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ARTICLE

What is ideal genetic counselling? A survey of current international guidelines

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The objective of this article is to review guidelines that address counselling in the context of genetic testing in order to summarise what aspects of counselling they consider most important, and to examine how they construct the ideal of genetic counselling. Guidelines were collected by examining the websites of different international professional, political, ethical and patient organisations, either previously known or found with the help of the Google search engine, and also using references listed in other studies. The most frequently mentioned topics in the collected 56 guidelines were sought, and this was carried out with the software package Qualitative Solutions and Research for Non-numerical Unstructured Data Indexing Searching and Theorizing. Topics related to genetic counselling that were mentioned in at least 30 of 56 collected documents were considered to be the most important aspects of genetic counselling. The ideal of genetic counselling is expressed in the analysed guidelines as being composed of (1) an appropriately trained professional who understands genetics and its ethical implications well; (2) relevant and objective information; (3) assurance of the counsellee's understanding; (4) psychological support; (5) informed consent; (6) confidentiality of genetic information; (7) considering familial implications; (8) appropriate handling of potential discrimination of testing; and (9) assuring autonomous decision-making by the counsellee. The ideal of genetic counselling is rather consistent in the guidelines, but there are some contradictions between the requirements of objective information-giving and adapting counselling to counsellee's circumstances.
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Introduction

Genetic counselling has been the topic of many studies and wide discussion because of its importance in providing and interpreting genetic information to patients and their relatives. There are several definitions of genetic counselling; probably the most often cited is that published in the *American Journal of Human Genetics*, in 1974, by FC Fraser ('Genetic counselling is a communication process which deals with the human problems associated with the occurrence, or risk of occurrence, of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained person to help the individual or the family to comprehend the medical facts, including the diagnosis, the probable course of the disorder and available

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management, to appreciate the way heredity contributes to the disorder and the risk of recurrence in specified relatives, to understand the options for dealing with the risk of recurrence, to choose the course of action which seems appropriate to them in view of their risk and their family goals and act in accordance with that decision, and to make the best possible adjustment to the disorder in an affected family member and to the risk of recurrence of that disorder').[1] The core of genetic counselling, according to this definition, is to present medical and genetic facts to the counsellees, and to help them to understand their meaning and choose the course of action most appropriate to them in relation to the genetic problem present in the family.

As the use of genetics in medicine has increased, both interest in and concern about the impact of genetic information on the life of individuals have been raised. For this reason, several organisations have produced guidelines for genetic counselling. The aim of this article is to review those international guidelines that address counselling in the context of genetic testing. Specifically, it aims to summarise the aspects in the guidelines that are regarded as forming elements of ideal genetic counselling. Topics that were considered to be the most important were sought in each document, regardless of the context, test type and situation. The ideal of genetic counselling, as expressed in international recommendations, is presented here by summarising the most frequently mentioned topics.

Before the first genetic counselling clinics were opened in the 1940s, the objective of genetics was to improve society through racial hygiene. As these eugenic goals began to fade, the interest of families became the main purpose of counselling. The need to dissociate genetic counselling from eugenics led to the current status of non-directive counselling.[2–4] The most salient aspects of counselling are now largely defined in the guidelines, and they play an important role in defining genetic counselling as they create the general framework for its best practice. They can be assumed to have an effect on the way genetic counselling is considered and practiced. The guidelines also fill the gaps in the legislation regarding genetic counselling, as the field of genetics is developing faster than the legislation, and as legislation might not be the solution that best adapts to all issues in genetic counselling.

Methods

The aim of this study was to collect, to the best of our knowledge, all available guidelines covering genetic counselling related to genetic testing, produced by the end of 2005, by global and European organisations. In addition to these, guidelines of relevant organisations from other continents and the USA were also sought. Most national guidelines were not included, not only due to their large number but also due to difficulties with language (National European guidelines have been collected by EuroGentest Unit 4 – Ethical, Legal and Social Issues Related to Genetic Testing.[5]). However, guidelines produced by the Nuffield Council on Bioethics and by the Genetic Interest Group, both based in the UK, were included because of their significant role in the discussion of genetic counselling. With guidelines we refer to, in addition to official guidelines, written statements, recommendations and reports that address issues related to genetic counselling.

The guidelines of 29 organisations were collected during 2005 by browsing their websites, either previously known or found with the help of the Google search engine or using references listed in other studies. This was part of Workpackage 3.1, in Unit 3 (Clinical Genetics, Community Genetics and Public Health) of 'EuroGentest – Genetic Testing in Europe', an EC 6th Framework Programme Network of Excellence.

The 56 documents collected were produced by political and ethical bodies, professional organisations, and patient associations (Supplementary Table 1; the URLs can be found on the www.eurogentest.org/unit3 webpage). Few of them were specifically written to address genetic counselling, but the topic was nonetheless covered in all of them. Their backgrounds differed, as some were official 'soft-law' statements, while others were bioethical declarations of principles, or more unofficial recommendations and reports.

Documents were coded for different topics and subtopics with the assistance of the software package QSR NUD*IST (Qualitative Solutions and Research for Non-numerical Unstructured Data Indexing Searching and Theorizing) that is designed to support the processes of coding data, searching for text and patterns of coding, and theorising about the data.[6–8] The most common topics related to genetic counselling were identified, their main contents reviewed, and the similarities and differences between the documents were analysed.

Results

In the guidelines analysed, nine topics related to genetic counselling were mentioned more often than the others. In this chapter, these topics will be discussed in the order of their appearance in the traditional genetic counselling session. All of these topics were mentioned in at least 30 of the 56 documents collected (Supplementary Table 1). These can thus be regarded as the most salient aspects through which ideal genetic counselling is defined. The topics are discussed here at a general level, even though the different documents placed differing levels of emphasis upon them. In addition to these topics, the guidelines also

brought out several other issues related to counselling in the context of genetic testing and these are presented in Table 1.

Table 1 The less frequently mentioned topics related to counselling in the context of genetic testing in the 56 documents studied

Topic	*Documents* (n) *mentioning this topic*
Equal access to genetic testing and counselling	25
Educating and informing the public	23
Genetic testing and counselling of children	21
Non-directiveness of counselling	21
Counselling in context of predictive testing	21
Timing of counselling	20
Right to know and not to know	18
Linking counselling better to testing services	17
Genetic testing and counselling of patients not able to give informed consent	16
Referring patients to other professionals	15
Professionals' conflict of values	15
Counselling in the context of prenatal testing	14
Differences between genetics and other health care	14
Counselling in the context of genetic screening	12
Well-being of the patient in genetic counselling	12
Follow-up of the patient	11
Problems of disclosure of genetic information	11
Considering cultural and ethnic aspects	10
Individual's interests at the core of the counselling	10
Focus on the risk of illnesses in the future	8
Counselling in the context of preimplantation testing	7
Role of patient organisations	7
Increasing role of counselling in the future	6
Genetic determinism	6
Alternative ways to present genetic information	6
Commercial genetic tests and counselling	6
Changing professional practices in the future	5
Rights and responsibilities in counselling	4
Counselling in the context of carrier testing	4
Opposing exceptionalism of genetic information	4
Genetic testing more into primary health care	3
Settings for genetic counselling	3
Counselling in the context of diagnostic testing	3
Lack of trained genetic counselling professionals	2
Costs of genetic counselling in the health care	2

Education and training of professionals

The appropriate education and training of the professionals who provide genetic counselling were seen as important in 42 of the 56 guidelines analysed. Given the complexity of the genetic information to be conveyed, the guidelines considered that only specialists in the field of genetics should perform counselling. Most guidelines required that professionals have 'appropriate training', without specifically stating what that training should be. It was also considered inevitable that the number of non-geneticists requesting genetic tests and disclosing test results would increase, as the use of genetic tests increases. Therefore, education in medical genetics as an integral part of health-care training was seen as much needed.

In addition to the emphasis on expert knowledge of genetics, the relationship between the counsellor and the counsellee was also considered an important aspect of training. The counsellor's role as a facilitator of the decision-making process was seen as important, and the counsellor was expected to understand the individual needs of the counsellee. The ideal relationship was considered to be one that is personalised and caring. Therefore, acquisition of good communication skills was seen as an important part of the training of professionals in genetics. In addition, patient organisations saw the education as a two-way process, where the geneticists could also learn from the experiences of their patients and the patient support groups.

The providers of genetic counselling were also expected to understand the ethical complexities of disclosing genetic information. Promoting equality and being aware of the social and ethical issues related to genetic testing were seen as important. Training in bioethics was specifically brought out in six guidelines.

Content of information to be provided

The topics that should be covered in genetic counselling related to genetic testing were defined in 42 guidelines. They included the following: (1) information about the condition in question, (2) treatment options, (3) the risk of having the condition, (4) the purpose, nature and consequences of the genetic testing in question, (5) the risks involved in the procedure, (6) the limitations of testing, (7) alternatives the counsellee should consider, (8) practical information on what will happen next, (9) the potential harm of testing, (10) the risks to family members, and (11) information on the support groups available.

Guidelines that concentrated on a specific test situation, such as prenatal diagnostics, specified the content of the information to be provided in that context. In most guidelines, the content of information was not discussed

in detail, but just listed. In general, the information was expected to be objective, adequate, balanced, understandable and adapted to the counsellee's circumstances.

The counsellee's understanding of genetic information

In the guidelines, genetic information was considered to be so complex that counselling would have to be very clear. In 30 guidelines, the counsellor was advised to check the counsellee's understanding throughout the consultation process and to focus on the aspects that the counsellee is really able to understand. Seven guidelines advised that the counselling should be given in the counsellee's own language, or that an interpreter should be used.

Psychological support

Psychological support and an empathic relationship between the counsellor and the counsellee were seen as important elements of genetic counselling in 30 guidelines. Some stressed the importance of support, stating that it is at least as important as the informational aspect, and that responding to the patient's emotional reactions may even take priority. Providing support in genetic counselling was seen as essential to enable the counsellee to make informed choices and cope with the test result.

The guidelines also advised the counsellor to consider that a test result – whether 'positive' or 'negative' – may alter the patient's self-concept. Relatives and friends, as well as support groups, were mentioned as important sources of emotional support. In the guidelines, counsellors were encouraged to suggest patients bring a support person, particularly when the test result is disclosed, and to refer the patients to appropriate professionals for further support whenever needed.

Implications for the family

Implications for the patient's family were brought out in 38 documents. Genetic information that is both personal and familial was seen as often raising questions about who the patient exactly is, and what the moral obligations of the professional are towards the relatives who are at risk of a genetic disease. Different documents emphasised disclosure of this information in different ways. All agreed that a patient has a moral obligation to share the genetic information with family members who are at risk, and that if this is relevant to other relatives, the patient should be recommended and even persuaded to disclose it. Some guidelines went further to suggest that the physician should be able to warn at-risk relatives when the disorder is serious and there is prevention or treatment available.

The guidelines also advised that the social impact that genetic information has on the family should be considered in counselling. It was seen as important that the genetic information is shared with the partner, especially if it affects the children or decisions regarding family planning. But, even when there are no plans to have children, the result of a genetic test was seen as having a considerable impact on the family life. The counsellor was advised in 10 guidelines to try to perceive the different roles of the family in different cultures and ethnic groups.

Confidentiality

Confidentiality of genetic information as a general principle that needs to be respected was brought out in 43 guidelines. There are two kinds of situations where confidentiality may be jeopardised. First, insurance companies, employers and school admission boards may be interested in the results of a gene test. The guidelines explicitly stated that privacy of information must be assured and the results must never be disclosed to third parties. Some guidelines noticed, however, that there may be exceptional cases, where disclosure may be in the best interest of the person in question, or of another person, whose life may be in danger.

Second, genetic information also relates to the family, which was seen to raise potential conflicts between confidentiality and the duty to warn the relatives. Therefore, confidentiality as an absolute principle was seen as problematic in many guidelines, and 12 documents stated that it may be breached in extreme cases when serious harm may be avoided, the at-risk relative is identifiable, and the disease is preventable or treatable. In these cases, the professional needs to judge the significance of the information, and discuss the conflict with other professionals. However, it was also pointed out that disclosure should not always supersede the individual's right to privacy, whatever the circumstances. Breaching confidentiality is an exception, and the professional always has to consider it very carefully.

Autonomy of the counsellee

In 38 guidelines, voluntary nature and autonomy in decision-making were regarded as the central issues of genetic counselling. Counsellees were expected to be able to choose freely whether to take the test, and what to do after receiving the test result. Appropriate, informed consent was seen as ensuring autonomy. Counsellors were expected to help the patients to make their own informed choices and to encourage independence of their decision-making process. No coercion or pressure was allowed – whether from the counsellor, state, community or family. Some guidelines specified that free choice means more than the absence of coercion: it means the practical ability to act afterwards on the decision that has been made. Thus, four guidelines considered the concept of autonomy problematic unless there is more than one viable alternative. The promotion of public education in genetics was seen as essential in order to protect free choice, as it was thought that an educated public is able and willing to make choices in its own best interest.

Genetic discrimination

Discrimination against a person on the grounds of his or her genetic heritage was seen as the major ethical question related to genetic testing that must be taken into account in genetic counselling. This was discussed in 32 guidelines that unanimously prohibited discrimination. The guidelines also suggested that society should support genetic differences. This means that there should be no indirect action either, such as limiting health-care access for patients with genetic diseases. In the guidelines of Disabled People's International, it was stated that the value of the human rights of the disabled is already diminished by the discriminatory attitudes in genetics. According to them, the stereotypic image of the perfect baby is promoted, and negative attitudes towards the quality of life of the disabled are encouraged.

Eugenics, as a form of discrimination, was discussed in 12 documents. It was considered that an essential distinction between genetics and eugenics is the importance ascribed to individual welfare, rather than to society. Four documents, produced by ethical boards and an organisation of the disabled, however, warned that new technologies have opened the door to a new eugenics, and that individual decision-making could collectively result in discrimination of minority decisions.

Informed consent

Informed consent from the patient to perform a genetic test was required in 39 guidelines. It was expected that the consent would be given freely after receiving the appropriate information. A written consent form was recommended in many guidelines, but verbal consent in some situations was considered to be sufficient. The possibility of freely withdrawing from testing at any time was described as an important part of the consent.

Discussion

The topics most frequently mentioned in the guidelines for counselling in the context of genetic testing indicate that ideal genetic counselling is comprised of three major elements that form the process of communication: information-giving, psychological support and ethical aspects.

Information-giving and psychological support

Traditionally, information-giving and support have been considered to form the core of genetic counselling.[1] This view was supported by the guidelines analysed. They listed what information should be given to the patient and also stated that the counsellor needs to provide psychological support. Some documents emphasised support over giving information. In the context of both information provision and support, adaptation to the patient's personal situation was encouraged. This was seen as being especially important when the patients have difficulties in understanding the information, and in cases where difficult decisions need to be made.

In the guidelines, genetic counsellors were expected to be trained professionals. In addition to education in genetic knowledge, training was seen as needed in communication skills and developing empathic relationships. The ideal counsellor was seen as someone who has good knowledge of human genetics and at the same time is an empathic person, whose communication is clear and who realises the special situations that patients are facing.

Ethical issues to be considered in counselling

In addition to the traditional conception of genetic counselling as a process involving elements of information-giving and support, the guidelines also highlighted the ethical issues involved in counselling. Genetic information was seen as ethically challenging, primarily because of its familial nature. Counsellors should not only be able to have a confidential relationship with the counsellees but also be able to deal with the impact that the genetic information may have on the family. In most guidelines, the counsellors were advised to ask the counsellees to disseminate information to their at-risk family members. Sometimes this is not possible, and the counsellors need to balance between confidentiality and the duty to warn, which has provoked wide discussion about the prioritisation between these values.[9] This is one reason why it was considered important in the guidelines that genetic counsellors have sufficient training in the particularities of genetic information.

Another challenge is ensuring the counsellee's autonomous decision-making. Consent was required by most guidelines to improve autonomy. There are, however, situations in which patients may not want to make autonomous decisions, or do not have the capacity to deal with the information. The counsellor was expected to be able to also deal with these situations, although autonomous decision-making could not be guaranteed. The extent, to which the principle of autonomy should be applied, was not discussed in most guidelines.

Autonomy is closely linked to the concept of non-directiveness, which has traditionally meant that the counsellor does not direct the decision-making process, but provides all the information needed for making an informed decision. This principle of non-directiveness has served as the central ethos for genetic counselling for the past decades, and has provided both practical and ethical guidance to professionals. There are several reasons why genetic counsellors may wish to claim that their work is non-directive. Respecting the patient's autonomy is a dominant principle of medicine, in general. There is also a desire to create a distance from the past of eugenics. The counsellors may also want to keep some emotional

distance from the counsellees and may not want to commit to their decisions, either legally or morally.[3,4,10]

Although the current model of genetic counselling is described as non-directive, this was not one of the topics most often mentioned in the guidelines. Of the 56 documents analysed, it was covered only in 21, whereas the autonomy of the counsellee was mentioned in 38 guidelines. When mentioned, non-directiveness was seen as a very important value of counselling, but some guidelines may have found it easier to agree on the importance of autonomous decision-making, a principle in medicine in general, than to discuss whether non-directiveness is, in the strict sense, even possible.

Avoiding discrimination on a genetic basis was seen by the guidelines as a remarkable challenge of genetics. Genetic counsellors were expected to understand the ethical questions posed by counselling, especially in the context of prenatal diagnostics. This is because counsellors may either promote or avoid discriminatory thoughts. The organisation of the disabled was especially worried that the lives of the disabled are defined in genetic counselling without the contribution of the disabled themselves.

The fear of discrimination in the context of genetics is so strong that it was conveyed in most of the guidelines. It was seen as important that genetic professionals actively avoid discrimination and eugenic thinking. For this, education in bioethics was considered desirable. Some theorists see, however, that the 'new eugenics' is essentially part of the practice of genetic counselling. Petersen writes that while the 'old eugenics' focused on populations and coercive control, the 'new eugenics' can be found in the counselling of individuals.[11,12] This 'new eugenics' has been seen to reflect professionals' values concerning diseases, disabilities and deviances as well as to involve a consensus of opinion on who should be born and who should not, even though the belief of objective information dominates the discussion.[13]

Contradictions in the guidelines

Although usually presented as containing no conflicts, some contradictions were apparent among the elements involved in genetic counselling. The guidelines expected the counsellor to provide objective information, to avoid being directive and to encourage independent choice. At the same time, they advised the counsellor to adapt the information needed to the patient's circumstances and to identify the particular situation of each patient. This means that the counsellor should select the appropriate information for each case and base the counselling sessions on the needs of each patient. Thus, in such cases, the information would not be objective, and the decision-making could be directed in some way. Objective information-giving and non-directiveness also do not conform with those guidelines advising counsellors to persuade counsellees to disclose the genetic information to family members.

Contradictions in the guidelines relate particularly to autonomous decision-making and non-directiveness. Non-directiveness has been considered problematic in many contexts before, and, even though it is the dominant model of counselling, it is not always supported as being the best one. Counsellees may not accept non-directiveness; they may experience it as sheer indifference, and may not want to make the decisions on their own. It has been argued that counsellors cannot act only as information providers, since they are asked to become facilitators in the decision-making.[14] Non-directiveness is based on the division between knowledge and values. Yet, this division is not easy. Experts choose the appropriate information and how to express it.[3] The genetic counsellor's values may come out, despite the effort to remain neutral. The institutional context is also said to imply the preferable course of action. Thus, it has been argued that non-directiveness limits the counsellor's ability to effectively serve the counsellee.[4]

Alternative models of counselling

As the currently dominant non-directive model is often seen as limited, new models of genetic counselling have been developed. In the shared decision-making model, counsellor and counsellee share information, and, in reaching a decision, the facts are integrated with the patient's emotions and personal values.[10] Charles *et al*[15] see that the advantage of this model is that it offers a potential middle way between the two polar extremes of the decision-making models: the traditional paternalistic model is characterised by the physician's dominance, whereas the informed decision-making model limits the role of the physician to transferring information, leaving all the decision-making control to the patient.

In addition, emotional support within counselling has been emphasised. Decruyenaere *et al*[16] offer a combination of information-orientated and psychological counselling. Jon Weil[4] argues that the central ethos of genetic counselling should be to bring the psychosocial component into every aspect of the work, instead of the emphasis on non-directiveness, as the fundamental role of genetic counselling is to help individuals to use the information of medical genetics to meet difficult situations. As the traditional definition of non-directiveness has been seen as limited, the concept has also evolved: it has been defined as a strategy directing counsellees to their own decisions, which requires interactive, skill-based counselling.[17,18]

Models that emphasise shared decision-making and psychological support can also be recognised in the analysed guidelines regulating genetic counselling. Most of them bring out the need to adapt to the counsellee's situation and to provide support when needed.

Diversity of the data

The three main elements of genetic counselling coexisted in the guidelines. They, however, were emphasised differently: while the professional organisations focused more on the practical issues, such as communication and the content of information, ethical boards and patient associations put the stress on ethical questions, such as autonomy and discrimination. As the guidelines reviewed had different purposes, the detail to which they addressed genetic counselling was extremely variable. They differed from each other by purpose and form, and dealt with different topics: some regulated a specific topic, such as a child-testing policy, while others discussed issues related to genetics on a more general level. Therefore, there are some issues that are not among the topics most often mentioned in these guidelines, such as predictiveness of a genetic test, even though this is often considered[19,20] to be one of the most problematic issues related to genetic testing. The topics most frequently mentioned in the guidelines rather describe ideal factors in each genetic counselling context, whether predictive, prenatal or diagnostic.

As there were some major differences in the data due to the purpose, form and topic of the guidelines, it can be questioned whether they were comparable at all. Since there were very few guidelines that were specifically written to address genetic counselling, it was necessary to include all the guidelines that mentioned it in the analysis. It can, however, be asked whether all the guidelines analysed were suitable for providing data. Another question is whether using QSR NUD*IST was the best method to identify the key issues. These considerations regarding the data and methods were taken into account while performing the study. The guidelines were reviewed critically and their differences recognised during analysis. QSR NUD*IST was used only as an assisting tool when coding the documents and searching for the main themes related to genetic counselling.

A uniform view of ideal genetic counselling

Despite the underlying contradictions between non-directiveness and adaptation to the counsellee's personal situation, the content of ideal genetic counselling was very uniform among the guidelines, whether they were of European origin or not. The same aspects were repeated from one guideline to another, forming a consistent vision of what may be seen as ideal genetic counselling.

This might be interpreted to mean that genetic counselling is, after all, not a very complex activity, but actually a rather simple encounter between the professional and the counsellee. It could equally mean that the difficult issues have not been examined properly as the uniform view is so strong. In this regard, Kerr and Shakespeare[13] argue that the international guidelines for genetics have created a global discourse of bioethics that is disappointing in its content. They hold that the regulations are conservative, uniform, narrowly defined, artificially divided and socially indifferent, and that they have a weak role as they seek to balance between different interests.[13]

The topics most frequently found in this survey have been interpreted to be the most important for genetic counselling, but this interpretation can be questioned, as the emphasis on certain aspects of genetic counselling is dictated by the particular topic being emphasised in the document, and thus the important topics are not always the same. It can also be argued that the most frequently mentioned topics are just the most obvious, and not always the most important ones. However, the presented topics came up in the analysis as general ideals of genetic counselling, not relating to any specific testing situation. Some topics that may be considered rather important as current challenges of genetic counselling, such as counselling in the contexts of common diseases, pharmacogenetics, and tests offered directly to consumers, were mentioned in only few guidelines. In general, the guidelines did not go into such detail. Another reason may be that these topics are still emerging, and it is too soon for a consensus. Thus, they may need more attention in future guidelines.

Conclusions

On the basis of an analysis of 56 guidelines, ideal genetic counselling, in the context of genetic testing, may be seen as consisting of an appropriately trained professional who understands genetics and its ethical implications well, relevant and objective information, ensuring of understanding of the patient, psychological support, informed consent, confidentiality of genetic information, dealing properly with the familial implications and potential discrimination caused by testing, and assuring autonomous decision-making by the counsellee. Most of these issues are covered by all the guidelines analysed, although the information, support and ethics may be stressed to different degrees.

The discussion on what ideal genetic counselling and its minimal criteria are based on is all in all more important as more genetic tests are being performed, and to an increasing extent non-specialised professionals provide the information when a genetic test is offered. As samples increasingly cross borders, particularly in the case of genetic tests for rare diseases, many molecular genetics laboratories would like to be sure that the ideals set in international guidelines are widely accepted and followed in different countries. If there is a desire to have a common view of what ideal genetic counselling is, the discussion needs to focus particularly on the differences and contradictions within and among the guidelines: (1) who may request a genetic test and provide genetic information in different testing situations? (2) How much education in the

psychological and ethical aspects of genetics should counsellors receive? (3) Should information be objective or adapted to the counsellee's situation? (4) Who should pass the information on to relatives at-risk? (5) Should autonomous decision-making be guaranteed, even if the patient does not want it? (6) Should genetic counselling include the perspectives of those who live with a genetic disease and their representative associations?

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References

1 Fraser FC: Genetic counselling. *Am J Hum Genet* 1974; **26**: 639–659.
2 Harper PS: *Practical Genetic Counselling*. London: Wright, 1988.
3 Clarke AJ: The process of genetic counseling. Beyond non-directiveness; in Harper PS, Clarke AJ (eds): *Genetics, Society and Clinical Practice*. Oxford: BIOS Scientific Publishers, 1997, pp 179–200.
4 Weil J: Psychosocial genetic counseling in the post-nondirective era: a point of view. *J Genet Couns* 2003; **12**: 199–211.
5 Borry P, Nys H, Goffing T, Dierickx K: *Genetic Testing and Counselling. European Guidance*, European Ethical-Legal Papers No 3, Leuven, 2007.
6 Barata PC, Gucciardi E, Ahmad F, Stewart DE: Cross-cultural perspectives on research participation and informed consent. *Soc Sci Med* 2006; **62**: 479–490.
7 Alexander J: Patients' feelings about ward nursing regimes and involvement in rule construction. *J Psychiatr Ment Health Nurs* 2006; **13**: 543–553.
8 Hueifang C, Shin-Ping T, Teh CZ *et al*: Lay beliefs about Hepatitis among North American Chinese: implications. *J Community Health* 2006; **31**: 94–112.
9 de Wert G: Cascade screening. Whose information is it anyway? *Eur J Hum Genet* 2005; **13**: 397–398.
10 Elwyn G, Gray J, Clarke A: Shared decision making and non-directiveness in genetic counselling. *J Med Genet* 2000; **37**: 135–138.
11 Petersen A: Counselling the genetically 'at risk': the poetics and politics of 'non-directiveness'. *Health, Risk Soc* 1999; **3**: 253–265.
12 Petersen A: The new genetics and the politics of public health. *Crit Public Health* 1998; **8**: 59–71.
13 Kerr A, Shakespeare T: *Genetic Politics. From Eugenics to Genome*. Cheltenham: New Clarion Press, 2002.
14 Shiloh S: Decision-making in the context of genetic risk; in Marteau T, Richards M (eds): *The Troubled Helix. Social and Psychological Implications of the New Human Genetics*. Cambridge: Cambridge University Press, 1996, pp 82–103.
15 Charles C, Gafni A, Whelan T: Shared decision-making in the medical encounter: what does it mean? (Or it takes at least two to tango). *Soc Sci Med* 1997; **5**: 681–692.
16 Decruyenaere M, Evers-Kiebooms G, Denayer L *et al*: Predictive testing for hereditary breast and ovarian cancer: a psychological framework for pre-test counselling. *Eur J Hum Genet* 2000; **8**: 130–136.
17 Kessler S: Psychological aspects of genetic counseling. XI. Nondirectiveness revisited. *Am J Med Genet* 1997; **72**: 164–171.
18 Weil J, Ormond K, Peters J, Peters K, Bowles BB, LeRoy B: The relationship of nondirectiveness to genetic counseling: report of a workshop at the 2003 NSGC annual education conference. *J Genet Couns* 2006; **15**: 85–93.
19 Davison C: Predictive genetics: the cultural implications of supplying probable futures; in Marteau T, Richards M (eds): *The Troubled Helix. Social and Psychological Implications of the New Human Genetics*. Cambridge: Cambridge University Press, 1996, pp 317–330.
20 Harper PS: What do we mean by genetic testing? *J Med Genet* 1997; **34**: 749–752.

Supplementary Information accompanies the paper on European Journal of Human Genetics website (http://www.nature.com/ejhg)