How to improve the quality of genetic counselling?

Kako izboljšati kvaliteto genetskega svetovanja?

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Ključne besede:
genetske preiskave, genetsko svetovanje, kakovost genetskega svetovanja

Key words:
Genetic testing, genetic counselling, quality of genetic counselling

Citirajte kot/Cite as:
Zdrav Vestn 2010; 79: 290–295

Prispelo: 30. sept. 2009,
Sprejeto: 5. nov. 2009

Abstract

Background: Genetic tests are used increasingly to improve the diagnostics and risk assessment. This has led to a growing need for genetic counselling, which helps patients understand genetics and genetic testing in their life situations. As genetic testing is often performed abroad, the need for harmonisation and quality issues of genetic testing process, including genetic counselling, on European level has been emphasized. Genetic counselling by appropriately trained professionals is indicated in many situations involving rare diseases: new diagnosis in the family, predictive or carrier testing and when considering prenatal or preimplantation diagnostics. On the other hand, comprehensive genetic counselling is usually not needed in situations related to risk profiling of common multifactorial diseases, pharmacogenetic testing or screening programs. Discussion on how to divide the work should continue. Developing tools for evaluating the quality of genetic services is underway as part of Eurogentest activities.

Conclusions: The need for genetic information including comprehensive genetic counselling is growing. Ample resources for genetic centres should be safeguarded and tools for evaluating the quality of genetic services should be developed. Simultaneously, all health care professionals should be appropriately trained to inform their patients about genetics and genetic testing and to refer them to comprehensive genetic counselling when indicated.

Izvleček

Izhodišča: Genetske preiskave se vse pogosteje uporabljajo za izboljšanje diagnostike in opredelitev tveganja. To je privedlo do naraščajočih potreb po genetskem svetovanju (GS), ki pomaga bolnikom pri razumevanju genetike in genetskih preiskav v njihovi življenjski situaciji. Ker se genetske preiskave večkrat izvajajo v tujini, se na evropski ravni poudarja potreba po harmoniziranju in izboljšanju kakovosti postopkov genetskih preiskav, vključno z GS.

Opredelitev problema: GS, ki ga izvaja ustrezno usposobljeno strokovno osebje, je indicirano v velikem števil primerov. Sem sodijo redke bolezni, pojav nove bolezni v družini, napovedno testiranje in testiranje pri prenašalkah oz. prenašalcih ter testiranje v okviru prenatalne ali predimplantacijske diagnostike. Po drugi strani pa celovito GS navadno ni potrebno v situacijah, ki so povezane z določenimi tveganji pri pogostejših multifaktorskih boleznih, pri farmakogenetskih preiskavah ali splošnih presejalnih preiskavah. Razpravljanje o tem, v katerih primerih genetsko svetovati, naj se še nadaljuje. Razvijanje orodij za vrednotenje kakovosti GS že poteka kot del dejavnosti v okviru EUROGENTEST-a.

Zaključki: Potrebe po genetskih informacijah, vključno s celostnim GS, naraščajo. Sredstva za delovanje centrov za GS je treba načrtovati in nadzorovati. Razviti je potrebno orodja za vrednotenje storitev GS, hktrati pa je treba zagotoviti, da bodo prav vsi zdravstveni strokovnjaki, ki se s tem ukvarjajo, primerno usposobljeni za informiranje svojih bolnikov o genetiki in genetskih preiskavah, in da bodo vedeli, kdaj je dejansko treba napotiti ljudi na celostne genetske preiskave.

Introduction

Molecular genetics has offered new, effective tools to find out the exact genetic etiology for a growing number of diseases. This has led to a growing need for clinical genetics services. Consultations of the genetic specialists help other clinicians in diagnosing rare diseases and choosing which genetic tests to use for their patients. Genetic tests improve the diagnostics and risk assessment. The role of genetic counselling is to help patients understand genetics and genetic testing in their life situations. A wide variety of different professionals are involved in this work: clinical geneticists, genetic nurses and counsellors, laboratory geneticists and all the diverse medical specialists. Adding to the complexity, samples for genetic testing often cross borders and, increasingly, also patients and families may seek genetic diagnostics and counselling from other countries. Recently, the need for harmonisation, coordination and quality issues in genetics, including genetic counselling, has often been mentioned as an important public health goal in Europe.[1-3]

What is genetic counselling?

The most widely accepted definition of genetic counselling was originally published by FC Frazer in 1974 and later slightly amended by BB Biesecker and KF Peters in 2001.[4,5] Recently, EuroGentest, a five-year Network of Excellence funded by the European Commission,[6] while creating European recommendations for genetic counselling related to genetic testing, used a slightly modified version of this definition which reads 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:

- understand the medical facts of the disorder;
- appreciate how heredity contributes to the disorder and the risk of recurrence in specified relatives;
- understand the options for dealing with the risk of recurrence;
- use this genetic information in a personally meaningful way that promotes health, minimizes psychological distress and increases personal control;
- 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
- make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder."

If this definition is taken literally, genetic counselling is a medical act performed by a highly specialized health care professional. These professionals have had a training, which could be assumed to guarantee sufficiently high quality of genetic counselling. In practice, however, there often are obstacles leading to a less satisfactory end result. This paper discusses some of these obstacles with the aim of suggesting solutions to overcome them and improve the quality of genetic counselling.

When is genetic counselling needed?

The following bullet points briefly present the need for genetic counselling in different clinical situations. Depending on the context, the disease in question and wishes of the individual and his or her relatives, there may be different needs. This paper focuses on average situations, realising that there may be exceptions in each category.

- When *a new diagnosis of a (rare) genetic disorder* has been set, the patient and his or her family usually need genetic counselling. Even if the disease has appeared in the near family before, the new diagnosis often creates new questions and genetic counselling should be actively offered.
- In case of a rare disease, even if it is non-genetic, or if *the diagnosis and thus the etiology remains unknown*, genetic coun-

selling may clarify the situation to the individuals involved and thus it is a good practice to offer counselling to them.

- If a healthy relative at high-risk for a specific later-onset monogenic disorder considers taking *predictive testing* , pre- and post-test counselling has to be offered. Predictive testing means a situation, where the mutation in the family leads to the disease or a considerably high risk for the disease (like in high-risk familial cancers). Even if the family has previously been counselled, further pre- and post-test genetic counselling is necessary to deeply discuss the implications that this kind of testing may have for the individual and the family. The counselling may be combined with psychosocial evaluation and support.
- Similarly, if *carrier testing* is envisaged, pre- and post-test genetic counselling needs to be offered. A carrier test means a genetic test that detects a gene mutation that will generally have limited or no consequence to the health of that individual, but 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).
- Always when *prenatal or preimplantation diagnostic genetic testing* is being planned, pre- and post-test genetic counselling for the prospective parents needs to be offered.

Even though genetic professionals widely agree about the need for "real" genetic counselling in the above-mentioned situations, patients and families are not always referred to a genetics unit. Many physicians representing other specialties do not comprehend the difference between genetic counselling and just shortly informing the patient, among other medical facts, that the disease is hereditary or that a genetic test may be considered. Patients themselves may not be aware of genetic services and thus are not able to ask for genetic counselling.

When would less comprehensive genetic information be sufficient?

Patients and families have different needs for comprehensive genetic information. Some may very well be satisfied with a short description of the heredity by any medical doctor or nurse while others seek for extremely timely and detailed information or need much support. However, it is generally agreed that there are situations related to genetic diseases or gene tests in which comprehensive genetic counselling cannot be considered very useful to the patient or offering counselling to all such patients would in practice be impossible. A list with examples of such situations is presented below.

- One example is *suspicion of a genetic diagnosis* and testing to confirm or rule out this suspected diagnosis. For instance, an infant with recurrent infections is often screened for cystic fibrosis mutations but a pre-test genetic counselling for the family at this stage might be too early. Instead, if the diagnosis is confirmed or strongly suspected, genetic counselling should be offered.
- Another example is a patient with *any common multifactorial disease*. In this situation genetic counselling is not necessary and can seldom add much to what the other specialists (for instance cardiologists, allergologists, diabetologists etc) can offer.
- Also, genetic counselling is not indicated if *pharmacogenetic testing* is being planned to evaluate genetic susceptibility for adverse drug reactions or efficacy of a drug treatment. They are ordered mainly by specialists other than clinical geneticists; proper genetic counselling by a genetic specialist might be needed only if the results have other implications for the person tested and his/her near relatives, than the decisions about drug treatment.
- *Genetic screening* means testing which is not targeted at high risk individuals or families but is systematically offered to the general population or a part of it

(e.g. newborns, young adults, an ethnic group, etc.). The need for and organisation of genetic counselling in genetic screening programmes[7] has been discussed elsewhere. Briefly, pre-test information and post-test information has to be an integral part of the genetic screening programs, 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 for a rare disease, as a result of screening, should be offered genetic counselling.

Do we need "ideal" genetic counselling?

According to its definition, genetic counselling aims at giving the counsellee knowledge and support to overcome difficult life changes and decisions. It requires specific experience and ample time from the professional. It is also very demanding for the patient or counsellee as he or she is expected to try to understand complicated medical and biological facts to be able to base future decisions on information that has really been understood. While all this is somewhat idealistic – the counsellor may lack time to go really deeply into the case and the counsellee may be unable to fully understand the message for emotional or intellectual reasons – it is the true aim widely shared in different guidelines and also among the professionals.[8]

In other fields of medicine, we are often quite satisfied with more pragmatic approaches. We understand that a patient cannot be carefully "pre-test counselled" before every thorax X-ray or gastroscopy, to prepare him or her to possible unexpected findings. We also don't expect (all) patients to fully understand why a specific antibiotic treatment was chosen or why we recommend physical treatment after an operation; we simply hope that the patient believes the doctor and obeys the recommendations.

The reason for the ambitious goal to assist the counsellee in achieving full understanding of the situation in genetic counselling is that the decisions made during or after the counselling are only sometimes "medical" but instead highly personal involving partnership, family planning, continuing or terminating pregnancy, future prospects related to education and profession etc. Thus, such decisions can be made only by the counsellee, even though medical and legal aspects have to be taken into account as well. In addition, clinical genetics as a rather new discipline suffers from lack of long experience of possible implications of, for instance, its predictive approaches and thus tries to create optimal services to prevent any adverse effects.

What happens during an average genetic counselling session?

Genetic counselling usually starts by the counsellee contacting or being referred to the genetics clinic where some preliminary preparations (asking family history, collecting consents and hospital files from relatives concerned) are usually made before the first visit, which in less complicated cases may remain the only visit to the clinic. The counselling session may take from 30 minutes to a few hours. Afterwards, there may be genetic testing or other additional investigations, collecting more family history and, in case of rare diseases, discussion about the case with other colleagues at the clinic or elsewhere. There may be another (or more) counselling session(s), and finally the content of the counselling is summarised in a letter to the counsellee; this usually comprises one or two pages. The letter is also sent to the patient's attending physicians, if the counsellee so wishes. When the counselling concerning the individual is completed, the question of whether to inform the relatives is usually discussed and this may lead to cascade counselling of further family members.

Genetic counselling may be considered expensive, as it is time consuming. However, as an individual or a family does not need genetic services often, the total cost for the health care will not be very high. Actually, genetic counselling can save money as

it may help to find the correct diagnosis and stop other diagnostic investigations, it may give the family information, which helps them to cope with the situation, and it may help the physician to treat the disease more adequately.[1] In several European countries, for instance the Nordic countries and UK, genetic counselling is part of public health care and the cost for the patient is very low. In some countries it may also be offered by private health care providers or by insurance companies and the costs for the patient may vary.

How to evaluate the quality of genetic counselling?

Evaluating the success and quality of genetic counselling is complex not only because of different approaches to genetic counselling, but also because it is difficult to define adequate outcome measures that are compatible with the aims of genetic counselling. How well do patients recall information? How has genetic counselling altered their plans? How have they in practice chosen to act on the basis of the information received? How satisfied are they with the process of genetic counselling? Have they experienced improvement in perceived personal control? Information, reproductive plans and reproductive behaviour cannot be considered as simple numerical measures of success or effectiveness of genetic counselling. Thus, some argue that a typical audit-approach for assessing the success or effectiveness of genetic counselling is not feasible and could give rise to misleading conclusions.[1]

Based on all this, EuroGentest Unit 3 (Genetic services) has created an instrument for evaluating the quality of genetic counselling, which can be applied in self assessment across different systems of genetic healthcare. It was developed in expert group workshops, including patient representatives, and discussed in an open workshop during ESHG Conference 2008. The decision was to emphasize the process instead of the outcome. The instrument consists of a set of standards and potential measurable outcomes for genetic counselling, including items from waiting times and physical clinical environment to access to peer support and continuing professional education, supervision of junior staff, and the actual communication with counsellees. The instrument, which is presently (July 2009) in pilot phase, can be approached at www.eurogentest.org.[6]

Are there ample resources in the genetics clinics?

The situations where genetic counselling is needed are becoming more and more frequent. Especially familial cancers have created a growing workload for genetic clinics. Also, prenatal diagnosis or detecting carriers has become more common as new genes and mutations have been increasingly identified. In many countries, this has led to lack of trained professionals to perform all the genetic counselling that is needed.

Genetic counselling is performed by genetics specialists (clinical geneticists and genetic counsellors/nurses) and these are available only in a small number of genetic centres, usually university centres. This leads to the unfortunate fact that, at least in sparsely populated areas, patients, families and counsellees may have to travel long ways to attend a genetic counselling session. The benefits of this kind of organisation, however, outweigh this drawback as genetics clinics may thus have larger teams with possibility to share experience and even subspecialize in a specific field within the clinic (e.g. cancer genetics). At present, a personal counselling session where the patient and counsellor are physically together is preferred to counselling by telephone or other remote methods.

There are some approaches aiming at dividing the work between genetics specialists and other health care professionals so that genetic professionals could concentrate on more complicated tasks in the field of genetic counselling. For instance, in the sensitive field of preimplantation genetic screening and testing, the professional societies in Europe have discussed and agreed upon recommendations on how to best divide the work.[9] In Finland, we have built a network

of specifically trained nurses in clinics of Finnish Cancer Society all over the country. Their task is to take family history and try to detect cases with suspected familial cancer to be referred to genetic specialists in the five university hospitals in Finland. Thus, these nurses do not need a very thorough training in genetics but only as much as is needed for detecting familial cancers. A somewhat similar approach was studied in the UK and found to be acceptable and cost-effective.[10] Similarly, specifically trained nurses have important roles in some screening programs, including maternity care clinics.

Independent of the organisation of the health care system, at least the very rare genetic diseases always require special attention because their rarity creates problems in suspecting and diagnosing them, having the experience for treatment and follow up as well as in genetic counselling of the family and relatives. Diagnostic consultations as well as genetic counselling in rare diseases are tasks for which the genetics clinics should be responsible and have sufficient resources.

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