Original Paper

Eur J Hum Genet 1994;2:96–102

Counselling following Diagnosis of Fetal Abnormality: A Comparison between German, Portuguese and UK Geneticists

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Abstract

The principle of non-directiveness in genetic counselling is embraced by all relevant professional bodies. Little is known about the extent to which it is endorsed by geneticists, or incorporated into their clinical practice. The aim of the current study is to document how geneticists in three European countries, Germany, Portugal and the UK, report counselling women at risk for having children with a range of conditions. While geneticists in all three countries reported counselling in a largely non-directive style, this varied both across genetic conditions and between countries. German and Portuguese geneticists were significantly more directive than UK geneticists, although they differed in the way in which they were directive. German geneticists were more likely to encourage continuation of pregnancies, while Portuguese geneticists were more likely to encourage termination of affected pregnancies. There was no strong consensus on approaches to counselling for any of the genetic conditions, defined as agreement between 70% of all three groups of geneticists. Despite strong professional codes of non-directiveness, geneticists report being somewhat directive in some counselling situations. Future research needs to focus on what geneticists are trying to achieve in genetic counselling, how they actually counsel, and with what effects.

Key Words
Counselling
Fetal abnormality
Geneticists
Germany
Portugal
UK

Received: August 12, 1993
Revision received: November 30, 1993
Accepted: December 29, 1993

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1018–4813/94/
0022–0096$5.00/0

The principle of non-directiveness in genetic counselling is embraced by all relevant professional bodies [1]. Little is known about the extent to which geneticists agree with this stance, or incorporate it into their practice.

The emphasis in genetic counselling has altered over the past 40 years from what Kessler [2] referred to as content-oriented counselling to person-oriented counselling. The purpose of genetic counselling has been variously described, but all definitions identify two key roles: the communication of factual information, and helping couples in their decision-making. Objectives of genetic counselling that have been put forward include reducing reproduction in those at high risk for recurrence of genetic disorder [3], and improved quality of life for the families that seek help [4]. Given the history of eugenics in this century [5, 6] there is an increasing awareness of the importance of assessing the success of counselling in terms of enhancing quality of life rather than in terms of reducing incidence of disease [7].

The manner in which counselling is conducted is also included in some definitions. Emery [8] views non-directiveness as a defining characteristic of genetic counselling, stating that: 'There is no place for directive counselling' (p. vii). To help people make decisions that are best for themselves, it has been argued that genetic counselling should always be non-directive [9]. Such an emphasis upon patient autonomy or non-directiveness has become emblematic of good genetic counselling. In part this probably reflects an attempt to distance current genetics from past abuses, in Nazi Germany, and to a lesser extent, in the UK and USA [5, 6].

Although there are many definitions of genetic counselling and many views as to how it is best conducted [10], there is little research documenting what counsellors describe themselves as doing during the counselling process. The aim of the current study is to describe how those practising genetic counselling in three European countries, Germany, Portugal and the UK, report counselling for a range of genetic conditions.

Methods

Samples

Medically and non-medically qualified geneticists were studied in each of three European contries: UK, Portugal and Germany. These countries were chosen in order to represent both northern and southern Europe. In the UK the geneticist sample was obtained from lists of geneticists employed in the main genetics centres in England and Wales, and the mailing lists of the Association of Clinical Cytogeneticists, and the Genetic Nurses and Social Workers Association. The response rate was 61% (137/225). In Portugal, a list of geneticists was obtained from the 4 hospitals in the country where geneticists are employed. The sample of 85 geneticists included both clinical and laboratory-based professionals. The response rate was 53% (45/85). In Germany, the sample of 230 geneticists was taken from the membership of the German Association of Human Genetics, whose membership spans former East and West Germany. Both clinical and laboratory-based geneticists were included. The response rate was 61% (140/230).

Clinical Experience and Counselling

The different samples in the three countries varied in the number of patients they saw a week. In the UK, 61% saw more than three per week; in Germany, this figure was 37%, while in Portugal it was only 17% ($\chi^2 = 67.01$; d.f. = 8; $p < 0.001$). The groups also differed in length of time since qualifying. Mean lengths of time were 17.2, 10.7 and 6.5 years for the UK, Germany and Portugal, respectively, (one-way Anova: $F(2) = 29.8$; $p < 0.000$).

Using direction and directiveness as dependent variables, two multiple regressions were conducted to determine the associations between clinical experience and approaches to counselling. The independent variables were: amount of patient contact, years since qualifying, gender, age, religion, and country. For directiveness, the only variable to enter the equation was country, which accounted for 9% of the variance in directiveness ($F = 23.5$; $p < 0.000$). Three variables predicted the direction of directiveness, but in combi-

nation accounted for less than 10% of the variance: age (accounting for 3% of the variance); country (2%) and amount of patient contact (3%).

Measures

Geneticists' approaches to counselling were assessed from responses to a questionnaire based upon one developed by Wertz and Fletcher (personal communication). These were translated into Portuguese and German, and piloted in all three languages. Respondents were asked to state how they would counsel women found to carry a fetus with 1 of 17 conditions, varying in severity, age of onset, and type of disability. Response options covered non-directiveness, and varying degrees of directiveness. The five response options were:

(1) encourage parents to carry to term;

(2) try to be as neutral as possible, but overall convey more positive than negative aspects of the condition;

(3) try to be as neutral as possible, covering both positive and negative aspects equally;

(4) try to be as neutral as possible, but overall convey more negative than positive aspects of the condition;

(5) encourage termination.

A scale of directiveness was derived to provide a summary score for each respondent based upon their responses to all 17 conditions. A score of two was given when a non-directive stance to counselling was reported for any one of the 17 conditions. Stating a tendency to emphasise positive or negative aspects of a condition was accorded a score of one. Encouraging termination or the continuation of a pregnancy was accorded a score of zero. The total possible score for each respondent across all 17 conditions was therefore 34. The higher the score, the more non-directive the counselling approach.

Direction of directiveness was determined by deriving a mean score of counselling approaches across all 17 conditions for each of the health professionals. A mean score of zero suggests a tendency to counsel non-directively; a score above zero suggests a tendency to counsel towards continuing affected pregnancies; a score below zero suggests a tendency to counsel towards terminating affected pregnancies.

Procedure

Questionnaires were sent by post using mailing lists of the relevant professional organisations. Stamped addressed envelopes were included for returning completed questionnaires.

Results

Two aspects of approaches to counselling were considered: degree of directiveness and consensus about directiveness.

Directiveness

Two aspects of directiveness were assessed: the extent to which counselling was directive, and whether directiveness was towards continuing or terminating affected pregnancies.

Geneticists in all three countries reported approaching counselling in a broadly non-directive style. Mean scores and SD for the UK, Portugal, and Germany were: 26.2 (6.6); 21.2 (6.4); and 21.1 (8.0), respectively. Geneticists in the UK were significantly more non-directive in approach than those from Germany or Portugal (one-way Anova: $F(2) = 17.21$, $p < 0.0001$; Tukey-B: UK vs. Portugal: $p < 0.05$; UK vs. Germany, $p < 0.05$).

The direction of directiveness evident in Portuguese and German geneticists differed. Mean scores and SD for UK, Portuguese and German groups were: −0.04 (0.4); −0.17 (0.5); and +0.08 (0.5), respectively. A multiple range test showed that the differences between the groups lay between German and Portuguese geneticists, and UK and German geneticists: Portuguese geneticists were significantly more likely to counsel in favour of terminating an affected pregnancy; German geneticists were significantly more likely to counsel in favour of keeping affected pregnancies (one-way Anova: $F(2) = 5.25$, $p < 0.01$; Tukey-B: Germany vs. Portugal, $p < 0.05$; UK vs. Germany, $p < 0.05$).

Consensus

Consensus between groups and countries on the extent to which they report counselling non-directively (defined as agreement between 70% or more respondents [11]) Nippertot evident for any of the 17 conditions (table 1).

Table 1. Percentages of geneticists reporting counselling non-directively and directively across different conditions

	Counselling non-directively[1]			Counselling directively continuing[2]			Counselling directively terminating[3]		
	UK	P	G	UK	P	G	UK	P	G
n	137	45	139	137	45	139	137	45	139
Cleft lip	33	31	24	66	67	74	1	2	2
Open spina bifida	47	26	36	2	0	2	52	74	63
Closed spina bifida	65	52	40	25	45	44	11	2	16
Anencephaly	36	19	18	0	2	1	64	79	82
Cystic fibrosis	73	48	63	6	2	6	21	50	31
Sickle cell anaemia	77	70	68	8	5	14	15	25	18
Huntington's disease	67	59	52	1	10	20	32	31	28
50% Alzheimer's disease	73	61	48	17	34	42	10	5	10
Alzheimer's disease	68	58	47	9	25	33	23	18	30
Turner's syndrome	54	68	32	41	7	61	5	24	7
Down's syndrome	70	36	59	2	5	10	28	60	32
Klinefelter's syndrome	59	68	38	32	7	56	8	24	6
Achondroplasia	74	45	56	14	13	25	12	43	20
PKU	60	50	45	29	41	46	11	10	10
Haemophilia	67	62	54	20	14	36	13	24	11
APKD	66	66	50	28	20	42	6	15	8
DMD	50	31	52	2	2	20	49	67	28

UK = United Kingdom, P = Portugal, G = Germany.

1 Percentages of respondents using response option (3) 'try to be as neutral as possible, covering both positive and negative aspects'.

2 Counselling directively in favour of continuing affected pregnancies. Percentages of respondents using response options (1) 'encourage parents to carry to term' or (2) 'try to be as neutral as possible but overall convey more positive than negative aspects of the conditions'.

3 Counselling directively in favour of terminating affected pregnancies. Percentages of respondents using response options (4) 'try to be as neutral as possible, but overall convey more negative than positive aspects of the conditions' or (5) 'encourage termination'.

If the cut-off for consensus is lowered to 50%, then consensus is evident for 7 conditions: cleft lip, sickle cell anaemia, Huntington's disease, haemophilia, adult polycystic kidney disease, anencephaly and severe open spina bifida. The consensus for the first was to encourage continuing to term, for the next 4 conditions, to counsel non-directively, and for the latter 2 conditions listed, to encourage termination of the pregnancy.

Consensus, as defined by 70% agreement, was also examined within each country. Amongst UK geneticists consensus was evident for 5 conditions (cystic fibrosis, sickle cell disease, 50% risk of Alzheimer's disease, Down's syndrome, and achondroplasia), the consensus being to counsel non-directively. German geneticists shared a consensus on 2 conditions (cleft lip and anencephaly), the consensus being to encourage keeping the

pregnancy in the former, and to encourage termination in the latter. Amongst Portuguese geneticists consensus existed for 3 conditions (anencephaly, severe open spina bifida, and sickle cell disease). Consensus for a non-directive approach to counselling was evident for sickle cell disease. For the other 2 conditions, the consensus was to encourage termination ($\chi^2 = 1.76$; d.f. = 2; NS).

If consensus is defined as agreement between 50% of respondents within a group, then UK geneticists reach consensus on counselling non-directively on 14 of the conditions. Portuguese geneticists reach such a consensus on 10 conditions, and German geneticists on 8 ($\chi^2 = 8.73$; d.f.: 2; $p < 0.02$).

Discussion

Geneticists in all three countries reported approaching counselling in a largely non-directive style. This varied however both between countries and across conditions. German and Portuguese geneticists were more likely than UK geneticists to counsel directively. Amongst German geneticists, this was in the direction of encouraging continuation of pregnancies; amongst Portuguese geneticists, this was in the direction of terminating affected pregnancies. Strong consensus on approaches to counselling, defined as agreement between 70% of the three groups of geneticists, was not found for any of the 17 conditions. When lowered to 50%, consensus was evident for 7 of the 17 conditions. It was most frequently found amongst UK geneticists, and less so amongst Portuguese and German groups.

Conditions tended to vary from each other in several ways, such as the degree of disability, the nature of the disability (physical vs. cognitive), age of onset (early vs. late), and treatability (lethal vs. treatable). This study therefore does not allow us to draw any firm conclusions about the way each of these were weighed by geneticists as reflected in their were counselling approach. Nonetheless, the directiveness and direction in which geneticists reported counselling was in part related to the condition. Directiveness was an approach adopted by many in the face of either a condition that was lethal (anencephaly) or relatively minor (cleft lip). Non-directiveness was more often in relation to late-onset disorders (Huntington's disease and adult polycystic kidney disease), and those with a variable expression (sickle cell disease and haemophilia).

The differences observed between the approaches to counselling taken by geneticists in Portugal, Germany and the UK may be explained in several ways. These include the broad tradition within a culture, as well as more specifically its history of genetics [12]. Political and economic factors may shape counselling styles in different countries. For example, the economic burden of having a child with a disability may be greater in Portugal than the UK and Germany. Hence, counselling in Portugal may incline more towards terminating than continuing with affected pregnancies. German geneticists were more likely to counsel in the direction of keeping affected pregnancies, which may reflect a historically motivated sensitivity and caution with which Germans approach genetic testing.

It should be emphasised that the data in this study relate only to self-reports of counselling. A further note of caution concerns the diversity of the samples of geneticists in this study. In all countries they comprised clinical and laboratory-based geneticists, as well as some non-medically qualified genetic counsellors. This was because there are few clinical geneticists in Portugal, and laboratory-based geneticists often see patients. Both the types

and amounts of counselling experience amongst these professionals varied widely. There was little evidence in the study to suggest that clinical experience, as defined by years since qualifying and frequency of patient contact, influenced counselling. However, more refined measures may reveal differences attributable to clinical experience. The response rates in each country were similar (53–61%). Given that no reminders were sent, this is an expected response rate. Nonetheless, we do not know how representative respondents are of the total sample.

The comparability of responses to questionnaires depends critically upon the comparability of wording. Sometimes subtlety of meaning is lost in translations. This study was conducted as part of a 1-year research programme funded by the EC [13]. Given the time constraints under which this study was conducted, it was not possible to back translate the questionnaires. As a result, some of the differences between countries may reflect differences in language rather than real differences. Nonetheless, the study provides useful data which will form the basis of further research in this area.

The results of this study show that not all geneticists consider non-directive counselling the approach of choice in all situations. We do not know the short- or longer-term effects of counselling approach either upon parents' decisions or emotional well-being. There is concern that counselling may influence decisions inappropriately, undermining patient autonomy. Yet patients sometimes look to health professionals for more direct advice than guidelines suggest is appropriate. In a postal survey of 791 families who had received genetic counselling, 42% stated that they wanted the counsellor's opinion of what they should do, in addition to the facts [14].

It is not known what proportion of those facing decisions about termination following detection of an abnormality wish for guidance. Preferences for decision making to be shared with health professionals were lower amongst women following detection of breast cancer, as opposed to the detection of a benign lump [15]. It may therefore be that following detection of a fetal abnormality, particularly one about which parents know very little, they actively seek guidance from the health professionals providing counselling. Whether fulfilling such requests is helpful in the short or longer run awaits study.

In summary, the results of the current study illustrate broadly similar approaches to counselling for different genetic conditions in three European countries. The results also demonstrate that not all geneticists see non-directive counselling as the approach of choice in all situations. These results suggest that decisions about continuing or ending pregnancies with certain fetal abnormalities may be more strongly influenced by geneticists than is recommended in guidelines for clinical practice. Future research needs to focus on what geneticists are trying to achieve in genetic counselling, how they actually counsel, and with what effects.

Acknowledgements

We are grateful to the EC for funding this study, which was funded as part of the EC Human Genome Analysis studies on the ethical, social and legal aspects (ESLA). We thank Dr. Dorothy Wertz and Dr. John Fletcher for allowing us to use their questionnaire as the basis for our own.

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