

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 on-going 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.

Subject	Summary of content
PERSONS INVOLVED IN COUNSELLING	
Counsellor	
<i>Profession</i>	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.
<i>Training</i>	Genetic counsellors should receive appropriate and on-going training in genetic conditions, risk assessment, psychosocial issues, bioethics, service system, communication and patient perspective.
<i>Co-operation within healthcare system</i>	Genetic counselling team consists of a team of professionals.
<i>Duties</i>	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.
Family	
<i>Common information</i>	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.
<i>Cultural issues</i>	When providing information, family's cultural and ethnic background should be taken into account.
Vulnerable patients	
<i>Not able to consent</i>	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.
<i>Children</i>	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.
Public	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.
PRACTICES OF COUNSELLING	

Setting	Genetic counselling should be given in a private and comfortable place.
When to counsel	
<i>Pre-test</i>	Pre-test counselling should be available.
<i>Post-test</i>	Post-test counselling should be available.
<i>Discussion before counselling</i>	It is recommended to discuss with patients before counselling to identify their needs, to increase their understanding of counselling and to gather background information.
<i>Periods</i>	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.
Consent	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.
Psychological support	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.
Referrals	Referrals can be made to other professionals when necessary. Reason for a referral can be e.g. support, treatment or ethical reasons.
Follow-up	Effective systems for follow-up are needed.
Counselling integral part of testing	Counselling is integral part of genetic testing and more links between laboratories and clinical services are needed.
INFORMATION-GIVING	
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	
<i>Predictive test</i>	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.
<i>Prenatal test</i>	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.
<i>Preimplantation test</i>	Appropriate counselling should be given.
<i>Carrier test</i>	Appropriate counselling should be given and reproductive options discussed.

VALUES	
Non-directiveness	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.
Autonomy	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.
Confidentiality	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.
Right to know and to not know	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 outweighs the right not to know.
Well-being	In counselling patient needs to be taken care of. Good should always outweigh harm.
Equal access	Everyone should have equal access to genetic testing and counselling.
Individual's interests	Interests of individual should prevail the interests of the society.
PROBLEMS	
Practical problems	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.
Ethical problems	
<i>Discrimination</i>	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.
<i>Conflict between values</i>	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.
<i>Disclosure</i>	It is sometimes difficult to decide what information to disclose, e.g. unexpected findings and information not related to disease.
<i>Society</i>	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.
<i>Determinism</i>	The use of genetic information may lead to deterministic thinking, and science may be considered as godhead.
<i>Cultural norms</i>	There are different cultural norms on health. Therefore dialogue is needed especially because of the cross-border patients.
RELATION TO OTHER HEALTHCARE	
Genetic testing is different	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.
Exceptionalism is not good	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.
FUTURE DEVELOPMENTS	
Techniques increase	Genetic testing techniques will increase and more guidelines will be needed.
Focus on risk	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.

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

Web address: http://portal.unesco.org/shs/en/ev.php-URL_ID=2228&URL_DO=DO_TOPIC&URL_SECTION=201.html

2. UNESCO: Preliminary Draft Declaration on Universal Norms on Bioethics, 2005

Web address: http://portal.unesco.org/shs/en/file_download.php/10d16a8d802caebf882673e4443950fdPreliminary_Draft_EN.pdf

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

- Web address: http://portal.unesco.org/shs/en/file_download.php/48de04a5e6de8bc4966add86540d6c71Confidentiality_en.pdf
5. **UNESCO: Report on Genetic Screening and Testing by David Shapiro, 1994**
Web address: http://portal.unesco.org/shs/en/file_download.php/bf5fa8468519b693df4bcd609c03b384GeneticTestingCIB2_en.pdf
6. **UNESCO: International Declaration on Human Genetic Data, 2003**
Web address: http://portal.unesco.org/shs/en/ev.php-URL_ID=3479&URL_DO=DO_TOPIC&URL_SECTION=201.html
7. **WHO: Statement of WHO Expert Advisory Group on Ethical Issues in Medical Genetics, 1998**
Web address: http://whqlibdoc.who.int/hq/1998/WHO_HGN_ETH_98.2.pdf
8. **WHO: Proposed International Guidelines on Ethical Issues in Medical Genetics and Genetic Services, WHO Human Genetics Programme, 1998**
Web address: <http://www.who.int/genomics/publications/en/ethicalguidelines1998.pdf>
9. **WHO: Review of Ethical Issues in Medical Genetics. Report of Consultants to WHO, Professors Wertz, D.C., Fletcher, J.C. & Berg, K., 2000**
Web address: http://www.who.int/genomics/publications/en/ethical_issues_in_medgenetics%20report.pdf
10. **OECD Programme on Biotechnology: Genetic Testing – guidelines for millennium**
Web address: <http://213.253.134.29/oecd/pdfs/browseit/9300051E.PDF>

European Institutions

11. **Council of Europe: Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine, 1997 + Explanatory report**
Web address: <http://conventions.coe.int/Treaty/en/Treaties/Html/164.htm>
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](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/Social%5FCohesion/Health/Recommendations/Rec\(1990\)13.asp#TopOfPage](http://www.coe.int/T/E/Social%5FCohesion/Health/Recommendations/Rec(1990)13.asp#TopOfPage)
14. **Council of Europe, Working Party on Human Genetics: Working document on the applications of genetics for health purposes, 2003 + explanatory note**
Web address: [http://www.coe.int/T/E/Legal_affairs/Legal_co-operation/Bioethics/Activities/Human_genetics/INF\(2003\)3E_Wkgdoc_genetics.pdf](http://www.coe.int/T/E/Legal_affairs/Legal_co-operation/Bioethics/Activities/Human_genetics/INF(2003)3E_Wkgdoc_genetics.pdf)
15. **Council of Europe, Steering Committee on Bioethics (CDBI): Working Party on Human Genetics Report, 1997**
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>
17. **European Parliament Temporary Committee on Human Genetics and Other New Technologies in Modern Medicine: Report on the ethical, legal, economic and social implications of human genetics by Francesco Fiori, 2001**
Web address: http://www.europarl.eu.int/compar/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**
Web address: http://europa.eu.int/comm/research/conferences/2004/genetic/pdf/recommendations_en.pdf
19. **European Commission European Group on Ethics in Science and New Technologies (EGE): Opinion No 6 Ethical Aspects of Prenatal Diagnosis, 1999**
Web address: http://europa.eu.int/comm/european_group_ethics/gaieb/en/opinion6.pdf

International professional organisations

20. **World Medical Association: Statement on genetic counselling and genetic engineering, 1987**
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>
22. **Human Genome Organisation: Statement on the Principled Conduct of Genetics Research. HUGO ethical, legal, and social issues committee report to HUGO Council, 1996**
Web address: <http://www.gene.ucl.ac.uk/hugo/conduct.htm>
23. **International Federation of Gynecology and Obstetrics (FIGO): Recommendations on Ethical Issues in Obstetrics and Gynecology by the FIGO Committee for the Ethical Aspects of Human Reproduction and Women's Health, 2003**
Web address: http://www.figo.org/content/PDF/ethics-guidelines-text_2003.pdf
24. **International Society of Nurses in Genetics: Position Statement: Informed decision-making and consent. The role of nursing, 2000**
Web address: http://www.isong.org/about/ps_consent.cfm
25. **International Society of Nurses in Genetics: Position Statement: Privacy and confidentiality of genetic information: the role of the nurse, 2001**
Web address: http://www.isong.org/about/ps_privacy.cfm
26. **International Society of Nurses in Genetics: Position Statement: Genetic counselling for vulnerable populations. The role of nursing, 2002**
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>
28. **European Society of Human Genetics & European Society of Human Reproduction and Embryology: The Interface between Medically Assisted Reproduction and Genetics: Technical, Social, Ethical and Legal Issues, 2005**
Web address: <http://www.eshre.com/emc.asp?pagelid=632>
29. **European Society of Human Reproduction and Embryology PGD Consortium: Best practice guidelines for clinical preimplantation genetic diagnosis and preimplantation genetic screening, 2004**
Web address: <http://www.eshre.com/emc.asp?pagelid=418>
30. **The European Association for Bioindustries (EuropaBio): Human Medical Genetic Testing. A EuropaBio Position Paper, 2004**
Web address: http://www.europabio.org/articles/article_317_EN.doc

Professional organisations from other continents

31. **American Society of Human Genetics (ASHG): Ethical, Legal, and Psychological Implications of Genetic Testing in Children and Adolescents Report, 1995**
Web address: <http://genetics.faseb.org/genetics/ashg/pubs/policy/pol-13.htm>
32. **American Society of Human Genetics (ASHG): Paper on Professional Disclosure of Familial Genetic Information, 1998**
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
34. **National Society of Genetic Counselors (USA): Position Statements, 1991-2002**
Web address: <http://www.nsgc.org/about/position.asp>

35. **National Society of Genetic Counselors: Genetic cancer risk assessment and counseling: Recommendations of the National Society of Genetic Counselors, 2004**
Web address: http://www.guideline.gov/summary/summary.aspx?ss=15&doc_id=5274&nbr=3601
36. **National Society of Genetic Counselors: Fabry disease in genetic counselling practice: Recommendations of the National Society of Genetic Counselors, 2002**
Web address: http://www.guideline.gov/summary/summary.aspx?ss=15&doc_id=3274&nbr=2500
37. **National Society of Genetic Counselors: Genetic counselling for fragile X syndrome: Recommendations of the National Society of Genetic Counselors, 2000**
Web address: http://www.guideline.gov/summary/summary.aspx?ss=15&doc_id=2546&nbr=1772
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://hum-molgen.org/NewsGen/06-2005/msg39.html>
39. **National Society of Genetic Counselors: Genetic counselling and screening of consanguineous couples and their offspring: Recommendations of the National Society of Genetic Counselors, 2002**
Web address: http://www.guideline.gov/summary/summary.aspx?ss=15&doc_id=3273&nbr=2499
40. **The American Geriatrics Society: Position Statement: Genetic Testing for Late-Onset Alzheimer's Disease, 2000**
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.annalssurgicaloncology.org/cgi/reprint/6/5/507>
42. **American Society of Clinical Oncology: Policy Statement Update: Genetic Testing for Cancer Susceptibility, 2003**
Web address: <http://www.jco.org/cgi/content/abstract/JCO.2003.03.189v1?ck=nck>
43. **American Academy of Pediatrics, Committee in Bioethics: Ethical Issues With Genetic Testing in Pediatrics, 2001**
Web address: <http://aappolicy.aappublications.org/cgi/reprint/pediatrics;107/6/1451.pdf>
44. **Human Genetic Society of Australasia (HGSA): Code of ethics, 2000**
Web address: <http://www.hgsa.com.au/>
45. **Human Genetic Society of Australasia (HGSA): Guidelines for the practice of genetic counselling, 1999**
Web address: <http://www.hgsa.com.au/>
46. **Human Genetic Society of Australasia (HGSA): DNA Presymptomatic and predictive testing for genetic disorders, 2002**
Web address:
47. **Human Genetic Society of Australasia (HGSA): Child testing policy**
Web address: <http://www.hgsa.com.au/>

Ethical boards

48. **Nuffield Council on Bioethics: Genetic Screening – ethical issues, 1993**
Web address: http://www.nuffieldbioethics.org/fileLibrary/pdf/genetic_screening.pdf
49. **Conference of European Churches Commission for Church and Society, Working Group on Bioethics: Genetic Testing and Predictive Medicine, 2003**
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

53. Genetic Interest Group: Guidelines for confidentiality, 1998

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/>

55. Disabled People's International Europe: Disabled People Speak on the New Genetics. DPI Europe Position Statement on Bioethics and Human Rights, 2000

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>