and patient perspective.</td>
</tr>
<tr>
<td><strong>Co-operation within healthcare system</strong></td>
<td>Genetic counselling team consists of a team of professionals.</td>
</tr>
<tr>
<td><strong>Duties</strong></td>
<td>Genetic counsellor has certain duties that include e.g. preparing for consultation, promoting public education, assisting in decision-making and being aware of the professional ethics.</td>
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<tr>
<td><strong>Family</strong></td>
<td></td>
</tr>
<tr>
<td><strong>Common information</strong></td>
<td>Genetic information is common to the whole family, which can cause conflicts. Counselling should be available to at-risk relatives. It should be considered before the test how they are contacted. Patients have a duty to contact their at-risk relatives, but this should not be a condition for test, as sometimes they have good reasons not to. Disclosure should be agreed between the patient and the physician.</td>
</tr>
<tr>
<td><strong>Cultural issues</strong></td>
<td>When providing information, family’s cultural and ethnic background should be taken into account.</td>
</tr>
<tr>
<td><strong>Vulnerable patients</strong></td>
<td></td>
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<tr>
<td><strong>Not able to consent</strong></td>
<td>Tests should be done in the best interest of the patients that cannot give informed consent, or sometimes in the health interest of their family. Authorisation of the representative is required, but the persons need to participate according to their capacity.</td>
</tr>
<tr>
<td><strong>Children</strong></td>
<td>Tests should be done in the best interest of the child, or sometimes in the health interest of their family. Authorisation of parents is required, but the child needs to participate according to his capacity. The age at which the child is able to decide on testing should be flexible, and the older the child is, the more significance should be put on his assent. Information should be appropriate to child’s capacity and needs, and the child should be involved in counselling as much as possible.</td>
</tr>
<tr>
<td><strong>Public</strong></td>
<td>Public should be educated about the availability of tests and about the impacts of genetic information. Public should also be involved in the dialogue on bioethics in general.</td>
</tr>
<tr>
<td><strong>PRACTICES OF COUNSELLING</strong></td>
<td></td>
</tr>
<tr>
<td>Setting</td>
<td>Genetic counselling should be given in a private and comfortable place.</td>
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</tr>
<tr>
<td>When to counsel</td>
<td></td>
</tr>
<tr>
<td><strong>Pre-test</strong></td>
<td>Pre-test counselling should be available.</td>
</tr>
<tr>
<td><strong>Post-test</strong></td>
<td>Post-test counselling should be available.</td>
</tr>
<tr>
<td><strong>Discussion before counselling</strong></td>
<td>It is recommended to discuss with patients before counselling to identify their needs, to increase their understanding of counselling and to gather background information.</td>
</tr>
<tr>
<td><strong>Periods</strong></td>
<td>There should be enough of time between counselling and decision-making. The waiting time should however not be too long, at least the test results should be disclosed as soon as possible. Patient should be informed about the time between the appointments.</td>
</tr>
<tr>
<td>Consent</td>
<td>Informed consent should be asked always. According to the situation, it should be either in written or verbal form, written form is often advisable. There should be a free withdrawal from the consent. Consent needs to be asked also to inform the relatives.</td>
</tr>
<tr>
<td>Psychological support</td>
<td>Psychological support should be given to patients when necessary to adjust them to the situation, to help them to cope with stress and to assure their individual decision-making. Patient’s psychological, emotional and social responses need to be taken into account. Sometimes it can be recommended to take a support person to the appointment.</td>
</tr>
<tr>
<td>Referrals</td>
<td>Referrals can be made to other professionals when necessary. Reason for a referral can be e.g. support, treatment or ethical reasons.</td>
</tr>
<tr>
<td>Follow-up</td>
<td>Effective systems for follow-up are needed.</td>
</tr>
<tr>
<td>Counselling integral part of testing</td>
<td>Counselling is integral part of genetic testing and more links between laboratories and clinical services are needed.</td>
</tr>
</tbody>
</table>

**INFORMATION-GIVING**

<p>| Content of information | Information should consist of medical facts of the disorder, risk figures, benefits and harms of testing, limitations of the test, reliability of the test, implications of testing, familial implications, probabilities of inheritance, prevention and treatment, information on available support and alternative choices. |
| What kind of information | Information should be appropriate, balanced, honest, objective, full, sufficient, accurate, age-appropriate and based on up-to-date knowledge. |
| Understanding | Patient’s understanding needs to be assured. Clear language should be used and interpreters invited when needed. Written summary of information is advisable. |
| Ways to give information | It is sometimes useful to use alternative ways to distribute information, such as leaflets, videos and visual presentation. |
| Test type – based information-giving | | |
| <strong>Predictive test</strong> | Appropriate counselling should be given. It should include advantages and disadvantages of knowing the result, psychological and social issues, complex appearance of the disease and difficulties to quantify probabilities, possibilities to treatment and planning of life. |
| <strong>Prenatal test</strong> | Appropriate counselling should be given. Options, limitations and risks of test should be discussed. Psychological aspects need to be taken into account. Medical facts and child’s future should be discussed. |
| <strong>Preimplantation test</strong> | Appropriate counselling should be given. |
| <strong>Carrier test</strong> | Appropriate counselling should be given and reproductive options discussed. |</p>
<table>
<thead>
<tr>
<th>VALUES</th>
<th>Non-directiveness</th>
<th>Non-directiveness needs to be ascertained to the patient. It does not mean abandoning patient, but personal conviction of the counsellor should not be involved. Non-directiveness is however difficult in practice and it needs to be considered case-specifically.</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Autonomy</td>
<td>Gene test should always be voluntary, only in rare cases can compulsory testing be accepted. Counselling needs to respect individual’s values and culture. No pressure should be put on the counsellee and every decision should be equally accepted. Independent decision-making should be encouraged, but counsellor can assist the patient.</td>
</tr>
<tr>
<td></td>
<td>Confidentiality</td>
<td>Confidentiality needs to be ascertained to the patient. Patient’s private information needs to be protected from third parties. In the situations that non-disclosure would cause severe harm, confidentiality can be breached.</td>
</tr>
<tr>
<td></td>
<td>Right to know and to not know</td>
<td>Right to know and to not know genetic information are both important, but sometimes, if it is in the patient’s interest, the right to know overweights the right not to know.</td>
</tr>
<tr>
<td></td>
<td>Well-being</td>
<td>In counselling patient needs to be taken care of. Good should always overweigh harm.</td>
</tr>
<tr>
<td></td>
<td>Equal access</td>
<td>Everyone should have equal access to genetic testing and counselling.</td>
</tr>
<tr>
<td></td>
<td>Individual’s interests</td>
<td>Interests of individual should prevail the interests of the society.</td>
</tr>
<tr>
<td>PROBLEMS</td>
<td>Practical problems</td>
<td>Varying practices related to genetic testing and counselling is a problem, as the services should be equal for everyone. Lack of trained professionals, inaccuracy of test results, risks involved in testing and costs of counselling are also problems to be solved.</td>
</tr>
<tr>
<td></td>
<td>Ethical problems</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Discrimination</td>
<td>There is a chance that there will be discrimination on basis of genes in the society. Testing can also stigmatize on basis of one’s ethnic origins. Testing for sex-selection should not be allowed in order to avoid discrimination on basis of sex. Testing creates attitudes towards disabled people, and in counselling the quality of disabled people’s lives is often defined without their contribution. Eugenics needs to be seen as a threat; a rich society includes all kinds of people.</td>
</tr>
<tr>
<td></td>
<td>Conflict between values</td>
<td>There is often conflict between confidentiality and duty to warn and provide care and between the right to know and the right to not know. These need to be handled sensitively.</td>
</tr>
<tr>
<td></td>
<td>Disclosure</td>
<td>It is sometimes difficult to decide what information to disclose, e.g. unexpected findings and information not related to disease.</td>
</tr>
<tr>
<td></td>
<td>Society</td>
<td>In genetic testing the beneficence of both the individual and the society needs to be thought. Increasing range of options that gene tests bring changes society. One of these options is commercial genetic testing in connection which proper counselling should be guaranteed.</td>
</tr>
<tr>
<td></td>
<td>Determinism</td>
<td>The use of genetic information may lead to deterministic thinking, and science may be considered as godhead.</td>
</tr>
<tr>
<td></td>
<td>Cultural norms</td>
<td>There are different cultural norms on health. Therefore dialogue is needed especially because of the cross-border patients.</td>
</tr>
<tr>
<td>RELATION TO OTHER HEALTHCARE</td>
<td>Genetic testing is different</td>
<td>Genetic information differs from other health information because it has familial and generational implications, it is predictive and the emphasis is not on treatment, it is severe, private and complex and deals with special ethical dilemmas.</td>
</tr>
<tr>
<td></td>
<td>Exceptionalism is not good</td>
<td>It is not good to think that genetic testing is too exceptional. This leads to deterministic thinking. Genetics should rather be integrated into mainstream healthcare.</td>
</tr>
<tr>
<td>FUTURE DEVELOPMENTS</td>
<td>Techniques increase</td>
<td>Genetic testing techniques will increase and more guidelines will be needed.</td>
</tr>
<tr>
<td></td>
<td>Focus on risk</td>
<td>Focus of testing will be more and more on risks of illnesses and the class of healthy ill will grow. This brings more choices and freedom but ethical dilemmas also need to be thought more carefully.</td>
</tr>
</tbody>
</table>
Professional practices

Patient and physician’s roles are changing and more interchange of information is needed. The role of counselling is increasing. Counselling needs to be evaluated and researched.

Healthcare and society

Genetics will be integrated more into everyday healthcare and healthcare costs need to be reviewed. Differences between countries continue to exist, as value basis is always different in different cultures.

* Abbreviations:

UNESCO: United Nations Educational, Scientific and Cultural Organization
WHO: World Health Organization
OECD: Organisation for Economic Co-operation and Development
WMA: World Medical Association
HUGO: Human Genome Organisation
FIGO: International Federation of Gynecology and Obstetrics
ISONG: International Society of Nurses in Genetics
ESHG: European Society of Human Genetics
ESHRE: European Society of Human Reproduction and Embryology
EuropaBio: The European Association for Bioindustries
ASHG: American Society of Human Genetics
NSGC: National Society of Genetic Counselors
AGS: American Geriatrics Society
SSO: Society of Surgical Oncology
ASCO: American Society of Clinical Oncology
AAP: American Academy of Pediatrics
HGSA: Human Genetics Society of Australasia
Nuffield: Nuffield Council on Bioethics
EURORDIS: European Organisation for Rare Diseases
GIG: Genetic Interest Group
IHA: International Huntington Association
DPI: Disabled People’s International

List of policies and guidelines related to genetic counselling

International institutions

1. UNESCO: Universal Declaration on the Human Genome and Human Rights, 1997
2. UNESCO: Preliminary Draft Declaration on Universal Norms on Bioethics, 2005
3. UNESCO: Report on Genetic Counselling by Michel Revel, 1995
   Web address: http://portal.unesco.org/shs/en/file_download.php/e5ec8f48c2de32a26171790bbdda05eccounsellingCIB3_en.pdf
4. UNESCO: Report on Confidentiality and Genetic Data by the working group of IBC, 2000
5. UNESCO: Report on Genetic Screening and Testing by David Shapiro, 1994


10. OECD Programme on Biotechnology: Genetic Testing – guidelines for millennium
Web address: http://213.253.134.29/oecd/pdfs/browsit/9300051E_PDF

European Institutions


12. Council of Europe: Recommendation No. R(92) 3 of the Committee of Ministers to Member States on Genetic Testing and Screening for Health Care Purposes, 1992
Web address: http://www.coe.int/T/E/Social_Cohesion/Health/Recommendations/Rec(1992)03.asp#TopOfPage

13. Council of Europe: Recommendation No. R(90) 13 of the Committee of Ministers to Member States on Prenatal Screening, Prenatal Genetic Diagnosis and Associated Genetic Counselling, 1990


Web address: http://www.coe.int/T/E/Legal_Affairs/Legal_co-operation/Bioethics/Texts_and_documents/DIR-JUR%2897%2913Genetics.pdf

16. European Commission Joint Research Centre: Towards quality assurance and harmonisation of genetic testing services in the EU, report 2003
Web address: http://www.jrc.es/home/pages/detail.cfm?prs=1124

Web address: http://www.europarl.eu.int/comparl/tempcom/genetics/final_report_en.htm

18. European Commission: 25 recommendations on the ethical, legal and social implications of genetic testing by an expert group of European Commission, 2004

International professional organisations

   Web address: http://www.wma.net/e/policy/c15.htm

21. World Medical Association: Declaration of the human genome project, 1992
   Web address: http://www.wma.net/e/policy/g6.htm

   Web address: http://www.gene.ucl.ac.uk/hugo/conduct.htm

   Web address: http://www.figo.org/content/PDF/ethics-guidelines-text_2003.pdf

   Web address: http://www.isong.org/about/ps_consent.cfm

   Web address: http://www.isong.org/about/ps_privacy.cfm

   Web address: http://www.isong.org/about/ps_vulnerable.cfm

European professional organisations

27. European Society of Human Genetics (ESHG): Provision of genetic services in Europe: current practices and issues policy, 2003
   Web address: http://www.eshg.org/PPPC.htm

   Web address: http://www.eshre.com/emc.asp?pageId=632

   Web address: http://www.eshre.com/emc.asp?pageId=418

   Web address: http://www.europabio.org/articles/article_317_EN.doc

Professional organisations from other continents


    Web address: http://genetics.faseb.org/genetics/ashg/pubs/policy/pol-29.htm

33. National Society of Genetic Counselors (USA): Code of Ethics
    Web address: http://www.nsgc.org/about/code_of_ethics.asp

    Web address: http://www.nsgc.org/about/position.asp


38. National Society of Genetic Counselors: Genetic evaluation and counselling of couples with recurrent miscarriage: Recommendations of the National Society of Genetic Counselors, 2005


Web address: http://www.americangeriatrics.org/products/positionpapers/gen_test.shtml

41. Society of Surgical Oncology: Statement on Genetic Testing for Cancer Susceptibility, 1999
Web address: http://www.annalsurgicaloncology.org/cgi/reprint/6/5/507

Web address: http://www.jco.org/cgi/content/abstract/JCO.2003.03.189v1?ck=nck

Web address: http://aappolicy.aappublications.org/cgi/reprint/pediatrics;107/6/1451.pdf

44. Human Genetic Society of Australasia (HGSA): Code of ethics, 2000

45. Human Genetic Society of Australasia (HGSA): Guidelines for the practice of genetic counselling, 1999

46. Human Genetic Society of Australasia (HGSA): DNA Presymptomatic and predictive testing for genetic disorders, 2002

47. Human Genetic Society of Australasia (HGSA): Child testing policy

Ethical boards

Web address: http://www.cec-kek.org/English/BioethGeneticTesting.pdf

50. President’s Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research: Screening and Counseling for Genetic Conditions. A Report of the Ethical, Social, and Legal Implications of Genetic Screening, Counseling, and Education Programs, 1983
Web address: http://www.bioethics.gov/reports/past_commissions/geneticscreening.pdf
Patient associations

51. EURORDIS – European Organisation for Rare Diseases: Guidelines for organisations providing information on rare diseases, 2004
   Web address: http://www.eurordis.org/IMG/pdf/pard3_guidelines_leaflet.pdf

52. Genetic Interest Group: Guidelines for genetic services, 1998
   Web address: http://www.gig.org.uk/docs/gig_guidelines.pdf

   Web address: http://www.gig.org.uk/docs/gig_confidentiality.pdf

54. International Huntington Association: Guidelines for the molecular genetics predictive test in HD, 1994
   Web address: http://www.huntington-assoc.com/

   Web address: http://freespace.virgin.net/dpi.europe/downloads/bioethics-english.pdf

56. Disabled People’s International Europe: Bioethics Declaration – The Right to Live and be Different, 2000
   Web address: http://www.johnnypops.demon.co.uk/bioethicsdeclaration/index.htm