

The Client's Perspective of Genetic Counseling—A Grounded Theory Study

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Previous studies of genetic counseling have mainly focused on outcomes defined by researchers or service providers, and have frequently related to changes in reproductive behavior and/or client knowledge. A longitudinal study of 43 families referred to a clinical genetic service was undertaken to ascertain client needs and expectations of the service, and to identify relevant outcomes from the clients' perspective. Semistructured interviews were conducted with each client, prior to and after genetic counseling. The transcribed interviews were analyzed using grounded theory. The need for certainty emerged as a powerful factor that motivated clients to pursue the genetic referral. The client's lay knowledge of the condition, satisfaction of the need for certainty, and the formation of a personalized relationship between the client and the genetics staff significantly influenced the central outcome, identified as a change in the client's psychological adaptation to the genetic condition in the family.

KEY WORDS: genetic counseling; need for certainty; lay knowledge; psychological adaptation to genetic condition.

INTRODUCTION

Genetic counseling is a relatively new field in health care, the first dedicated genetics clinic being established in the United Kingdom in 1946 (Carter *et al.*, 1971) and in the USA in 1940 (Kevles, 1985). The definition of genetic counseling published by the Ad Hoc Committee on Genetic Counselling, American Society of Human Genetics (1975) emphasizes that it is a process of communication

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that addresses the problems and challenges that accompany the risk of a genetic condition in the family.

Genetic counseling in the United Kingdom is provided by professional teams in regional genetic centers, funded by the National Health Service. The clinical teams consist of medical geneticists, and nonmedical genetic counselors. Genetic counselors have either a background in nursing, with appropriate additional training, or have completed a Master's degree in genetic counseling.

The outcomes of genetic counseling have been investigated by a number of researchers. However, this research has tended to be concerned with assessing changes in knowledge of the condition (particularly recurrence risks), and changes in reproductive behavior or intention as a result of the genetic counseling. As early as 1971, Carter *et al.* (1971) tried to assess the effectiveness of genetic services by studying recall of risk figures and postcounseling reproductive behavior in a cohort of 438 couples who had attended a genetics clinic about 3–10 years earlier. One striking aspect of this study is the researchers' assumption that parents who were judged to have planned their families appropriately after counseling (i.e., parents with high risk limited their family, those with low risk had further children) must have done so because they understood the information given to them. Only a year later, Leonard *et al.* (1972) started to discuss the burden of risk as a factor in the reproductive decision-making process, and more in-depth work by Lippman-Hand and Fraser (1979) confirmed it was not the risk figure that influenced family size, but the burden of risk to that family. Sorenson *et al.* (1987) showed in a study of couples seeking genetic counseling prior to starting a pregnancy that the couple's precounseling intentions were the most significant factor in their decision-making, while the risk information given in the genetics clinic was secondary. Work by Frets *et al.* (1990) indicated that both the couple's desire for children and their experience of the problems associated with the disorder were key factors influencing their family plans.

However, research using reproductive behavior and risk recall as outcomes of genetic services does not fully address the value of genetic services to clients. This is demonstrated by the findings of Rona *et al.* (1994) in a study that assessed accuracy of risk recall, reproductive intentions, and levels of anxiety after genetic counseling. Whilst only a third of couples correctly recalled the risk figure applicable to them, 84% were pleased to have had genetic counseling, indicating that risk recall may not be an important outcome to clients. Gagnon *et al.* (1996) found that the anxiety levels of women at risk of familial breast cancer were reduced after genetic counseling, regardless of the information obtained. It did appear therefore that clients derived some benefit from genetic counseling, although the nature of that benefit was unclear.

In order to assess outcomes, the needs and expectations of clients also need to be addressed. Michie *et al.* (1997) contributed to this discussion in a paper on the client's expectations. The response categories (information, explanation,

reassurance, advice, help in decision-making) were generated by providers of the service, rather than the users.

This paper describes a longitudinal study of genetic counseling from the clients' perspectives. As this study was primarily concerned with ascertaining the views of clients, it was important to choose a methodology that enabled clients to express themselves freely and to raise issues that were of relevance to them. Qualitative methodology is increasingly used in health services research, and has been used effectively in the area of genetics research (Armstrong *et al.*, 1998; Frets *et al.*, 1990; Lippman-Hand and Fraser, 1979).

Field-based methods enable the experience of the client to be heard in a more natural context, and the grounded theory method was chosen over other qualitative methods such as phenomenology and ethnography for several key reasons. Phenomenology has been criticized as enabling only "superficial narratives" of phenomena to be produced (Grbich, 1999), and there is some dispute as to whether development or integration of theory should be a feature of ethnographic studies (Hammersley, 1992), where the subjects' voices may be taken to be sufficient, without the imposition of researcher interpretation. The use of grounded theory enables the researcher to be guided and directed during the actual research process by the data, and was therefore suitable for use in an area of study in which there has been relatively little previous research. It also offered the opportunity to develop a theory concerning genetic services.

The aims of the study were (1) to enable clients to describe their own needs and expectations of the genetic counseling service prior to any contact with the staff of the service, (2) to assess the outcomes the clients perceived as important following genetic counseling, and (3) to develop a grounded theory describing factors that influence the outcomes of genetic counseling and showing the relationship between these factors.

STUDY METHOD

In order to follow the client through the process of the genetic counseling, from the referral to follow-up, a longitudinal design was chosen. This enabled the researcher to obtain data on the client's expectations, before these were altered by the client's contact with the genetic service, as well as providing the opportunity to assess how the perceived needs, expectations, and outcomes changed over the period of contact. Each subject was interviewed on three separate occasions (30–90 min duration each) using an interview schedule. The first interview was conducted prior to any contact between the client and the genetic service, to assess the client's needs and expectations of the service. The second interview took place 2–4 weeks after the episode of genetic counseling had been completed, and the issues explored included the extent to which the client's needs and expectations had

been met, and the client's perceptions of the outcome(s) of the genetic counseling process. The third interview, 6 months after counseling was used to assess both the stability of perceptions that clients reported after the consultation, and changes to the client's life that had occurred in the medium-term. Data was collected by means of audiotaped semistructured interviews, not only enabling the researcher to guide the conversation so that relevant issues were covered, but also enhancing the ability of clients to introduce issues spontaneously.

The grounded theory approach to data analysis and development of theory was devised by Glaser and Strauss (1967). However, following their early publications, the two authors followed divergent pathways with respect to some aspects of the method (Turner, 1981), and the analytical process which has been used in this study is that described by Strauss and Corbin (1998).

The Process of Analysis Using Grounded Theory

1. The area for study was identified as a broad topic. One study aim was to determine whether it was appropriate to develop an overarching theory relating to genetic services as a whole, and this would not have been achieved by restricting the study to particular client groups.
2. A preliminary exploratory study (not reported here) of 10 families was conducted. Initially, the interview schedule was unstructured, consisting of 5–6 broad topic areas, but as the study progressed it was evident that there were several strong recurring themes. A semistructured questionnaire was developed to enable these themes to be explored more fully with a further 43 families.
3. Interviews were analyzed during the data collection period, and the interview schedule was amended according to the emergence or nonoccurrence of themes.
4. The phenomena that were described by clients were grouped into categories. In the initial period of the study, every statement was categorized (open coding). The categories were grouped into more abstract themes (axial coding). As the analysis progressed there was a focus on those categories that appeared to be of greatest relevance to the developing theory (selective coding). Negative examples were noted and explored with clients, as well as confirmatory data.
5. All clients were interviewed for a second and third time, to enable all those previously interviewed to report their perceived outcomes. Again, categories and themes were extracted from all the transcripts, although the analysis was informed by the themes that had emerged from the data obtained at previous interviews.
6. Throughout the process of planning, data collection and analysis, detailed contemporaneous notes were kept by the researcher. The discussion of the

study and the grounded theory hypothesis were based upon the material in these notes.

7. Following the development of categories and themes, three independent researchers were asked to each code three different interview transcripts, using a number of codes defined by the researcher. This exercise was undertaken to try to ensure that the researcher's coding of the interview data was consistent.
8. The grounded theory developed gradually as conditions that produced the observed phenomena were sought throughout the course of the study. In accordance with the method described by Strauss and Corbin (1998), conditions that were both causal and consequential were explored, and the data examined repeatedly to derive any existing links between the various relevant conditions. Diagrammatic representations were used to demonstrate the interconnecting processes involved in genetic counseling from the client's perspective. Finally, a substantive grounded theory was devised. A report on the analysis was also sent to each client interviewed with an invitation to comment, to enable discrepancies or inconsistencies to be identified.

STUDY PROCESS

Over a period of 6 months, 107 families were referred to the local genetics service, and 73 eligible families were approached by means of a letter from the researcher, for inclusion in the study. Those excluded were clients less than 18 years of age, clients with serious learning or psychiatric problems, those requiring an urgent appointment (in which case a precontact interview was not possible), and clients who had undergone recent psychological trauma such as bereavement or already known to the researcher or the genetic service. Because of the exclusion criteria, some caution is needed when interpreting the results of the study with respect to clients who have recently undergone acute psychological trauma. The letter inviting families to participate was sent as soon as possible after the referral was received by the genetics department, prior to any contact from the clinical genetics team. Of the eligible clients, 45 families accepted the invitation, 2 were later excluded for ethical reasons. Table I shows a comparison of participants and those who declined. The study method did not enable the researcher to investigate reasons for nonparticipation, although several people who declined left messages to say they were too busy to commit the time. There was no attempt to focus upon particular groups of clients by condition. This was because the study was concerned with exploring whether there were common themes that might be relevant to genetic counseling overall, or whether families differed in their needs and expectations according to condition group.

Table I. Demographic Comparison of Participants and Nonparticipants

Variable	Families in study	Families who declined to participate
Age	Mean age 35.5 years Range 18–58 years	Mean age 36.11 years Range 20–53 years
Sex	Male 26.9% Female 73.1%	Male 16.7% Female 83.3%
Marital status	Married 73.1% Living as married 9.6% Single, never married 7.7% Divorced 1.9% Intending marriage 7.7%	Married 70.8% Living as married 8.3% Single, never married 8.3% Divorced 12.5%
Condition type	Mendelian 50% Multifactorial 13.5% Cancer 13.5% Chromosome 5.8% Delay/dysmorphism 17.3%	Mendelian 29.2% Multifactorial 12.5% Cancer 20.8% Chromosome 16.7% Delay/dysmorphism 20.8%
Who affected in family	Self 11.5% Family member 28.8% Child 48.1% Self and others 11.5%	Self 16.7% Family member 25% Child 50% Self and others in family 8.3%

A total of 43 families completed the first interview, 40 the second, and 38 completed all three interviews. In cases where a child was the primary subject of the referral (e.g. because a diagnosis was being sought), both parents were asked if they would like to be interviewed, and were interviewed together as a couple, although individual differences in their responses were noted and explored. In 9 of the 43 families both partners chose to be included in the study, and in 34 cases there was only one family member interviewed. There were therefore 52 individual participants overall.

After obtaining consent from the client, an appointment was made to interview the client(s) in his/her own home. Ethical approval was obtained from the relevant Local Research Ethics Committees before the study commenced.

Whilst the researcher was affiliated with the genetics unit concerned, during the period of the research she withdrew from all involvement in the provision of clinical service to these clients to maintain a strict barrier between the research and service domains. This was possible because of separate funding for the research that allowed the researcher to be released from clinical duties.

RESULTS

Demographic Profile of the Sample

A demographic profile of the sample is presented in Table I. The disease group categories are based on groups used by the South-West of Britain Clinical

Genetics Audit Group (1999) that have been developed over at least 8 years of clinical audit practice for analysis of data on clinical genetics services.

Comparison of Participants and Nonparticipants

When demographic data on clients who declined to be involved in the study was compared with the participant group there were no detectable differences between the groups, except in the condition categories (see Table I). The reasons for the differences are unclear and may simply be due to the small numbers.

Type of Contact with Genetic Service

Of the 43 families studied, 5 (11.6%) were seen by a genetic counselor only, and all of these contacts took place in the client's home. Thirty-six (83.7%) saw both the genetic counselor for a preclinic contact and a medical geneticist. One person (2.3%) saw a medical geneticist only. At the time of the study, it was the policy of the genetics unit to offer clients a preclinic home visit by the genetic counselor to obtain information that would assist preparation for the clinical consultation and to provide the family with information about the service.

The results herein are reported under the themes that emerged from the analysis. Figure 1 provides background information on clients quoted in the text.

Major Theme 1: Need for Certainty

Clients in this cohort appeared to have a general preference for certainty in a variety of areas of daily life. This was tested after the need for certainty had been identified as an important theme by means of several general questions about obtaining certainty (not reported here). Clients were also keen to obtain certainty with regard to the genetic condition, but there was frequently an identifiable trigger that had precipitated the genetic referral at that precise time in the client's life, for example a new diagnosis in a relative, or impending marriage (Table II). It was possible to identify specific issues related to uncertainty for the clients, which they hoped would be clarified.

Manifestations of the Need for Certainty

- i) Need to prepare for the future: The search for information to help the family prepare for the future was emphasized by the majority of families (51%). This was especially true if a child in the family was affected. For example, Georgia (S29) was concerned to obtain information about her daughter's skeletal condition.

Pseudonym	Condition	Main concern
Rachel	Mendelian condition affecting vision	Carrier status
Melanie	Child with learning difficulties	Diagnosis and prognosis for child
James	Mendelian neuromuscular condition	Risk to his adult children
Marilyn and Harry	Child with chromosomal abnormality	Prognosis for child
Maria	Familial cancer	Risk to herself and children
Josie	Mendelian neurological condition	Child might inherit condition
Gloria	Familial cancer	Risk to herself and children
Sarah	Mendelian neuromuscular condition	Risk to self and future children
Gail	Family history multi-factorial condition	Recurrence risk for future children
Martin	Mendelian neurological condition	Predictive test for self
Lydia	Familial cancer	Risk to self and arranging clinical surveillance
Vera	Mendelian recessive disorder and familial cancer	Risk to children of both conditions
Molly and Ben	Child with learning difficulties	Prognosis for child and recurrence risk
Marg and Simon	Child with learning difficulties	Diagnosis for child
Malcolm	Mendelian skeletal condition	Risk to future children
Julie	Mendelian musculo-skeletal condition	Diagnosis for self
Kirsty and Frank	Recessive condition in family member	Risk to future children
Lucy and Stuart	Child with X-linked condition	Prognosis for child

Fig. 1. Background information on clients quoted in the text.

S: . . . whatever we find out, it will help us tailor, basically, our services to her needs, we can best deal with her needs, the more we know about it the better we can help her.

I: . . . So is that the outcome you do hope for really that you will have more information that helps you to answer her needs.

S: . . . Yes provided, to give her the best quality of life that we can.

The need to prepare for the future was also expressed as the identification of genetic risk for themselves or their offspring for 24 (56%) of families studied, and as a request for prenatal testing made by 2 (5%) families.

- ii) Need to understand the present: Families who were unsure of the diagnosis were desperate to know the reason for their child's condition. This is clearly demonstrated by John's parents, Ben and Molly (S26). John had developmental delay and dysmorphic features.

S: Molly . . . it seems all we do is we're waiting for something.

I: Your life is kind of on hold?

S: Molly Yeah it is, definitely

I: And how long has it been like that?

S: Molly Since we've known,

S: Ben Since we . . . since he got referred to doctors and we realized something was up, . . .

I: And that's making it harder for you to sort of just get on with daily life.

Table II. Classification of Events that Triggered Referral

Category	Number of families in study	Notes/examples
Self or family members reaching a different life stage	10	E.g., affected child starting school or offspring starting to date
The diagnosis of a affected relative	10	In 5 cases the relative was the client’s child and further information was being sought
Planning to start or increase family	9	
Ongoing quest for diagnosis	5	All referred at suggestion of paediatrician
Planning marriage/cohabitation	5	
Alerted to possibility of screening for cancer	2	
Alerted to genetic basis of condition by media	1	
Death of an affected relative	1	
Total	43	

S: Molly Yeah once we, once, that’s what I said to Ben, as long, if they can give me a name then I’ll be alright, and I can say “Right, he’s got that, that’s it he’s got that and that’s the end of it,” let’s just get on with our lives and worry about it as it comes along, but not all this not knowing, god it’s awful, frustrating . . . waiting, waiting, waiting, waiting for something I don’t know what we’re waiting for, a miracle. That’s what it feels like, you’re waiting for something.

Certainty in dealing with the present was also the underlying reason for the request for clinical screening for cancer that was made by a further two clients.

Anticipated Worst Outcome

The majority of clients had emphasized their desire for certainty, whatever the news. This need for certainty was further tested by asking clients what they would consider to be the worst outcome of the referral. The responses fell into two categories, the first being uncertainty. Seven (16%) of the families felt that the worst outcome they could envisage would be to emerge from the genetic counseling process without the answer they sought, i.e. to be left with uncertainty. Julie (S45) was worried about being left with no firm diagnosis.

I: *What’s your worst case scenario?*

S: *Umm, it comes back and they say, you haven’t got Marfan’s but you have got something wrong and we don’t know what it is*

The second category could be summarized with the phrase “bad news.” The remainder of the cohort were afraid of either being told the prognosis was poor, that

their child had a shortened life expectancy, or that their children were at genetic risk. Janice (S24) had a daughter with learning delay.

S: Like I said if they come back and say it's some such and such and the life expectancy's shorter than an average life expectancy, that's all I'm worried about, you do hear, you know, people's worlds turned upside down cause they find out their child's got such and such and they're not expected to live, I mean at the end of the day when you have children you don't expect to bury them.

However, after citing the worst possible outcome, a number of clients stated that even though this was the worst news they could receive, they would still rather hear the news than remain ignorant.

In all there appeared to be 12 families who had obtained a measure of certainty which fulfilled their needs. Josie (S11) had the news that her daughter was at risk of Huntington disease:

S: I think basically really, it's a, put my mind at ease that I know more about it, because like I said, if anything did happen to Sean, at least I know that there is a chance that she could have it,

There were clients who felt that obtaining certainty had enhanced their lives. One such client was Lydia (S50), who relied heavily on the genetic counselor's certainty for reassurance:

S: . . . she was quite sure about it, she wasn't sort of . . . half telling me, or saying well I don't think you'll get it, she was quite sure about what she was saying, she was quite confident about it, and she brought books with her, and she showed me exactly how the pattern works, and how she could be sure that I wouldn't have any more risk than anybody else

Where families had not obtained certainty, for example where a syndromic diagnosis had not been made in a dysmorphic child, many families were able to rationalize the outcome, and felt some closure had been achieved by virtue of the fact that they had at least pursued another avenue of inquiry. These families obtained a type of certainty by finding out that the information they sought was not available, even to "experts." This appeared to give them peace of mind. However, four clients who previously felt they had a firm diagnosis that was thrown into doubt by the staff of the genetics service were extremely distressed. Maria (S25) explained how she felt when the geneticist said that her son's previous diagnosis was incorrect:

S: I'd accepted, I mean I've always known Stevie's had something the matter with him, and I accepted what they (pediatrician) told me, and I was quite happy with that, I coped with that, perfectly well, but now I'm sort of left, wondering, what on earth's going on, really

These data confirmed that in this study obtaining certainty was a primary factor in motivating clients to seek genetic counseling and in influencing their satisfaction with the outcome. Both positive and negative instances confirmed

the importance of this component, and the need for certainty became an integral concept in the overall theory.

Major Theme 2: The Personal Relationship Between Client and Counselor

The importance of the relationship between client and counselor became apparent when clients were asked for their opinion of the role of the genetics service. During the first round of interviews that clients had difficulty describing the role of a genetic service, largely because of their lack of knowledge of the genetic service. Prior to contact with the service, clients were particularly concerned with the practical issues such as where the consultations would be held, what type of staff they would see, and what information would be asked of them.

After their contact with the genetics service, the majority of those interviewed expressed the role in terms of what they individually had desired or received. For example, those primarily seeking a diagnosis felt that clinical genetics was primarily a diagnostic service, whilst those seeking a risk assessment focussed upon that aspect. However, several stressed the need for psychological support from the staff during the process. Martin (S15) put it this way:

I: What would you say then is the role of the genetic service?

S: Ahh, I think really to sort of bolster people's confidence I think, and always be there as a backup when they're actually needed, because there's very deep-seated feelings running through people, umm, when they are involved in something like this, they need somebody there that they can rely on, and they are very good at, you know they're there, and they're always there, they'll always help you if you need it

Marilyn and Harry (S17) cited provision of both technical information and counseling support as being part of the role, but felt the counseling aspects were most useful. They were emphatic about the need for emotional care from the genetics service:

S: Oh I think the counseling side is, is the most important thing of it, really

Many clients spoke of the need for support to be available, even if they felt they had not required it themselves. There was a general acknowledgment that the issues involved could be very sensitive, and that there was potential for clients to become distressed. This appears more likely when the family involved are coming to terms with new or different circumstances, as was the case for the two families quoted earlier. However, it was evident that families who were not particularly distressed also appreciated warmth and personal interest from the counselor. Several who spoke positively of their experience cited personal comments from the counselor as indications they were not just "part of the assembly line." Several people commented that the information they received from others, especially other family members, might be biased to protect them from worry. They described the genetics staff as professionals who would tell them the truth,

indicating an element of trust in the staff. The importance placed by clients on their relationship with the counselor indicated that this was a significant factor in the process, and required inclusion in the overall theory. “Negative examples” further demonstrated the importance of the relationship. Andrea (S18) had not been visited by a counselor prior to her clinic appointment, and felt that she was not treated individually by the staff in the clinic. Although she reasoned that she had received the information she required, she was very disillusioned by her contact with the service, which she felt was “uncaring.” To explore responses to the service further, each client was asked if they felt they would contact the staff again, if they had queries about the information discussed. There were clear patterns that emerged as a response to this question. Some felt they could contact the staff again if they needed clarification or support; these clients tended to be those who had found the contact with the genetic counselor helpful and felt they had been treated as individuals. There were those who simply did not feel that it was appropriate to contact the service again, because the genetics service was seen as an adjunct to the more individual care offered by their GP or pediatrician. In a third group were those who felt alienated from the staff and indicated they would not contact the genetic service again. These clients had felt distressed by the process and appeared to feel that the staff had not demonstrated care for their individual concerns.

Major Theme 3: Integration of Lay and Scientific Explanations

The need to make sense of the present situation as previously described also influenced the construction of lay explanations by clients. Whilst there were few people who could explain the occurrence of the genetic condition in their family, most had given thought to possible causes and had a candidate theory about the inheritance. These theories tended to be individual, and partly based on related, but not necessarily relevant, information. For example, three women at risk of breast cancer used information from media sources about gene mutations, combined with the pattern of cancer in their own families. Paternal exposure to chemicals before conception was the explanation used by a woman with Marfan syndrome and a man with bony exostoses, whilst one woman attributed her retinitis pigmentosa to prematurity. However, it was the parents of affected children who struggled hardest to examine all aspects of the pregnancy and birth in an effort to determine influential factors. For example, a pediatrician had told Marilyn and Harry (S17) that their child had a chromosomal abnormality. Marilyn put together a hypothesis as to how the abnormality could have arisen:

I: And you said that you had blood taken because it could be hereditary, does that mean that his, you understand his condition might not be hereditary?

S: Umm, that's right.

I: How would that have happened then?

S: Umm, I don't know, I don't if um, I know when you're pregnant you take milk to drink, and not smoke, cause that can cause um, mental illness, physical handicap, so I

assume it would be something along those lines. I don't know. Very very early on in a pregnancy.

I: *So are you saying it might have been something environmental?*

S: *I don't know. (laughs). I don't know, cause um, I mean things, at the end of the day things don't just happen, there has to be some sort of reason for them happening.*

I: *If it wasn't hereditary, in your case, can you, do you sort of think about what it might possible have been in your own case?*

S: *Um, yeah.*

I: *Have you got any ideas?*

S: *No. Um, I said to my husband, do you think the occasional drink had done it, but, I mean, I guess it's only probably about three times a month, wasn't it (laughs nervously) probably, and then it wasn't every month, so I mean it would have only been one or two glasses of wine*

At least 20 (51%) of clients had been seeking information. To examine the extent to which this need was realized, they were asked by the researcher how much they had learnt as a result of the genetic counseling process. In all cases the client felt that they had learnt something about the natural history, prognosis, genetic testing or genetic cause of the condition. However, despite the number of clients who said they were seeking risk assessments prior to genetic counseling, actual numerical risk figures did not appear to be of importance to many and was poorly recalled. Where clients did want information about their risk, an explanation of the way the figure was obtained helped the client to integrate their lay knowledge and the new information. Four families felt they had not had a satisfactory explanation of the way the recurrence risk had been assessed, and therefore had difficulty making sense of it. Anthea (S22) was confused:

I: *Umm, did she talk about how (condition) might be inherited then?*

S: *She said there was a 50/50 chance of it going, sort of down, she said it did tend to sort of skip some generations and then come back*

I: *Right, so it could skip generations?*

S: *Yeah*

I: *Right. So where does the 50% come from, did you understand why it's 50%?*

S: *Umm, no, she just said it was sort of just a 50/50 chance as to whether the faulty gene came out as faulty, or whether it came out as normal*

Whilst the 50% risk was remembered by Anthea, she had clearly not grasped that an unaffected person could not pass on the gene mutation, so although she had been told she did not appear to have inherited the condition, she was still concerned that her children were at 50% risk. Two families found it difficult to understand why the geneticist concentrated on one side of the family rather than both sides, and appeared to be left with the feeling that the process had been incomplete. Jenny's (S33) daughter was being investigated for a connective tissue disorder. Although she was aware that tall stature was a sign of the condition, she was still not satisfied with the apparent focus on one side of the family:

S: . . . only they seemed to concentrate on his side rather than my side, which I thought was a bit strange, cause I did ask but they just said that they were just gonna get his side sorted out because they had more, seemed to have more symptoms, but that might be the case, I don't know.

However, the clients who were satisfied that they understood the explanation reported “peace of mind” after the consultation. Gloria (S39) had been concerned about her family history of breast cancer, but was satisfied with the explanation for her low risk:

S: He said there was no problem, I wasn't really high risk, I was no different than anybody else of my own age, that the risk wouldn't really be any different, and . . . simply because mum didn't have breast cancer, and she lived till she was 69, and both me sisters being at the age . . . where the hormones change and things that it was just more or less one of those things, and that my chances were no different really than anybody else's

Major Theme 4: Outcome Related to Psychological Adaptation

Overall, 39 families of the 40 studied postcounseling felt that the referral had been worthwhile for them. The clients in this study did not report significant practical changes to their lives as a result of receiving genetic counseling. Few perceived changes in reproductive plans, work, insurance, lifestyle, or family relationships. However, when asked about changes to their lives, clients typically cited psychological changes related to their own adaptation to their situation, often using terms such as “peace of mind.” Lydia’s (S50) comments reflected the feelings of many others:

S: Well, umm, yes, yes, peace of mind, it really is, it has made a difference because I, I just, my mental outlook

Ben (S26) was relieved to know his son’s disability was due to a genetic cause, rather than birth trauma, although the exact diagnosis was not made:

S: I think it did (help) a bit, because it's now out of my mind, whereas before I kept on thinking about this one man (the obstetrician), not all the time, but when I did it would wind me up a little bit really, and now I know it's what would have been anyway, so it's made it easier

This “peace of mind” was not confined to those given a low risk, as all those who received news that they or family members had a moderate to high risk, responded that they felt better knowing and understanding the risk.

But there were also those who felt their peace of mind had been disrupted by further uncertainty, such as Marg (S38):

S: No, no, we didn't get anything, for that fact they raised more questions for us, and more problems for us, than . . . anything which had, so that, I mean we were more, more confused and more upset and more . . . anxious to find out things about Kyle, but we didn't get told anything

Development of a Grounded Theory

Using the principal themes that emerged from the study, a grounded theory was developed. This is represented in diagrammatic form in Fig. 2. The central

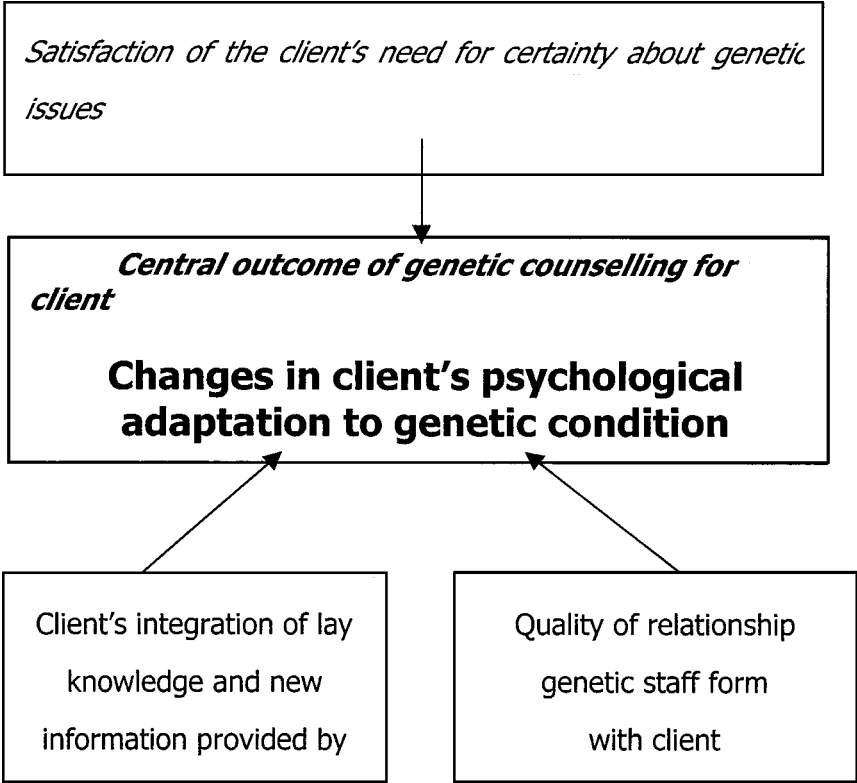


Fig. 2. Representation of the theory: Factors that influence the outcome of genetic counselling from the client's perspective.

outcome of genetic counseling for clients at risk of a range of genetic conditions has been identified as a change in the client's psychological adaptation to the situation. This outcome is influenced by three key factors, the satisfaction of the client's need for certainty, the relationship between the client and the staff of the genetic service, and the extent to which the client is able to integrate the information provided by the genetic counselor with his or her lay knowledge of the condition.

It is proposed that psychological adaptation, principally 'peace of mind,' is directly connected with the extent to which the genetic counseling process is able to address the client's concern for certainty with regard to the genetic condition in the family and the effect of that condition on the lives of family members. The family's lay knowledge of the genetic condition is also relevant to the central outcome, as the extent to which certainty is obtained will be conditional on the client's ability to satisfactorily integrate lay and scientific

knowledge. Where incongruence exists between the two versions, it is difficult for the client to obtain certainty. However, even where integration is possible at the time of consultation, the effects of the genetic condition on the family will alter over time. Explanations given by the genetic counselor therefore need to encompass the family experience and also be robust enough to accommodate changes. The relationship between counselor and client also needs to be appropriate so that clients have confidence that the information pertains to them directly and so they feel able to recontact the counselor if further explanation is needed.

DISCUSSION

Need for Certainty

Clients in this cohort demonstrated a general preference for certainty. Webster and Kruglanski (1994) describe the concept of the need for cognitive closure as an individual drive for certainty and a discomfort with ambiguity. It may be that clients who request or prompt a referral to genetic services have a greater need for closure than those at genetic risk who avoid or do not seek genetic counseling. This could explain the variation in approaches to genetic counseling between members of the same family, which is clearly described by Lynch and Lynch (1994).

It would appear that obtaining certainty is one way of enabling a client to wrest control of the situation. Berkenstadt *et al.* (1999) devised an outcome measure for genetic counseling, called Perceived Personal Control, and found that counselees who obtained a more certain diagnosis or specific recurrence risk were found to feel a greater sense of control.

One feature of the genetic service that was emphasized by clients was the need for honesty from those providing the service. Even when the client dreaded bad news, they stressed the desire to know what the situation was, so they could learn to cope with it. There was an acknowledgment that family members might blur the truth, in order to save the client from worry. This phenomena (family protectiveness) has been documented in other fields of health care (Claffin and Barbarin, 1991; Pursell, 1994; Tapp, 1993) as well as by authors of other studies relating to genetic disease (Barette and Marsden, 1979; Skirton, 1998). These issues are interesting from an ethical perspective, particularly when considering the concept of nonmaleficence (Hurwitz and Richardson, 1997). Giving 'bad news' could in some cases be seen as causing harm, although not to give the news would be seen as damaging to patient autonomy (Gadow, 1981). These clients, however, are strongly expressing the view that to withhold bad news would be seen by them as both harmful and a threat to their autonomy.

The Counselor/Client Relationship

The relevance of the association formed between professional staff and the client is discussed by few other authors in relation to use of genetic services. One publication, which does address the issue of personal relationships between client and genetic counselor is that by Lynch and Lynch (1994) on the subject of counseling for familial cancer. Trust and confidence between all the protagonists involved in the process is stressed as being essential to the counseling process. It is also considered important that there is a positive relationship between client and professional to facilitate the dissemination of information throughout the family. Shiloh and Berkenstadt (1992) have emphasized the importance of effective communication between client and counselor, and the cases in this study appear to confirm that effective communication at least partly depends upon the client believing that the counselor is interested in their personal circumstances. This point was also demonstrated in a paper by McCarthy Veach *et al.* (1999), in particular in a report of a client who described her adverse reaction when the counselor proceeded to give her the standard information about termination, despite the client having expressed her revulsion for the topic.

Lay and Scientific Knowledge

Clients in this study had developed their own lay explanation for the occurrence of the condition in the family. This finding is consistent with the views of Hallowell and Richards (1997), who believe that genetic counseling cannot be effective unless the lay knowledge of the client is considered as part of the process.

In the present study, some clients did not understand the reasons for particular actions by the genetics staff. For example, several families were unclear as to why certain tests that they had been expecting were not performed. This finding was also reported by Hallowell *et al.* (1997) in the study of women with a family history of breast cancer, 9% of whom said the consultation had not satisfied their expectations because they had expected to have a blood test. Armstrong *et al.* (1998) refer to the expert knowledge that guides the genetic staff in the pursuit of information, a knowledge that is not matched by the client's. It appears that within the genetic consultation there are a number of clinical decisions made that make sense to the professionals involved, but not to the client. Discussion with the families during the research interviews clearly showed that these unexplained features of the consultation remain as riddles to the family, inhibiting the overall integration of the material discussed into the client's knowledge of the condition. This knowledge is dynamic and evolutionary, therefore any explanation that is made needs to be sufficiently robust to accommodate further information or circumstances as they come to the attention of the family.

Clients felt that the outcomes that were important to them related to their psychological ability to deal with the condition. This is certainly consistent with the findings reported by McCarthy Veach *et al.* (1999), who documented the significant emotional responses described by clients after the genetic counseling session, in particular "relief." Development of measures to assess this type of outcome may be more useful for audit of genetic services than assessing changes in risk knowledge or reproductive behavior.

CONCLUSION

This longitudinal study has followed clients through the process of genetic counseling, to identify those aspects that are meaningful to them. The use of reproductive behavior and retention of knowledge used in other studies as outcome measures provide, at best, an incomplete picture of the effectiveness of genetic services. Whilst practical changes are difficult to discern in the lives of many clients, psychological changes do occur that may enable the client to deal more effectively with the condition or the threat of the condition. The development of a grounded theory may assist practitioners to identify features of the genetic counseling process that have a significant impact on the outcome for the client. In particular, it appears to be important to discuss the family's lay construct of the cause and inheritance of the condition to assist them to assimilate the genetic information given, and to address the client's need for certainty. The influence of genetic counseling on the client's life may be positive or negative, but such subjective psychological changes are difficult to quantify and therefore to assess. Future studies to develop a tool for the assessment of genetic services based on the findings of this study are planned.

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