

Have patients' opinion been asked? A review of 102 studies from the years 1997–2007 on patient perspective of genetic counselling services in genetic testing situations

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Background

EuroGentest Network of Excellence (www.eurogentest.org) aims at improving the quality of genetic testing. As patients' understanding of the results and consequences of the test is an integral part of genetic testing, EuroGentest also aims at improving the quality of genetic counselling services associated with genetic testing, across Europe. For this purpose, recommendations for genetic counselling in connection with different testing situations have been issued and instrument(s) to measure the quality of genetic counselling services are being developed.

To achieve all this, the group assigned by EuroGentest for this task has collected and analysed international and European non-national guidelines and policies related to genetic counselling, as well as some relevant national recommendations and other documents.¹ In addition, legislation related to genetic counselling in EU countries has been collected.² Data has also been collected with the help of surveys.³ Finally, it was considered essential to investigate also the experiences of patients and other clients on genetic counselling in connection with testing situations.

Objective

The aim was to find out what kind of research has been done on patient perspective on genetic counselling. Further, our purpose was to determine the possible issues that need more research. To achieve this, we reviewed studies on patients' opinions and experiences about genetic counselling related to genetic testing published in the period 1997–2007. In this paper we summarise the results of this review.

Methods

Data sources

At first, we searched the PubMed database for articles on genetic counselling related to genetic testing from patient's perspective. Our purpose was to determine the issues explored in research. On the basis of identified issues, we then selected main topics for this review: patients' expectations of genetic counselling, their experiences of genetic counselling, and their knowledge, after genetic counselling, about the issues discussed during the counselling session.

The final search was conducted on the databases PubMed, Cinahl, and PsycINFO for literature from 1997 to June 2007, using the terms *genetic counseling/counselling* in combination with the following terms: *counselee/counselee*, *expectation(s)*, *perception(s)*, *needs*, *satisfaction*, and *knowledge*.

Inclusion criteria

Studies were eligible for inclusion if they were published in English; if they included individuals who were referred for or had received genetic counselling related to genetic testing, or individuals accompanying those referred at the counselling sessions (e.g. parent or spouse); and if they examined at least one of the following topics: expectations of genetic counselling, experiences of genetic counselling, or knowledge after genetic counselling. In addition to actual counselling, various aspects closely related to genetic counselling were considered (e.g. physical environment, waiting times, or summary letter).

Papers were excluded from the review if the study was not related to such genetic counselling situations where genetic testing was actually offered, if genetic counselling was conducted in the context of a genetic screening programme, or if they were reviews. Studies dealing exclusively with prenatal genetic counselling were excluded if the reason for referral for counselling was not a genetic disorder in the family or a positive/ambiguous prenatal diagnostic test result (i.e. the reason was advanced maternal age or abnormal maternal screening result).

As the number of the studies included in the review was large, we used the statistical software package SPSS (version 14.0) as a tool to assist the analysis.

Results

We identified 102 studies that examined patients' expectations of genetic counselling related to genetic testing, their experiences of genetic counselling, or their knowledge after genetic counselling.

35 studies were published in genetics journals, 26 in counselling journals, 22 in oncology journals, seven in psychology journals, four in public health journals, three in nursing journals, two in internal medicine journals, and one each in neurology, pediatrics and medical ethics journals.

65 studies included authors from the discipline of genetics, 52 from the discipline of public health, 45 from the discipline of oncology, 42 from the discipline of psychology, 12 from the discipline of social sciences, 11 from the discipline of pediatrics, eight from the discipline of nursing, six from the discipline of surgery, and four from the discipline of obstetrics & gynecology. 17 studies included authors from various disciplines, such as biology, biostatistics, internal medicine, and economics. We could not identify representatives of patient organizations or other patient groups from the lists of authors.

33 studies were from the United Kingdom, 27 from the United States, 14 from Australia, nine from the Netherlands, four each from Canada and Israel, three each from Sweden and France, and one each from Brazil, Finland, Germany, India and Norway.

Seven studies were published in 1997, nine in 1998, five in 1999, 11 in 2000, eight in 2001, 11 in 2002, 10 in 2003, nine in 2004, 16 in 2005, 12 in 2006, and four in 2007.

The research method used was in 39 studies posted questionnaire, in 16 studies questionnaire completed at the clinic, in 19 studies interview, and in two studies group interview. 26 studies included both questionnaires and interviews. 13 studies compared different counselling methods.

The number of research participants ranged from 6 to 833. The mean number of participants was 120.34. In 30 studies research participants were relatives at risk, in four studies affected, in four studies parents, in one study support persons, in one study spouses, and in one study pregnant women. 56 studies included combinations of various types of participant.

101 studies focused on counselling sessions that had been performed in genetic counselling clinics. In 39 of these studies counselling sessions were performed specifically in familial cancer clinics. One study did not mention at all where the patients received genetic counselling.

In 14 studies genetic counselling was provided by clinical geneticist, in 12 studies by genetic counsellor/nurse, and in one study by other specialist. In 33 studies counselling sessions were given by more than one kind of above-mentioned professionals. In eight studies patients received counselling via device (computer program, pamphlet, audiotape of the counselling session, real time videoconferencing technology), and in five of these studies also from counsellor.

Great majority of the studies related to cancer (n=72), three to cystic fibrosis, two to chromosome disorders, one to Huntington disease, one to hemochromatosis, and one to Duchenne muscular dystrophy. 18 studies concerned combinations of various diseases.

The type of genetic testing which the counselling situations concerned was in 31 studies predictive testing, in eight studies diagnostic testing, in three studies prenatal testing, and in two studies carrier testing. In 51 studies genetic counselling concerned various types of testing.

Three studies specifically addressed the question whether patient perspective to genetic counselling was affected by the gender of the patient.

Patients' expectations were explored in 40 (39%) studies and experiences in 72 (71%) studies. The table below presents the percentages of studies assessing patients' expectations and experiences of different aspects of genetic counselling.

STUDIES ASSESSING EXPECTATIONS (n=40)	Percentage of studies
Topics about information - e.g. general information, information about different matters	90 %
Topics about quality - e.g. quality of information, atmosphere, communication style, procedural aspects	30 %
Topics about procedures - e.g. access to genetic testing or screening, examination by doctor	35 %
Topics about advice - e.g. advice about different matters, decision-making preferences	38 %
Topics about the benefit of others - e.g. to help family members, to help scientific research	23 %
Topics about psychologically positive outcomes - e.g. reassurance, emotional aspects	40 %
Others	43 %
STUDIES ASSESSING EXPERIENCES (n=72)	Percentage of studies
Topics about information - e.g. satisfaction with general information, satisfaction with information about different matters	58 %
Topics about quality - e.g. usefulness, quality of information, atmosphere, procedural aspects, evaluation of counsellor, evaluation of facilities	92 %
Topics about procedures - e.g. access to genetic testing or screening, examination by doctor	7 %
Topics about advice - e.g. advice about different matters, evaluation of directiveness	15 %
Topics about psychologically positive outcomes - e.g. reassurance, helped cope better, alleviation of guilt	46 %

Patients' knowledge was explored in 48 (47%) studies. The table below presents the percentages of studies assessing patients' knowledge, after genetic counselling, about the issues discussed during the counselling session.

STUDIES ASSESSING KNOWLEDGE (n=48)	Percentage of studies
General knowledge	50 %
Questions about own risk of developing the disease	46 %
Questions about population risk of developing the disease	13 %
Questions about other risk information - e.g. risk factors, family members' risk	25 %
Questions about inheritance	17 %
Questions about procedures - e.g. genetic testing, surveillance	10 %
Questions about test results	8 %
Questions about condition	4 %
Questions about treatment	4 %
Questions about family implications	2 %
Questions about future planning	2 %
Questions about scientific research	2 %
Questions about contacting other centres	2 %
Questions about reason for referral	2 %
Questions about healthcare practitioner knowledge	2 %

Discussion

Genetic counselling is usually understood as a communication process between an appropriately trained person and the client(s), as defined by Frazer.⁴ In addition, patients may get less comprehensive information by health care professionals from other fields of medicine, especially before and after diagnostic genetic tests in symptomatic individuals. In the reviewed studies "genetic counselling" had been performed in genetic units in all studies with the exception of one study mentioned above. Thus the great majority of the studies can be deduced to have evaluated "real" genetic counselling and not pre- and post-test information provided by non-geneticist professionals.

There is a consensus among clinical geneticists that many of the genetic testing situations, especially those predicting future illnesses, are so complex that comprehensive genetic counselling is needed. The satisfaction of patients/clients to such genetic counselling services has been investigated, also in the studies review in this paper, and these studies have usually revealed high satisfaction. According to our search, patient perspective to the less comprehensive information that is given in diagnostic situations and when the aim is mainly to rule out a possible diagnosis has not been evaluated.

The studies included in our review considered particularly patients' expectations about information, and patients' experiences of the information received and of the quality of

different aspects of their counselling sessions. Considering patients' knowledge, the most explored single topic was their own risk of developing the disease in question. However, as none of the studies initiated from patient organizations or other patients groups, some issues that could be very significant to patients may not have been covered at all in the studies.

One possible bias in our reviewed papers may be that most of the studies related to cancer. This is apparently because of the time period 1997–2007 that we investigated. The breast cancer genes *BRCA1* and *BRCA2* had been cloned in 1994⁵ and in 1995⁶, and hereditary non-polyposis colon cancer genes *MLH1* in 1994⁷ and *MSH2* in 1993⁸ and thus genetic testing for cancer predispositions was a growing part of the work of genetic counselling clinics during our research period. Predictive testing of monogenic adult onset disease (e.g. Huntington's disease) was already more established and patient perspective related to those tests had been investigated earlier. As people generally are well aware of cancer as a disease and detection of predisposition to familial cancer leads to follow up programs, the situation is very different when compared to hereditary neurodegenerative diseases. Thus patients may have different expectations and fears concerning the counselling process. Comparison between studies concerning cancer and studies concerning other diseases is not possible based on the papers in this review because the number of the latter was not adequate for this.

¹ www.eurogentest.org/web/info/public/unit3/summaryofguidelinesMay06.xhtml,

www.eurogentest.org/web/info/public/unit3/final_recommendations_genetic_counselling.xhtml

² www.eurogentest.org/web/info/public/unit3/regulations.xhtml

³ www.eurogentest.org/web/info/public/unit3/regulations_genetic_counselling.xhtml

⁴ Frazer FC. Genetic counselling. *Am J Hum Genet* 1974; 26: 636–661.

⁵ Miki Y, Swensen J, Shattuck-Eidens D, Futreal PA, Harshman K, Tavtigian S, Liu Q, Cochran C, Bennett LM, Ding W, Bell R, Rosenthal J, and 33 others. A strong candidate for the breast and ovarian cancer susceptibility gene *BRCA1*. *Science* 1994; 266: 66–71.

⁶ Wooster R, Bignell G, Lancaster J, Swift S, Seal S, Mangion J, Collins N, Gregory S, Gumbs C, Micklem G, Barfoot R, Hamoudi R, and 29 others. Identification of the breast cancer susceptibility gene *BRCA2*. *Nature* 1995; 378: 789–792.

⁷ Papadopoulos N, Nicolaides NC, Wei YF, Ruben SM, Carter KC, Rosen CA, Haseltine WA, Fleischmann RD, Fraser CM, Adams MD, Venter JC, Hamilton SR, Petersen GM, Watson P, Lynch HT, Peltomaki P, Mecklin JP, de la Chapelle A, Kinzler KW, Vogelstein B. Mutation of a mutL homolog in hereditary colon cancer. *Science* 1994; 263: 1625–1629.

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