2000

GENETIC COUNSELLING: A REVIEW OF THE LITERATURE

A Pilnick
R Dingwall
E Spencer
R Finn

Trent Institute for Health Services Research
Universities of Leicester, Nottingham and Sheffield

DISCUSSION PAPER 00/01

Published by the Trent Institute for Health Services Research

ISBN 1-900733-37-4

Referencing information:

Pilnick A, Dingwall R, Spencer E, Finn R *Genetic Counselling: A Review of the Literature.* Sheffield: Trent Institute for Health Services Research, Universities of Leicester, Nottingham and Sheffield, 2000. Discussion Paper 00/01.

Further copies of this document are available (price £15.00) from:-

Information Resources
Trent Institute for Health Services Research
Regent Court
30 Regent Street
SHEFFIELD S1 4DA

Tel 0114 222 0703
Fax 0114 272 4095
E-mail scharrlib@sheffield.ac.uk

ABOUT THE TRENT INSTITUTE FOR HEALTH SERVICES RESEARCH

The Trent Institute for Health Services Research is a collaborative venture between the Universities of Leicester, Nottingham and Sheffield with support from NHS Executive Trent.

The Trent Institute:

- undertakes Health Services Research (HSR), adding value to the research through the networks created by the Institute;
- provides advice and support to NHS staff on undertaking HSR;
- provides training in HSR for career researchers and for health service professionals;
- provides educational support to NHS staff in the application of the results of research;
- disseminates the results of research to influence the provision of health care.

The Directors of the Institute are: Professor R L Akehurst (Sheffield);
Professor M Clarke (Leicester); and
Professor H Williams (Nottingham).

Professor Clarke currently undertakes the role of Institute Co-ordinator.

A Core Unit, which provides central administrative and co-ordinating services, is located in Regent Court within The University of Sheffield in conjunction with The School of Health and Related Research (ScHARR).

CONTENTS Page

EXECUTIVE SUMMARY

The rapid scientific and technological advances in the field of human genetics have created an ever-widening gap in knowledge and understanding between those specialists who are involved with them and the general public who are the intended beneficiaries. Genetic counselling services are seen to be an important way of addressing this gap. As a result, there is a growing body of literature considering genetic counselling services in a variety of clinical settings. This literature encompasses both predictive and diagnostic testing, from the viewpoints of service providers and recipients. It also embraces a wide range of conceptions of the nature and goals of genetic counselling. However, research in this area has been criticised for a focus on *outcome* rather than *process*, and it has been suggested that this focus limits its practical use. The purpose of this review is twofold: to describe the varying concepts of counselling which appear to be utilised in published work; and to discuss the possible applications of this work to practice. The review is not an exhaustive survey, in the manner of a systematic review, but examines examples of the types of study which have investigated genetic counselling and represents the limited range of methods commonly used.

Genetic counselling is a process through which people affected by, or at risk of, a hereditary disorder are told about the possible consequences, the probability of developing or transmitting it, and the ways in which this may be prevented or ameliorated. Whilst this definition evidently underpins most of the research which is discussed here, there are significant differences in how the definition is practically applied and what is considered to fall within this remit. Existing research has been largely informed by a fairly basic, quantitative psychological paradigm, relying on psychological tests and scales, attitudes to testing, questionnaires, or post-counselling interviews. The limitations of these methods in relation to understanding the process of genetic counselling is discussed, and the review concludes with suggestions drawn from other medical settings for broadening the methodological agenda.

INTRODUCTION

The rapid scientific and technological advances in the field of human genetics have created an ever-widening gap in knowledge and understanding between those specialists who are involved with them and the general public who are the intended beneficiaries. Like any discovery, this new information may be used for good or for evil. In the light of the historic abuse of genetic information for eugenic purposes by authoritarian governments, this ethical problem is well recognised and has been the subject of considerable philosophical discussion. Counselling is seen to be an important part of the answer.

Within the setting of the clinic, ethical problems are implicitly present but may not be explicitly addressed. The counsellors' job is defined in terms of neutrally summarising and passing on sufficient information for clients to make their own decisions. The ethical responsibility for those decisions is thus transferred from the providers to the users of genetic services.

It is, though, far from clear how users are to make sense of the genetic information that they are given, having arrived at the clinic with little or no previous knowledge. Similarly, they are expected to make significant ethical decisions without prior preparation and in a context where experience may have led them to expect that professionals will make recommendations for them. When we move to these issues about the delivery and receipt of information, we are moving from philosophical to social scientific questions. How is information to be given in a neutral fashion? How are users to be encouraged to give appropriate moral consideration to the implications of their individual decisions? How is prescription to be avoided in a setting where people may be searching for it? Ethical pontification comes in the end to very practical questions about the way professionals and clients, men and women alike, talk to each other.

This review first seeks to establish what is known from empirical studies about the actual interactional performance of genetic counsellors. It then moves on to consider the state of knowledge against the background of theoretical possibilities offered by the general research literature on professional/client interaction. In the conclusion, we set out some of the directions which might profitably be followed by future research in this area.

Search Strategy

In order to present an accurate overview of the existing literature in this field, a comprehensive search was carried out. The following databases were searched from 1980 onwards: Social Science Citation Index, International Database of the Social Sciences, Medline, EDINA Biosis, and the Biomed Core Biomedical Collection. The search terms used were genetic counsel(l)ing, genetic counsel(lor), and genetic testing. All of the articles which were located through this search and which constitute empirical research have been reviewed here, regardless of study design or methodological rigour. However, on occasion, this presents difficulties in making comparisons between findings: few studies, for example, are designed with a control group, and some studies give only very limited methodological details. A further issue is that whilst the majority of the studies that are reported here concern themselves with prenatal testing, there is also a sizeable body of work on predictive genetic testing. Evidently, these are distinct activities which raise different issues and implications. These differences aside, the choices of study tools in the work presented in this review would seem to suggest that this is a research area which is strongly influenced by a quantitative, social psychology paradigm.

1. COMMON THEMES IN RESEARCH

Existing research has relied mainly on psychological tests, questionnaires or post-counselling interviews. A significant body of this research is concerned with attitudes to testing, often asking people to put themselves in a hypothetical situation in relation to a specific genetic disorder, and aiming to discover how they think they would act if they were to be in this situation. Whilst there are clearly problems with assuming a simple link between hypothetical attitudes and actual behaviour, the validity of those methods which attempt to analyse actual counsellor/client interaction can also be called into question on methodological grounds. The fundamental issue here is that any kind of post-counselling analysis depends largely on the recall abilities of subjects. What these studies do show is a great deal of variation amongst perceptions of what counselling is supposed to be and what it is intended to achieve. Genetic counselling may be broadly defined as the process by which patients or relatives at risk of a disorder that may be hereditary are advised of the consequences of the disorder, the probability of developing or transmitting it, and of the ways in which this may be prevented or ameliorated. Whilst this definition evidently underpins most of the research which is discussed here, there are significant differences in how the definition is practically applied and what is considered to fall within this remit. Where these studies do concur is in indicating a very low level of awareness, even among populations who might be thought to have particular reasons to be informed, about either technical or policy issues in genetics, let alone in genetic counselling.

1.1 The Impact/Effectiveness of Genetic Counselling

Relatively few researchers have attempted to evaluate the actual effectiveness of genetic counselling. This reluctance may be explained in part by the fact that how efficacy might be defined in this context has been the subject of much debate (Clarke, 1991a; 1991b). The studies that have been located and discussed here use a variety of methods to tackle the problem of efficacy, both directly and indirectly in terms of impact. These studies also demonstrate the variation in what is actually meant by `counselling'.

Frets and her colleagues (1991) analysed the problems people experience in making reproductive decisions following genetic counselling. Counselling was given to couples following a positive carrier result. The investigators followed up 164 couples, between two and three years after their initial genetic counselling session. As many as 43 per cent were shown to be experiencing problems in reproductive decision making. For couples deciding to have children, fewer decision-making problems were experienced when prenatal diagnosis was not available. This is particularly important when we

consider that one justification for investment in the new technological advances in genetics is said to be the choice it offers to at risk individuals. If people experience fewer problems when there is no prenatal test available, this argument loses some of its force. We should not necessarily assume that genetic counselling is a straightforward means of helping individuals to reach a decision, since the real likelihood is that additional information will make this decision-making process more complex.

A further finding of Frets et al is that problems for couples often become apparent after rather than during counselling. They suggest a structured follow up three to six months after the initial counselling session so as to identify those who would benefit from additional supportive counselling. They also encourage counsellors to explore other feelings and emotions in more depth in the first session. Here counselling is seen to have a psychosocial element rather than just the giving of information.

Another study by Lynch and Watson (1992) reported responses to counselling by their patients who were at risk of hereditary breast or ovarian cancer. Responses ranged from objective acceptance to disbelief or confusion. More genetic and/or psychological counselling was needed for some. A fear of disclosure of information was a common concern. Although many expressed relief at knowing, some of those shown not to be carriers expressed disbelief, which was dealt with in counselling. The authors conclude that physicians must know how to interpret molecular genetic and gene linkage findings and how to inform and manage high-risk cases. These authors appear to regard counselling primarily as the interpretation of technological information with a psychosocial dimension being distinctly secondary, since they stress emotional impact as an issue which will need to be dealt with at a later date.

Other studies discuss and assess counselling in terms of its impact on reproductive decision making, e.g. Somer, Mustonen and Norio (1988), D'Amico, Jacopini, Vivona et al (1992), and Czeizel, Metneki and Osztovics (1981). The difficulties associated with this choice of outcome measure in the context of public perception of the aims of the new genetics are perhaps best illustrated by a paper entitled 'Impact of Genetic Counselling After Neonatal Screening for Duchenne Muscular Dystrophy' (DMD) by Hildes et al (1993). The authors report a pilot neonatal screening programme which offers the opportunity of prenatal diagnosis for future pregnancies in at risk females. As a result, prenatal diagnosis is carried out in only 2 out of 7 subsequent pregnancies. Some thoughtful points are raised in consideration of this: for example, the authors raise the complex psychology behind the decision to request termination of an affected pregnancy. Nonetheless, they conclude that highly accurate carrier testing and prenatal diagnosis, very early carrier identification, and genetic counselling after identification of DMD males in a population based neonatal screening programme "*may not be an*

effective way of decreasing the number of repeat cases of DMD within families or the overall population frequency of DMD." It would be easy to conclude from this that the aim of the programme, and of the new genetics as a whole, is the reduction of births with genetic disorders. This is not meant as an accusation of eugenics, however: it may be that the number of births of babies with a particular disorder becomes a kind of default goal for these kind of programmes, since it is a concrete outcome measure. Nevertheless, it is easy to see that the perceptions this may create are potentially extremely problematic.

Although this paper's primary focus is on the impact of genetic counselling, rather than the effectiveness, it is still possible to draw some conclusions from the results along these lines. For example, it would seem that the results are suggesting that genetic counselling does not have any particular impact upon high-risk individuals' reproductive behaviour. In which case, if we were to assume that one aim of genetic counselling is the reduction of DMD births and to define effectiveness as an actual reduction in births, we could conclude that genetic counselling with regard to this particular illness is not effective. Counselling in this study seems, implicitly at least, to be evaluated as a persuasive act, where information is communicated in a way intended to achieve a particular goal.

A different stance is taken by Swerts (1987), who looks at the impact of both genetic counselling and prenatal diagnosis for Downs Syndrome (DS) and neural tube defects (NTD). The information given during counselling was reported as influencing more than half of the parents of a child with DS, and more than 80% of a child with NTD to plan another pregnancy. For more than half of the families of children with both disorders, the availability of prenatal diagnosis was considered to be of crucial importance in the decision to plan future pregnancies, which suggests that they intended to make use of these services. In terms of genetic counselling alone, a significantly better recall of the relevant risk figures was found in the counselled group as compared with the group who had not received any counselling. There is certainly no suggestion of any kind of goal of reduction of affected births in this study, and it appears that counselling here is treated as straightforward information delivery. As a result, its efficacy can be assessed through client recall.

Effectiveness in terms of knowledge acquired or information recalled after counselling appears to be a commonly used proxy in this literature, demonstrating the emphasis which is often to be found on the educational aspects of counselling. A number of other studies were found which compared 'before' and 'after' knowledge of genetic factors (e.g. Seidenfield and Antleys' (1981) study amongst mothers of children with Down's Syndrome, Michie, McDonald and Marteau's (1990) study involving an

unspecified group of 32 counsellees) or of risk and diagnosis (e.g. Sorenson, Kavanagh and Mucatel's (1981) large scale study of 2,220 counselling recipients.

The reported findings of Watson, Mayall et al (1992) are consistent with those discussed so far. People who were counselled following screening for CF showed a reasonable knowledge of the disorder and its inheritance six months later. However, this study takes a broader view of screening, by assessing its effects on anxiety levels, attitudes and actions of participants as well as knowledge. It is not clear what form the counselling component took, or how it was carried out. In conclusion, the authors report that varying degrees of anxiety were found to be associated initially with a positive result, but most of this was allayed by genetic counselling. Unfortunately, however, this study did not have a control group, so it is difficult to know how much weight to attach to these findings. The concept of counselling here, then, combines information-giving and some form of psychosocial intervention.

Highlighting another issue, Tibben et al (1992a; 1992b) report how, within the confines of service provision, counsellors tend to pay less attention to individuals who participate in pre-symptomatic DNA testing for Huntington's disease but test negatively. However, the authors reported the existence of long-term numbing and survivor guilt amongst those at risk individuals receiving a negative test result. Having shaped their lives against a background of possible Huntington's Disease, the emotional effects of a negative test could still be considerable, and some of those testing negatively reported feeling that their siblings would now be more at risk, or that they now had an obligation to help sufferers in their family. The paper concludes that carriers, non-carriers and partners must all have their specific needs attended to by genetic counselling for at least one year after testing. Here the emphasis goes back to the psychosocial meaning of counselling rather than the information- delivery aspect, and encompasses all service users, regardless of their eventual status.

Apart from the advocacy of further or expanded counselling by some of these studies, and the useful findings of Frets et al, there is little one can conclude about the effectiveness of genetic counselling, beyond echoing Clarke's (1991) suggestion that it is very difficult to define efficacy in this setting. Indeed, the studies are most useful in illustrating the inconsistency and/or reluctance to define what a particular counselling programme actually consists of and what it is intended to achieve. Much of this reflects the evasiveness of the new genetics about its goals and the standards by which effectiveness could be measured. Unless it is possible to specify an intended outcome more clearly, it will be always be difficult to evaluate the effectiveness of the counselling process. Is it to be assessed by the level of understanding achieved, by the resolution of psychological distress or by a

specific change in behaviour? Each of these implies something different about the nature of the counselling process itself and the skills that would be required to carry it out.

Other research has attempted to tackle this problem from a different viewpoint. Clients' reported satisfaction with counselling has been used as a proxy measure for effectiveness, e.g. in Bleiker, Aaronson, Menko et al's (1992) study of 36 individuals who had received genetic counselling for cancer. These individuals were asked to rate their satisfaction both with the care provided by the clinical geneticist they had seen, and with general procedures at the clinic. Zare, Sorensen and Heeren (1984) used similar methods in asking clients to rate counselling sessions in terms of clarity, depth of discussion and clients' willingness to raise issues; their particular interest, however, concerns whether the sex of the counsellor affects these ratings. Interestingly, both of these studies acknowledge their limitations, stressing the importance for findings such as these to be related to or validated by qualitative analysis of the actual counselling session. Reported 'satisfaction' is highly questionable as a valid or useful measure of the counselling process, since it does not necessarily provide any information about what has actually occurred during counselling. Indeed, this problem seems to be common to most of the literature purporting to look at effectiveness of counselling: whilst the research focus has been on *outcome* measures, effectiveness is also fundamentally related to the delivery of service, or *process*. Clarke, Parsons and Williams (1996) highlight this distinction, suggesting that whilst outcome measures are valid in a research context, they are useless in practice; this is echoed by Kessler (1997) who argues that outcome measures are methodologically inadequate, as well as inappropriate, alone.

There are studies which go some way towards meeting this call for a focus on process when examining effectiveness, for example by comparing different methods of counselling and assessing the outcome in some way. Whilst this does not entirely solve the methodological problem, it does address questions about *how* counselling should be conducted in order to be effective. Strategies used here include the use of videotaped information alongside counselling (Cull, Miller, Porterfield et al, 1988), or the use of differing counselling 'formats'. Work by Fisher, Rowley and Lipkin (1981) and Rowley, Lipkin and Fisher (1982) reports the use of 3 counselling formats: 'conventional', 'programmed' (again using video tape) and 'patient structured', and uses outcome measures of recall and satisfaction to assess these. Young, Jorgenson and Shapiro (1986) also compare three formats (in this case they are group, tape and individual counselling), and in assessing recall, conclude that none of these are significantly more effective at conveying information. Similar studies have incorporated other process variations, e.g. Loader, Sutera, Walden et al's (1991) use of dramatisation and role models, but all have in common the *outcome* measures used as assessment. In addition, in

all these cases, there is limited information on how these differing kinds of format were interactionally achieved or carried out, or indeed how the client's recall relates to the actual counselling session. The only study found here which attempts to address the latter part of this problem is by Michie, French, Allanson et al (1997), which compares counsellors' summaries of consultations and the information recalled by clients with tape recordings of the actual sessions. For their sample, the authors concluded that counsellors' summaries were a valid baseline against which to measure patient recall. Unfortunately, this still fails to address the more fundamental problem of assessing the interactional process of counselling, since the process component here is used indirectly to validate a particular outcome measure, rather than as a resource in itself. It seems that while the importance of the counselling process has been recognised, the search for effectiveness measures has yet to fully incorporate this.

1.2 The Role of the Genetic Counsellor

At least part of the problem of defining a suitable measure of effectiveness, and finding ways to analyse this, appears to stem from broader debates on the role of the genetic counsellor, particularly in relation to the complex ethical considerations that are an integral part of the genetic counselling process. A clear definition of what counselling, and the role of the counsellor, should consist of, is an essential basis for any evaluation. There is already a recognition of the need to research what occurs in the process of counselling, and also to evaluate counselling on the basis of process factors. Fundamental then is an agreement of what should characterise this process in order to evaluate it. Unfortunately, there is a clear lack of consensus, and much debate, concerning how the counsellor role and counselling process should be characterised. Furthermore, there must be a recognition that often, 'taken for granted' terms and concepts relating to role can carry a multiplicity of meaning, and involve contested definitions. Which definition is being used has important implications for the assessment of any study.

The key area around which this conflict appears to revolve is non-directiveness. A significant amount of literature focuses on the role of the genetic counsellor in relation to the guiding principle of non-directiveness, and generally, research suggests that directiveness is not uncommon (Michie, Bron, Bobrow et al, 1997). However, as Kessler (1997) notes, the issue of definition of this term is critical, since it may be invoked to incorporate anything from solicited advice giving to persuasive coercion. Once again, the interactional dynamics of counselling are a fundamental issue here: whilst a clear definition might make the task of evaluation easier, the situations arising in counselling sessions are too complex to be easily suited to rigid adherence to particular principles. Indeed, some of these

principles, such as client autonomy, may not be desirable to all those receiving a counselling service. Just as there are differing professional perspectives on what a counselling session should constitute, so there are amongst clients. The next two sections of this review consider the research examining these lay perspectives.

1.3 The Lay Person's Knowledge and Attitudes

Lay perspectives do receive some attention in the literature, with the majority of research focusing specifically on lay knowledge and/or attitudes to genetic services. The research studies which fall into this category have used a variety of survey designs and strategies for their data collection. Again, actual genetic counselling clients, members of a client's extended family and the general public have all been used in these studies. Both prenatal and predictive testing have also been considered. These studies can be seen as a way of defining the baseline knowledge about genetics in the population which may come for counselling and of the attitudes with which potential recipients approach the encounter. However, there seems to be no work which deals directly with the non-professionals' specific knowledge of or attitude to genetic counselling, as opposed to screening in its most general sense.

We will look first at different populations' attitudes towards carrier testing and prenatal diagnosis (see Decruyenaere et al 1992a, 1992b; Hildes et al 1993; Mitchell et al 1993; Watson et al 1992a, 1992b) and then move on to look at levels of knowledge about these services.

Decruyenaere and his colleagues (1992b) questioned 169 women aged between 21 and 35. They were asked to state what their attitude to predictive testing for Huntington's disease would be, if they were to have a 50 per cent risk of developing the disorder. The use of such hypothetical exercises is not unproblematic as a research tool, since they rely on the respondents' ability to imagine themselves in the specified situation. The validity of this assumption frequently goes unexamined. These reservations aside, however, the results are typical of this type of study in the field. Half the group, although expressing interest in predictive testing, would be reluctant to tell their employers and insurers if they received a positive test result. This shows that people are aware of the dangers such knowledge about one's own condition can bring. Having ascertained attitudes towards predictive testing, the authors go on to establish whether prenatal testing would be acceptable. The study also found the typical response that, despite a high acceptance of prenatal testing in theory (50%), only one quarter of the women thought they would terminate a pregnancy following a positive result (see Evers-Kiebooms et al 1993; Denayer, Evers-Kiebooms et al 1992; Decruyenaere et al 1993). It would

seem that the efficacy of genetic counselling as measured by a reduction in affected births is likely to be not only unacceptable, but also inaccurate.

Denayer, De Boeck et al. (1992) and Denayer, Evers-Kiebooms et al (1992) both looked at reproductive decision making, risk perceptions and attitudes towards carrier identification and prenatal diagnosis among aunts and uncles of a child with cystic fibrosis (CF). Again the findings were consistent with similar studies. About three quarters would (probably) make use of heterozygote detection and would (probably) ask for prenatal diagnosis should they become pregnant, but less than half thought that they would opt for termination given an affected fetus. The intention to use prenatal diagnosis was significantly associated with age and educational level. Other studies which found a correlation between education level and age with knowledge and/or decisions made include Tibben et al 1992a, 1992b; Wertz and Sorenson 1986 and Decruyenaere et al 1992a, 1992b. Despite the correlation between an apparent willingness to utilise genetic services and educational level, however, Decruyenaere et al draw a more general conclusion: that the rate of technical advance in genetics contrasts sharply with the slow diffusion of information on this subject to the general population (Decruyenaere et al, 1992b: p189).

Three other studies have looked at people's attitudes towards carrier testing and population screening (see Mitchell et al 1993; Watson et al 1992a, 1992b). Mitchell and colleagues looked at what young people (i.e. between 15 and 17 years) think and do when the option for CF carrier testing is available. There was a high participation in the screening and the authors found that carriers had positive attitudes towards the education and testing experience. Parents of children with CF were targeted by Watson et al to study their attitudes towards both prenatal diagnosis and carrier screening. They discovered that as many as 92 per cent of their sample would support the introduction of a population screening test to detect carriers of CF but only 19 per cent thought it should be mandatory. 64 per cent would not choose to have any more children if it were known that they were both carriers, 74 per cent would have a prenatal test if pregnant, 44 per cent would not and 23 per cent were unsure. Overall, by whichever means, 72 per cent would seek to avoid having another child with CF. A second study by Watson, Mayall et al (1992) examined the psychological and social consequences of community carrier screening programme for CF. Over 3000 individuals were screened. All those involved were reported to be in favour of screening and were glad to have been tested, suggesting that screening may improve understanding of CF among the population.

From these surveys of peoples' attitudes towards, or perceptions of, particular services we can see some common findings:

1. People are generally in favour of the idea of prenatal diagnosis, but are less clear about whether they would opt for therapeutic abortion in response to a positive result. Since many of these studies have been carried out using members of the population not known to be at genetic risk, it is easy to see why they find it hard to make decisions involving a hypothetical situation that may be far removed from their realm of experience.

2. People would usually want to keep the knowledge about their own condition within a close network of family and friends. This also raises issues of personal choice, and an individual's right to choose not to undergo testing. Issues of ownership of knowledge are also pertinent: if an individual's family may also be at risk, who has the right to tell or withhold this information?

3. It appears that people's level of education and age are significantly associated with whether or not they find particular genetic services acceptable as an option for them. There is also a proposed correlation between class position and attitude towards genetic services (Wertz and Sorenson 1986). It is unclear if these reported attitudes are related to an individual's perceived understanding, and hence usefulness, of a service, since if a person feels they will not be able to understand or apply information of any kind they are unlikely to seek it in the first place.

The following two examples focus more directly on people's knowledge about genetics, again looking either at the general public or at the extended family of an affected individual.

The first example examined community knowledge about carrier screening and prenatal diagnosis for CF (Decruyenaere et al, 1992b). The findings show a very poor knowledge of the nature of CF and an even more limited awareness of its inheritance by the group. Again, knowledge was mainly associated with educational level. Varekamp et al (1993), in their retrospective study of women who were possible or obligate carriers of the haemophilia gene, found four factors to be statistically related to their sample's knowledge about carrier testing for haemophilia: information via the media; an affected relative in the nuclear family; medical severity of haemophilia (three levels of severity); and information from patients' organisations. The last two factors were related to usage of carrier screening by those with knowledge. The following three factors were also found to be related to usage: attitudes towards abortion due to haemophilia; educational level; and marital status. Respondents stated that relatives, especially parents and sisters, were the most important source of information about both genetic counselling and carrier testing.

Extended family members have been the target for much of the research in this area (see Denayer et al 1992a, 1992b; Evans et al 1993; Suslak et al 1985). Perhaps because of their family status, it seems to have been assumed that this group of people should be better informed than the general public. Studies have looked at the way in which information has been transmitted among family members and the degree to which counselling has been understood. Only one asked direct questions about the transmission of information between family members (Suslak et al 1985). The authors interviewed 12 balanced translocation carriers who had 36 siblings and 21 parents between them. Out of these, four siblings had not been informed (from two families) and only 16 of those informed had had tests. Of the parents, 14 had been informed and three couples had been tested. This again raises the question as to whether genetic professionals have a right, or indeed a duty, to disclose genetic risk information to siblings or other individuals at possible risk. Results from the work of Denayer et al (1992a; 1992b) show a very poor understanding of CF by aunts and uncles of the affected child. Only a small proportion of the respondents (around one quarter) were aware of their approximate risk of being a carrier of the CF gene and/or of the risk of having a CF child. However, almost one third used their subjective understandings of this risk in reproductive decision making. So, whilst the general conclusion seems to be that the majority of the respondents were not reluctant to discuss their condition with family members and friends, it is not clear that they did so in sufficient detail to establish that relatives understood the implications of the information for them. Again, there are implications related to the ownership of knowledge here.

From these studies it is apparent that knowledge about genetics generally, or particular genetic services, is quite poor even among individuals who may either be at risk or thought to have special knowledge due to their kin relationship to an affected person. However, the subjects in these studies had not had any prior direct experience with a genetic clinic or genetic counselling. The literature dealing with this latter group of people can be divided into those studies which are carried out before counselling, those which are carried out after, and those which make comparisons of findings from both situations. In some cases, 'before' assessments are used as a tool to develop appropriate counselling, e.g. Hallowell, Murton, Statham et al (1987), whereas in others it is unclear whether the preferences or expectations elicited are ever carried through into practice (e.g. Audrain, Rimer, Cella et al, 1998). Michie, Marteau and Bobrow's (1997) study, focusing on expectations of counselling among people who have not yet received the service, actually presents an evaluation of how these expectations are fulfilled.

Other studies have more in common with the 'satisfaction' surveys previously discussed here, e.g. Shiloh, Avdor and Goodman's (1990) examination of satisfaction levels, and determinants for this

satisfaction, amongst genetic counsellees; some research combines this with an assessment of knowledge post-counselling, e.g. Markova, Forbes and Inwood's (1984) study amongst those who had received counselling for haemophilia, or encompasses more broadly social aspects such as attitudes to *dealing* with the knowledge received (Tyler and Harper, 1983).

Obviously, attitudes and expectations of clients are important for service providers, particularly with regard to satisfaction. This becomes even more important if satisfaction is to be used as a measure of effectiveness. However, lay attitudes to how counselling is carried out is only one part of the equation: as Tyler and Harper's work suggests, lay perceptions of the information given during counselling and the ways that this is dealt with are also fundamental.

1.4 The Lay Person's Perception of Risk

A number of studies have attempted to elucidate and understand lay perceptions of genetic risk. These studies fall into two groups: those which examine this issue in the absence of counselling or before it takes place; and those which examine the issue post-counselling. The former group, in the shape of aunts and uncles of children attending a cystic fibrosis (CF) clinic, are the subject of the study by Denayer, Evers-Kiebooms et al (1992). Their finding was that only one quarter of their sample of aunts and uncles of a child with CF were aware of their approximate level of risk of being a carrier of the CF gene, and/or of the risk of having a CF child. However, at least 39 per cent of the total sample used their subjective understandings of this risk in reproductive decision making, underlining the importance of lay knowledge in making reproductive judgements. These findings are fairly similar to the other studies considered here. Interestingly, the study is inconclusive in terms of future use of screening services, with 75% of the sample saying they would (probably) ask for prenatal diagnosis, and less than 50% saying that they would terminate the pregnancy if they found the fetus was affected. The authors suggest that those non-respondents to their questionnaire tended to be the relatives of those newly diagnosed with CF: there may be important issues here around attitudes towards coping with CF, as well as issues of recall, which are not discussed in this study.

Evans et al (1993) looked at the perceptions of risk in women with a family history of breast cancer before a counselling session. The questionnaire asked how women perceived the risk of breast cancer in the general population, as well as their own personal risk. These estimates were then used as a basis for the counselling. They found that only 11 per cent were able to identify the correct population risk. Over 50 per cent of these women were unable to assess their own lifetime risk. Whilst there were both under estimators and over estimators, the under estimators tended to significantly underestimate.

As a result of this study, the authors raise an important issue which is easily overlooked: for a significant proportion of these women (the under estimators), the counselling could have been a worrying or threatening experience.

Returning to the generally poor assessment of risk, Huys, Evers-Kiebooms, and d'Ydewalle (1990) explain similar findings in a pre-counselled group as the result of the difficulties people have in understanding risk expressed in terms of probabilities. More than half of the women in their sample gave an incorrect answer to questions about their own risk. Some reformulated the risk of an affected child, framed by the counsellor in terms of a healthy child, into a risk framed in terms of an affected child.

The final studies considered in this group are by Parsons and Atkinson (1992) and Parsons and Clarke (1993). The earlier study looked at lay constructions of genetic risk and found fundamental differences between medical and lay understandings of the statistical issues involved. The authors express concerns over the possible important consequences these differences may have for the women's reproductive behaviour. The later study reported the same significant differences between lay and health professionals' perspectives, especially regarding the thresholds used to distinguish high and low risk. Some of the women, when quoting their risks in mathematical form, confused their carrier risks with their reproductive risks, and several did not retain their risk in numerical form at all. These women had translated their risk into a descriptive category which resolved it into greater certainty, e.g. not so bad.

This last finding highlights a concern expressed by Barbara Katz Rothman (seminar 17-5-95 Nottingham University) regarding the ambiguity of prenatal diagnosis and genetic testing more generally. Many of the respondents in her research had disclosed that they would only consider therapeutic termination in response to a positive result from prenatal testing if the disorder was 'severe'. It could be argued that if clients find risk difficult to comprehend, the ambiguity of testing may well be involved as a causal factor in this confusion. Mary Seller (1982b) warned about the potential misinterpretation that can be caused when talking about 'risk' or 'abnormal child'; "...it is important to remember that what one person thinks of as 'risk' or 'abnormal' is not necessarily what another may believe". This problem is surely only going to be heightened with the added problems of clients misinterpreting what health professionals actually mean, and the ambiguity surrounding severity which comes with testing for almost all disorders.

Once again, little research appears to exist on how risk is presented and received in actual counselling sessions, i.e. the *process* influences. However, there is a growing body of research which adopts a more theoretical psychological approach to this problem, discussing what lay risk assessment actually is, and attempting to elucidate the processes or cognitions that underlie this assessment. As in the previous section, some of these studies deal with research subjects asked to place themselves in hypothetical situations, whereas other use actual counselling clients. Kessler and Levine's (1987) study falls into the former group, using undergraduate students. The authors examined the subjective assessment of risks provided in fake counselling sessions as both percentages and proportions, and found that there was a significant difference in perceived magnitude between these two presentations, so that percentage figures tended to be seen as greater. Interestingly, the group of subjects that reported using a 'person strategy' to assess risk (i.e. visualising mental images of persons or parts of persons) were in contrast to the rest of the group, perceiving proportions as having greater magnitude. This study highlights the importance of linguistic framing, and the influence this can have on the processing and understanding of risk information. Similar results were found by Chase, Faden, Holtzman et al (1986) in relation to the influences of the numerical frameworks used. Other studies employing similar theoretical paradigms stress the importance of context for processing and memorising genetic information (e.g. D'Ydewalle and Evers-Kiebooms, 1987). However, all these studies use fake clients in artificially manipulated situations, and so their practical use is limited.

A study by Wertz, Sorensen and Heeren (1986) is one of the few found here which examines issues of risk assessment using counsellees in relation to real counselling sessions. This study also compares clients' subjective or qualitative assessments of risk (e.g. high, low) with the numerical risks presented by counsellors. Once again, discrepancies were found across the categories, but importantly, counsellors were also asked to make a qualitative assessment; the authors found that this discrepancy remained here, with clients' modal interpretation of a 15-50% risk figure being 'moderate', while for counsellors it was 'high' or 'very high'. Ekwo, Seals, Kim et al (1985), also working with a group of female clients in relation to actual counselling situations, also found a generally low correlation between objective and subjective risk estimates. These studies might be described as process research in that through using psychological models of learning theory, they attempt to show *how* as well as what risk perceptions are made, and to gain a deeper understanding of the mental process. Once again, however, how this information is conveyed interactionally is lost in the reporting of the studies.

To conclude this section there are four observations to be made. Firstly, it would seem that all these studies, whether pre- or post-counselling, have found that there is a minimal understanding by clients

of actual genetic risk. This is true even of those populations who might be thought to have some genetic knowledge through contact with family members with genetic disorders. The second common finding is that there is a high rate of confusion amongst lay people when it comes to translating medical information into something they can understand. This is evident in the different vocabularies of lay and health professionals when discussing issues like risk. The third point concerns the confusion which arises from different types of risk (e.g. carrier and reproductive), and the ambiguity that comes with the results. The fourth is that, in common with many of the studies in this area, no information is provided concerning how or what the clients were told as opposed to what the counsellors knew they were supposed to tell them or reported telling them. Does the confusion arise from a confusing message in the first place?

1.5 Psychological Effects of Genetic Counselling

The psychological effects of genetic counselling receive greatest consideration in the literature in relation to diagnostic testing. There are two distinct sorts of genetic test which may be carried out: predictive testing, where an individual is informed if they have inherited a gene which will cause them symptoms in later life (e.g. for Huntington's Disease); and prenatal testing. Prenatal testing may be further subdivided into two groups: those performed specifically for couples known to be at risk of early onset severe genetic disorders, such as CF; and routine antenatal screening tests which can identify abnormality in the baby. In these latter circumstances, delivery of the diagnosis/prognosis and other information is rarely in the hands of the geneticist, and will not be considered at length here.

Mennie et al (1993) researched the psychological effects of prenatal screening for CF. 1798 women accepted the offer of testing, and 64 were identified as carriers. The major finding from this study reveals that, compared to control groups who were identified as non-carriers at testing, both carriers and their partners showed a significant increase in generalised psychological disturbance which could be attributed to anxiety and depression. This was during a four day period when they were awaiting the test results of the partners. After the results had been disclosed both parties returned to their control levels.

Two further studies also report anxiety in their samples. Evers-Kiebooms et al (1988) investigated the psychological impact of amniocentesis in three different risk groups: mothers of advanced age, mothers who had a previous child with DS, and mothers who had a previous child with NTD. They found that anxiety levels in relation to the test differed between the groups, but also showed

considerable variation within the group. Overall, however, the evaluation of the procedure was said to be positive, and a high subsequent uptake was both stated and carried out.

Watson, Mayall et al (1992) examined anxiety in those being screened to ascertain whether they had CF carrier status. They report that this anxiety was mostly allayed by genetic counselling. However, since the screening process was simply to determine (asymptomatic) carrier status, which is not necessarily problematic in reproductive terms, this seems likely to have had an impact on expressed anxiety. Similarly, Livingstone et al (1994) detected no anxiety among a cohort of the population they screened for CF carrier status. In addition, they found that 99 per cent of the questioned participants expressed satisfaction with the concept of couple screening for CF. Clearly, different screening or testing programmes have different practical, moral and temporal implications for the participants, and so there are no easy comparisons to be made or conclusions to be drawn here.

However, what these results do imply is that, in any kind of testing, anxiety can be great during the waiting for results and following a positive result. This echoes one of Barbara Katz Rothman's concerns, that prenatal testing creates an unnatural and unnecessary 'emotional hell' which is caused by the availability of tests. As she points out, terminating a wanted baby due to unfavourable test results is not the same thing as what she calls an 'un-pregnant' termination, where a pregnancy is terminated regardless of genetic status because it was unwanted from the outset.

If we return to Tibben, Neirmeijer et al (1992), it is evident that the authors are reporting similar emotional problems in their subjects. Tibben and colleagues questioned those people who did take up the opportunity of pre-symptomatic DNA testing for Huntington's disease, and those non-participants who were at 50 per cent risk. They found that the ones who did attend were the ones who had predicted that they would not be emotionally affected by the outcome. They were also highly educated and had anticipated that the results would help them plan and control their own future. Overall, the non-participants had more fears and more negative attitudes about the possibility of a negative result. Whether or not emotional stress or anxiety is directly related to age or education level is unclear, and is not an issue which is addressed in the majority of the literature, but it may be an important factor in the kind of psychological reaction an individual will experience.

The final study to be considered in this section relates to the issue of what clients themselves report to be their influences and reactions to genetic counselling. Wertz and Sorenson (1986) had 628 clients complete questionnaires six months after their genetic counselling session. 273 (43.5%) of these reported that the session had had an influence on their reproductive plans. Again, it was found that

these clients on average had a higher level of education than others in the sample. However, the change and stability of reproductive plan patterns of both groups - self confessed influenced and not influenced - were similar. The authors discuss the possible reasons why some people report expert influence and others do not when their behaviour is similar (see Strong, P, M. 1980, for an interesting addition to this debate). They conclude, amongst other suggestions, that this response is related to social class. The problem, however, is that it is virtually impossible to measure the actual utilisation of the medical information given during counselling as a factor in eventual decision making.

Although the majority of literature in this area appears to relate to genetic testing, there are also some studies which focus directly on the counselling experience. Fisher, Rowley and Lipkin's (1981) study has been previously discussed in terms of its assessment of the impact of different 'formats' of counselling. However, this study also examines the psychological impact of counselling on mood. Mood change is assessed by using a psychological scale, both before and after counselling. The authors conclude that the use of different formats made no significant differences to mood changes, although there is no discussion of what these mood changes actually were. Cull, Miller and Porterfield (1988) carried out a similar assessment of differing counselling formats using the Spielberger State-Trait Anxiety Inventory, and also found no significance between groups. Similarly, Michie, Marteau and Bobrow's previously discussed study examines not only clients' expectations of counselling, but also psychological impact, again using the Spielberger Inventory. Here, the common sensical conclusion is drawn that there was reduced adverse psychological impact when patient expectations were met in the counselling session. All of these studies, however, are concerned with comparisons of counselling methods rather than the counselling itself. The only study identified for this review which has as its explicit focus anxiety levels as a result of counselling is Lloyd, Watson, Waites et al's (1996) work on familial breast cancer. The authors found that counsellees had higher breast cancer specific distress rates after counselling than before, despite being more informed. Once again, the potential for counselling to be a worrying or threatening experience is underlined. Yet to consider an issue which has not yet been raised, as Jarman (1982) points out, counselling can also have an adverse psychological impact on the counsellor. The interactional dynamics of delivering uncertain or unwelcome information are a two way process, and it is to studies considering the counsellor's perspective that we now turn.

1.6 Counsellor/Practitioner Knowledge and Attitudes

Despite the recognition of the psychological dimension of counselling for the *counsellor*, most of the work that appears to exist in this area focuses on the examination of professional attitudes and knowledge of genetic issues as they relate to service provision. More specifically, the focus is on

ethical issues arising in practice. Holtzman's (1993) study of practitioner knowledge and attitudes in the US, for example, reports not only that medical geneticists and genetic counsellors have a far greater knowledge of even the basics of genetic risk than primary care physicians, but also that non-geneticists were more likely to be directive given a hypothetical counselling scenario. However, it could also be assumed that genetic professionals would have a greater knowledge of what would be deemed ethically correct in this situation, and that their responses reflect this ideal rather than actual practice.

Burke's (1992) study also has a clear ethical focus, examining US genetic counsellors' attitudes towards fetal sex identification and selective abortion through interviews. Overwhelmingly, non-directive counselling was endorsed, and the use of prenatal diagnosis for sex selection purposes was condemned. Interestingly, when client views conflicted with their own, counsellors reported using psychological mechanisms to cope, such as redefining the category of 'unwanted pregnancy' to include 'wrong sex'. The possibility of differing cultural values is also highlighted: this is echoed in Wertz and Fletcher's (1988) and (1998) studies of sex selection in 18 and 37 nations respectively. Again, the conflict for counsellors between maintaining personal integrity and serving their clients needs is stressed. Differing counsellor characteristics are also assessed in these terms, e.g. Wertz's (1993) study which discusses the impact of gender on ethical decision making in the provision of genetic services, and concludes that women are likely to be less directive than men. Unfortunately, all these studies are based on *reported* action, so the relationship between what people do and what they say they do remains unknown. In addition, with the exception of Burke's study, and Wertz's 1993 work, all of this research involves invoking hypothetical situations. There is little debate that genetic counsellors are *aware* of the ethical dilemmas involved in the provision of services, but it would be interesting to see how they deal with these difficulties in actual practice.

1.7 Potential Clients

Another group of studies have tried to predict the take up of particular genetic services by focusing on more narrowly specified groups seen as potential users. These groups were then questioned about whether they would use any of the services available. There has, however, been a much lower uptake than these studies predicted, which raises the question of whether the right groups of people were targeted for the research. Did the projected clients actually see themselves as such? This section considers who the assumed potential clients for genetic clinics are and some of the reasons researchers have identified to explain the unexpectedly low uptake.

The studies in the first group of this section have concentrated on predicting the take up of particular genetic services. The second group have tried to find explanations for why the take up was much lower than the first studies suggested.

All the studies in this first group have relied upon either interviews or questionnaires for their data collection. The target populations are extended family members of an affected individual. These people are all either at risk of being a carrier of the disorder in question and/or of having an affected child.

One study (Swerts 1987) divided the subjects into three groups: one having received genetic counselling; one having had an amniocentesis performed; and the third group having neither. In this study the participants either had a child with Down's Syndrome or with a neural tube defect. Swerts reports that the information given during counselling influenced more than half of the parents of children with DS, and 80% of the parents of children with NTD, to plan another pregnancy. She also states that for more than half of the families with a child having either disorder, the availability of prenatal diagnosis was of crucial importance in the decision to plan future pregnancies. The considerable proportion of parents who rejected amniocentesis most often cited moral convictions or fear for the risks of the procedure as their reasons. So, it would seem that those who received genetic counselling did not necessarily go on to use the services available for their own particular needs. It appears that a better, if less concrete, measure of the efficacy of genetic counselling might be the proportion of clients who feel empowered to make their own personal decision as a result of the counselling process.

A second example in this first group of studies is by Decruyenaere et al (1993) who focused upon young women's attitudes towards predictive testing and prenatal diagnosis for Huntington's disease. The authors found that, although half would consider prenatal diagnosis, only one quarter would consider termination following a positive result. In a large-scale Belgian survey, Evers-Kiebooms et al (1989) found an even higher proportion of at risk persons and their partners (66 per cent and 74 per cent respectively) intending to make use of predictive testing for Huntington's disease. Although Evers-Kiebooms and her colleagues found that the motivations behind their respondents' decisions were very complex, they could specify that they were not related to socio-demographic characteristics.

Of course, someone does not have to undergo diagnostic testing to be defined as a user of genetic services. From these studies, however, we can see that researchers expected the potential clients of genetic services to be those individuals who were already known to be at risk for one reason or

another. All the studies predicted that there would be a fairly high uptake of specific genetic diagnostic tests by this target population. However, what people say is not necessarily the same as what they will do, and in particular the link between attitudes and behaviour is complex and poorly understood. In the event, all of these studies were subsequently proved wrong by a much lower uptake. However, there are significant differences between predictive testing for late onset disorders such as Huntington's disease, and prenatal testing for disorders which will have a lifelong impact, e.g. Downs Syndrome. Prenatal tests are very rarely performed for adult onset disorders, since this would involve the consideration of therapeutic abortion as a solution to something far in the future. This point takes us straight to the results of the first study in our second group.

Adam et al (1993) found that demand for prenatal testing for Huntington's Disease amongst 425 at risk couples in Canada was only 18%. The most frequently cited reason for this decision was the hope that a cure would be found in time for their children. A lower percentage of actual patients reporting an influence on their reproductive planning was apparent in Eggers et al (1993) study of people with Facioscapulohumeral Muscular Dystrophy (FSH). In this small study of 46 patients, most stated that they would have liked to have known their diagnosis earlier, for example in order to prepare themselves emotionally, or to choose an appropriate profession. Although they were generally in favour of the option of prenatal diagnosis, only 2 said they would consider therapeutic abortion in response to a positive result. Genetic counselling seemed to have little influence on family planning and the authors report that AFSH does not seem to reduce reproductive performance in our population. Whilst the possible reasons for this are not explored in this study, these patients may be seen to be in something of an expert position, since they know first-hand the impact their condition has on everyday life, and are able to make reproductive decisions on this basis.

Tibben, Niermeijer et al (1992) discovered that the few participants who did actually take up the opportunity for predictive testing for Huntington's disease in their study were more highly educated than average, and had already anticipated that they would not be emotionally affected by the outcome. Instead, they had the test for similar reasons given by the group with FSH: in order to control and plan their own future. The same study found that those who did not participate in the testing (who were also at 50 per cent risk) seemed to express more fears and more negative attitudes about the possibility of a positive result.

Summarising these studies there are three main observations to be made. Firstly, the nature of the disorder, the kind of testing, and the possible support or treatment services available seem to be crucial factors in decision making. In a study by Houlston et al (1990) much higher responses to

screening were found among relatives of patients with colorectal cancer, where there is a recognised medical treatment regime, for example. If the disorder is late onset, such as Huntington's disease, couples may turn down prenatal diagnosis on the basis that a cure may be found before their child develops symptoms, or that it is impossible to make a decision based on a temporally distant outcome. The decision to undergo prenatal diagnosis for a condition with no potential for a cure (e.g. CF), or for treatment (e.g. Down's Syndrome), appears to be strongly linked with peoples attitudes to abortion, with some individuals rejecting the testing as a result, and others stating that the test will help them to plan or give them control.

Secondly, the perceived severity of an illness influences people's reproductive planning. In particular, experience of an illness relates to perceptions of severity. As we have seen above, individuals at risk of FSH were not overly concerned about restraining their reproductive behaviour because of this fact.

Finally, a person's educational level and attitudes towards information about their carrier status affects their decisions to utilise genetic services. However, a person's educational level is not a predictor of whether and to what extent they will be affected emotionally. People who predict that they will not be negatively affected emotionally by the information are more willing to make use of the services available. This again raises the issue of how individuals receive and handle the information delivered by a genetic counsellor, which will be discussed in more detail below.

1.8 Process Research in Genetic Counselling

Throughout the literature previously examined, we have drawn attention to the need for research on the *process* of genetic counselling. This final section of the first part of this review considers the process research which has been carried out thus far, in order to suggest a way forward.

Once again, a wide variety of methods have been used in this area. What authors have in common is recognising that the focus on *outcome* measures alone is of little use for the development of practice, in terms of evaluating and specifying training needs and issues. In common with other genetic research areas, a psychological theoretical framework underpins the majority of the work here, e.g. Kessler's (1981) analysis of a transcript of a counselling session. The author explicitly states that his motivation for research is the limitation of outcome studies which results from a lack of knowledge of the actual content, structure and dynamics of the session evaluated. He presents a transcript of a pre-amniocentesis genetic counselling session, which is analysed in terms of such issues as procedures used by the counsellor, the style of counselling, and the nature of counsellor interaction

with counsellees. Kessler concludes that the session was content-oriented, and that the counsellor avoided or evaded effective issues. He also identified elements of directiveness. However, it is important to remember that this is a study of only one consultation, in relation to a particular genetic service. Whilst it may provide issues and pointers for training, a larger body of research is clearly needed.

Transcript analysis also features in Kessler and Jacopini's (1982) study, but this time the analysis is quantitative, building on hypotheses developed in the previous study concerning counsellor style and behaviour. The Bales system of scoring interaction is used, which involves the applications of categories and scores to small subsections of the transcript. These categories include 'positive' and 'negative' reactions, questions, 'agreement', etc. However, the end result is that, through use of these scores, we are taken further away from the actual interactional processes that Kessler himself suggests are so important: findings are reported such as "the counsellor initiated 54% of all units", but it is not clear for example whether there was client resistance to this on any or all occasions.

Transcripts are again used to slightly different ends by Kessler, Kessler and Ward (1984). Here, both actual and potential counselling situations are explored and discussed in terms of what is done by the counsellor and what should be done. Interventions for effective management, in particular in dealing with guilt and shame, are discussed, and a number of tactics for dealing with these issues are suggested, such as use of authority, normalisation and limiting liability. However, some of these situations are completely hypothetical, and it is unclear what advice is given to the counsellors on *how* to incorporate these tactics interactionally. Once again, the focus is shifted from the two way interactional processes of *actual* counselling sessions.

One difficulty in exploring actual sessions is how to record or preserve these. Van Zuuren's (1997) investigation into neutrality, and Van Zuuren, Van Schie and Van Baaren's 1997 work on uncertainty both examined verbal exchanges in 30 counselling sessions, as they were recorded in writing by a psychologist. The choice of methods, however, raises issues of validity and reliability, particularly in terms of the influence of the interpretation of the transcriber. The authors themselves advocate the use of audio tape for future validation. Additionally, in both cases, material deemed to be relevant to the research question was extracted from the body of the consultation, raising issues about the context of interactional exchanges. The results:- that there is a bias towards the use of genetic services, and that uncertainty is a factor in client stress- are consistent with outcome research studies discussed earlier.

The remaining studies to be discussed in this section all make use of audio or video recordings. Whitten, Thomas and Nishiura's (1981) evaluation of sickle cell trait counselling involved the audio recording of 193 structured counselling sessions. Once again, the authors' stated aim is to address the lack of knowledge about content, quality and instructional activities of counsellors which results from outcome measures alone. Their quantitative analysis rates counsellee success in acquiring information on 10 components defined as key aims of this particular counselling programme. This was assessed by ascertaining whether counsellees' answers to a number of questions asked were satisfactory. However, it is unclear whether the authors' conceptions of what is satisfactory correspond with those of the counsellees. Adequacy of counsellor performance was also assessed in relation to information transfer. This study represents a practical evaluation of an actual programme, although once again its focus is on specific components of the counsellor's role. In addition, the authors' determination of what is to be treated as satisfactory raises questions about how this relates to counsellor and client experiences of the process.

Specific aspects of counselling were also investigated by Mendez and Shymansky (1984), using video tape as a mechanism to highlight physician verbal patterns and behaviours during 49 counselling sessions. The authors stress the importance of analysing communication as a chain of events, rather than abstracting single events and considering them in isolation. Using tools called the Physician Communication System (PCS) and Interviewing Attributes Questionnaire (IAQ), encounters were categorised and coded, and physicians were categorised into groups according to their score. The results show differences in verbal behaviour and verbal patterns as analysed by IAQ group. Although some of the practical details of this are then discussed, (e.g. physicians with low scores are more prompt to emphasise cognitive aspects of the interactions, and to avoid silence) this has the effect of distancing the reader from the actual interactions. Once again, there is no suggestion of how these variations were received by clients, or how physicians who have low scores might acquire better skills in practice.

Chapple and May's (1996) work using two case studies to examine genetic knowledge in the context of family relationships also makes use of video data. The authors claim that their use of ethnographic methods enables not only detailed insight into family relationships, but also examines this in relation to the actual process of counselling, without transforming it through application of a coding tool or pre-defined scheme. With the co-operation of 30 families, consultations were video-recorded, and post-counselling interviews were also carried out both with the counsellor and the client. Counsellors were asked a number of questions relating to what their aims and intended achievements for the session had been, and whether or not they felt this had gone well. Clients were asked about their expectations

of the service, whether they had any concerns about the consultation, and their views of what had taken place. They were also re-interviewed after 6 months, and asked whether the consultation had been helpful and whether there had been an effect on family relationships. The discussion here focuses on two cases, a child with fragile-X syndrome and a mother and children identified as carriers of Patau's syndrome. Analysis suggests that psychosocial issues such as guilt, stigmatisation or possible damage to social relationships were rarely discussed, and the authors conclude that genetic counsellors must be trained to deal with these issues as well as diagnosis and risk calculation.

A further publication using this body of data (Chapple, Campion and May, 1997) looks at the use of clinical terminology and its effects on anxiety and confusion of clients. The findings here suggest that the language used in counselling is often confusing and misunderstood by families, with unfamiliar terms conjuring up alarming images. The practical implication is that counsellors should try to use simple, understandable language, and give clear explanations of unfamiliar terms that cannot be avoided. A careful choice of words, the authors suggest, can reduce risks of labelling and stigma, but can also prevent unnecessary anxiety experienced by clients when they hear unfamiliar medical terms, such as eponymous syndromes, employed in diagnosis. This study not only provides practical resources for counsellor training, but also demonstrates the fundamental significance of the communication process. Clearly, further research is needed which demonstrates this connection between process and outcome variables.

2. THE WAY FORWARD?

This part begins by summarizing and presenting some of the issues raised by the research discussed in Part 1. It then considers some of the most relevant concerns emerging from the theoretical literature. Section 2.3 then discusses some research undertaken in comparable fields using conversation analysis and ethnographic methods. Conclusions are then drawn about the way forward for research in the area of genetic counselling.

2.1 Issues Raised by Research

This section reviews the issues which previous researchers into genetic counselling have most frequently addressed or discussed.

One recurrent issue relates to the transmission of information to family members (Suslak et al. 1985). It seems that there is some controversy over who exactly should be responsible for this task - should it be the duty of a professional or left to the individuals themselves? This is of particular importance when it comes to targeted screening programmes. If an affected individual has decided not to inform other family members about their condition or status, should the professional involve them in the programme? The matter can be further complicated by the finding that people are well aware of the potential problems which can arise with insurers and employers as a result of knowing one's own status (see Decruyenaere 1992b).

The second issue is directly related to the problem of the somewhat ambiguous role of the genetic counsellor. This is still controversial, and there is certainly public confusion over the aims of the new genetics. Researchers have found that people are potentially interested in specific genetic tests through prenatal diagnosis. However, this does not necessarily indicate willingness to consider terminating a pregnancy (Eggers et al. 1992; Evers-Kiebooms et al 1993; Denayer et al 1992b; Decruyenaere et al 1993). If *pretest* counselling is not always offered, the clients may not be fully aware of the consequences of participating in testing.

Researchers have commented on the insufficiency or inadequacy of available counselling (Frets et al 1991). There seem to be issues emerging about its timing and the appropriate recipients. Petra Frets points out how problems in the decision making process often become apparent after, rather than during, the counselling session. She argues, therefore, that counselling cannot be seen as a discrete process, and that follow up counselling should be increased. Tibben et al (1992a; 1992b)

suggest that non-carriers and partners of carriers tend to receive less attention from counsellors, but that they may still have specific counselling needs related to survivor guilt.

The extent to which people actually understand the counselling they are being given remains obscure. Clearly, the counsellors' task is set against a background of a general lack of knowledge in the population as a whole (Decruyenaere et al 1992a), although a lack of knowledge or counselling does not necessarily result in a reduced intention to use diagnostic genetic services (Swerts 1987). After exposure to counselling, individuals often do not recall their own risk accurately (Swerts 1987; Evans et al 1993). There is repeated evidence suggesting that people reach a subjective understanding of the information they receive which does not necessarily accord in any mathematical or objective way with the information thought to have been transmitted (Denayer et al 1992a; Swerts 1987). This subjective interpretation is the basis on which people's reproductive decision making actually seems to rest (Parsons and Atkinson 1992; Parsons and Clarke 1993). Parsons and Clarke believe that the gap arises from fundamental differences between lay and professional perspectives about the meaning of risk, especially when this information is given in statistical form (see also Huys et al 1990). Added to this `vocabulary gap', is the real complexity of the science involved in the new genetics. This is expressed in the differentiation of the types of risk involved and the uncertainty about the severity of any genetic abnormality detected through testing (Katz Rothman 1994).

Many studies were intended to predict the take up of particular genetic services (e.g. Swerts 1987; Decruyenaere et al 1993; Evers Kiebooms et al 1989), although in some studies it is unclear what level of counselling was involved. Whilst some of these studies clearly have their focus on diagnostic testing, it might be better to conceptualise uptake of genetic services in terms of the number of individuals seeking information about their own risks, regardless of whether they then decide to have a specific test. As we have noted, these studies tended to overestimate test uptake. One reason for this could be that researchers targeted the wrong groups of people for questioning. Another could be that people may, in some circumstances, prefer uncertainty to certainty. These studies showed a reluctance to consider abortion in the hope of a cure in time for late onset disorders. However, since much of this research is Belgian, and appears to involve a considerable proportion of Roman Catholic respondents, this may in part be an artefact of the sites where they were carried out. Given the scientific uncertainty about the interpretation of test results and the psychological disturbance that can be caused by testing, it seems that the kind of clear-cut services people actually want are not yet available (Eggers et al. 1992; Evers-Kiebooms et al 1993; Denayer et al 1992a; Decruyenaere et al 1993; Adam et al 1993; Katz Rothman 1995; Livingstone et al 1994; Evers-Kiebooms et al 1988; Watson 1992b; Mennie et al 1993). The demand for testing may be quite selective until cures or

effective treatments become available, rather than relying on selective termination or reproductive decisions.

This becomes clearer when we look at the other survey evidence. A large proportion of researchers pointed out the correlation between the respondent's age and education level, and decisions made, attitudes towards and/or knowledge of genetic services. For example, those who decided to use genetic services in a study by Tibben and Neirmeijer (1992) had a higher education level and predicted that the results would not affect them emotionally. They believed that the information would help them to control their own future. On the other hand, those who decided against testing displayed fear and a negative attitude towards the possibility of a positive result (Varekamp et al 1993; Decruyenaere et al 1993; Wertz and Sorenson 1986).

2.2 Common Concerns in the Literature

This section focuses on some of the main issues to have been addressed in the theoretical literature. We begin with the debate surrounding the role of the genetic counsellor. The majority of people who come into contact with genetic counselling have an opinion on this issue at some level, but the authors discussed here have made particular contributions to the debate.

Central to discussions about the role of the counsellor is the problem of directiveness. There is an inherent tension in genetic counselling between the counsellor as a non-directive information giver and as someone who may be seen by clients as capable of giving them an expert opinion. In addition, whilst counsellors may be clear that they are not bound by a mission to promote certain decisions which are in accord with the presumed values of a society or of some other normative code, clients may be less so.

According to Clarke (1991a; 1991b) a non-directive counsellor should avoid openly suggesting termination as a response to a positive test. If the termination of an affected fetus is treated as an explicit goal, then non-directive counselling has been replaced by public health policy or eugenics. Others disagree, on the grounds that this still involves making decisions for people. This is not the geneticist's job as he or she is not the one who has to live with the consequences of an affected child. Clarke goes on to criticise counselling practice by arguing that the offer of prenatal diagnosis carries a built-in recommendation to terminate in case of abnormal results. In response to this Super (1991) argues that geneticists do not offer prenatal diagnosis but simply discuss the possibility. Clarke concludes that geneticists must recognise their responsibility to communicate the full costs of prenatal

diagnosis to clients and health service management, in addition to caring for individuals affected by genetic disorders. In response to this suggestion, Pembrey (1991) insists that medical and social care is not the responsibility of the clinical geneticist.

However, Clarke's argument is in line with those researchers who have pointed out the need for more counselling to be available at all levels, but specifically at the pretest stage. However, this would not solve all potential problems in the clients decision making process. Many future parents would only consider termination as a solution to a positive result if they could be sure that the disorder is severe. As Morrison and Nevin (1991) point out, factual advice is often not possible as physical or mental impairment varies, as does an individual's assessment of severity.

Wertz and Sorenson (1986) also stress the importance of non-directive counselling but argue that counsellors are often unintentionally directive, especially with vulnerable, poor and less educated clients, or clients who may actively be seeking solutions. Raeburn (1994) is another to emphasise the importance of non-directive counselling. He describes this as the counsellor being honest about his own position, which he calls a kind of `formal neutrality'. This involves the counsellor avoiding any directive statements but still exerting a considerable steer on the outcome. In practice the counsellor aims to befriend the clients and encourage them to talk from an early stage in the session.

Exceptions to the non-directive principle, in Raeburn's opinion, include cases where there is possible danger to the person, to the person's family members, or where he feels that not testing constitutes the better option for those involved. He realises that this directive counselling may also at times lead to a breaking of confidentiality if persuasive tactics fail. The Nuffield Council also advise directive tactics, and revise their rules on confidentiality when family may be at risk.

One definition of genetic counselling is set out in a paper by Linhout et al (1991) for the American Society of Human Genetics. Genetic counselling is described as a communication process that deals with problems associated with the occurrence, or the risk of recurrence, of genetic disorder in a family. This, the authors claim, is not essentially different from definitions and attitudes in most European countries. However, this is not to suggest that there are not specific cultural problems which can arise as a result of genetic counselling in some societies. Carmi (1991) and Naveed (1992) both report their experiences of these problems. Carmi cites the case of testing for Maple Syrup Urine Disease in Bedouin Arab society in Israel. The high incidence of the disease was explained in terms of consanguinity, but as counselling continued, a social problem emerged: potential male carriers were looking outside of their tribe for wives. Since marriages are traditionally pre-arranged amongst this

group, this created problems for families who were then left with a dependent adult female. Naveed (1992) reports the case of a Muslim husband anxious to know if his wife alone could be responsible for the occurrence of thalassaemia in their child, so that this could be used as grounds for a divorce. He also presents a similar occurrence in a Hindu family with a child suffering from Duchenne Muscular Dystrophy, as an indication of how the knowledge imparted in genetic counselling sessions may be exploited or misinterpreted. Evidently, these kinds of situations are likely to be more problematic in X linked recessive and autosomal dominant disorders, where the wife is the carrier of the trait. Commenting on Carmi's experience, Borgaonkar (1991) emphasises the necessity for genetic counsellors to take into account societal factors in conducting premarital or preconceptual counselling, not only in a traditional society but in any society.

It seems to be the case that the genetic counsellors' role cannot be discussed in isolation from the debate over the aims of the new genetics. This issue is still unclear and controversial, but due to the level of the technology available, in most prenatal testing situations it is difficult to offer any intervention other than abortion. Indeed, there are those who openly advocate the goal of using the new genetics to seek an actual reduction in the number of births with a genetic disorder. Of course, this support will vary with the nature and severity of the disorder. Health Authority screening programmes, however, are more commonly concerned to study the early natural history of a disease (before clinical presentation), and to provide genetic counselling before another child is conceived (e.g. Green et al 1993).

A further major concern regards the financial aspect of genetic counselling. This inevitably affects the definition of the aims of the new genetics and the counsellors' role. Raeburn, for example, emphasises the difficulty in defining exactly what genetic counselling is, partly due to its rapidly changing nature, but also due to economical considerations. A typical process used to involve something like the following:

1 - patient enters clinic via referral --> 2 - information gathering home visit by genetic counsellor --> 3 - patient sees geneticist at hospital. (Raeburn 1994)

The second stage in this process is now regularly neglected for financial reasons.

The dependency of genetic counselling on public funding has led to pressures to find ways of measuring the work of counsellors with quantitative tools in order to determine its efficiency and cost-effectiveness. The problem of identifying suitable outcome measures has drawn the attention of many

authors in the field. Chadwick (1993) argues that the recent emphasis on efficiency in health services, and on finding ways to measure this, has given a central place to the issue of what counts as `success' in genetic counselling. Clarke (1994) suggests that this should be based on indicators of workload rather than, say, the number of pregnancies terminated as a result of counselling or the contribution to some national eugenics target. Chadwick supports this argument but argues that a measure of workload is insufficient because the goals of genetic counselling must be connected to the genetic health of the population. Clarke goes on to dismiss the idea of cost-benefit analysis of screening as unacceptable. He believes that the secondary effects of screening, such as the inadvertent diagnosis of unsought conditions, and the social effects on the disabled must be assessed before they can be evaluated.

Before concluding this section it is important to note the extensive literature relating to the ethical aspects of genetic counselling (for a detailed discussion on the ethical and legal implications of the new genetics see; Wertz 1992). The most common focus for attention has been the possibility of eugenic interventions. Previous abuses of genetic information have set the stage for genuine concern about the direction of the new technology. This is reflected in Clarke's concern that the goal of the new genetics should not be the reduction of births affected by genetic disorders. However eugenic goals are seductive, especially when supported by the pragmatic argument that fewer disabled births will reduce both private and social costs and the unhappiness of victims, families and the communities in which they live. There seems to be support for screening programmes among the respondents in the studies presented here (e.g. Decruyenaere et al (1992b), but only if usage is through individual choice rather than mandatory regulation.

In response to these ethical concerns, the Nuffield Council emphasises the voluntary nature of the screening process. It recommends that adequately informed consent be obtained for all genetic screening programmes. It suggests that information supplied should cover the implications for other family members, and should be delivered in both oral and written forms. Genetic counselling should be available to all being screened at all levels of the process.

2.3 Conversation Analysis and Ethnographic Research

At almost every point in the research and policy debates examined, the issue of the delivery and understanding of information from counsellor to client is evidently important. Was an individual influenced by the information gained? What do people understand about their risks? What psychological effects does the information have on people? How well is information transmitted within

an affected family? How successful is the counsellor in aiding the client in decision making? All these questions, and more, depend at least in part, on understanding how individuals receive and handle information, and how counsellors deliver it.

This section presents some studies which have used either conversation analysis or ethnographic methods to look at similar issues. Most have not examined genetic counselling settings but have studied comparable medical and therapeutic settings.

Before considering these studies, it will be helpful to sketch out how these methods, particularly that of conversation analysis, actually work.

As mentioned above, previous researchers have regularly begun their studies with prior assumptions about the nature and aims of genetic counselling. Qualitative research tends to be inductive, open-ended and discovery-oriented. Both these qualitative methods, and especially conversation analysis, aim to operate without *a priori* assumptions about what the data will reveal.

The distinctive characteristic of these methods is their concentration on actual episodes of face to face interaction and the details of participants' talk and activities within a given setting. In particular, conversation analysis is concerned with both the contextual sensitivity of language use and talk as a vehicle for social action (Drew & Heritage, 1992). That is, it is based on two important principles which differentiate it from other forms of social or language analysis. Firstly, it emphasises and studies the ways in which talk in any setting is both context-shaped and context-renewing. And secondly, it focuses on the interactional accomplishment of social activities. In this way it can identify sequences of talk as relevant to a particular setting, how they contribute to creating that setting, and finally how they achieve the social activity in hand.

Conversation analysts work with audio or video tape recordings of the exchanges between participants in an interactional situation. These recordings are transcribed using a notation system aimed at preserving as much detail as possible. Analysts work with both the recordings and the transcripts. This process of immersion in the data allows researchers continually to refine and improve the level of detail in the transcription. At a second stage, the researcher tries to identify emerging patterns in the interactions examined, later comparing these categories with the data. In this stage its units of analysis are sequences of activity and their component unit turns as turns-within-sequences. From these sequences, at a very simplistic level, the analyst can determine how a particular utterance is understood by its treatment with reference to the preceding utterance. In all published studies the

segments of talk are presented to the reader, thus allowing the analyst to demonstrate the findings, referring directly to the actual speech exchange.

Conversation analysis was initially applied to mundane conversations in different everyday situations. More recently it has been applied to institutionalised settings, such as news interviews, political speeches, mediation sessions, and the like. More interestingly for our purposes, a number of studies have examined the interaction between patients or clients, and public sector professionals. A brief overview of some of these studies can therefore provide some useful insights in the context of genetic counselling encounters.

The first paper which presents some relevant points, by Strong (1980), is a critique of a study looking at medical care and public attitudes towards physician authority. This quantitative study (Haug and Lavin, 1978) looks at factors such as age and educational level in relation to challenge to physician authority. Drawing on his own ethnographic work examining the physician /patient encounter, Strong highlights the limitations of this kind of approach to the study of interaction, and in particular the ways in which power and authority may be subtly manifested by the doctor. Responding to this, Haug and Lavin (1978) cite increases in complaints against British doctors as evidence for the growth of patient challenge to medical authority. However, these complaints typically occur after consultation and treatment, and not in face-to-face interaction with the doctor. In response to this claim, then, Strong quotes other British observational studies of doctor patient interaction which all emphasised the heavy medical control of the interaction and the highly tentative and indirect fashion in which such challenges were typically made (see Bloor 1976; Stimson and Webb 1975; Strong and Davis 1977, 1979).

However, this kind of ethnographic work has also been challenged from elsewhere in sociology for its inability to identify any particular action as being specifically relevant to a medical encounter. Sharrock (1979), for instance, argued that an 'interactionist' analysis of doctor/patient interaction can only tell us that the encounter is readily recognisable as medical work. Although the actions of the participant are negotiated and easily intelligible as medical, the organisations identified may be expected to occur in all kinds of social relationships. 'If we do want to isolate 'tactics' that doctors and patients use to assert or challenge each other's autonomy we shall only be leaping to premature conclusions if we do single out episodes of interaction without a clear awareness of the way in which those episodes are engaged in doing medical work' (Sharrock 1979: 144).

There is, of course, much more to this debate than there is space to present here. In short, however, it is sufficient to say that the challenge is, in part, unfounded. If an ethnographer finds through his or

her studies that authority is present within the interaction, it is not necessarily furthering their argument by attempting to find that this interactional strategy is exclusively medical. It is, however, partly to take the argument further, that a combination of ethnographic and conversational analytic tools is advocated in a study of the genetic counselling.

Heath (1992), who also documented the asymmetry evident in the doctor/patient relationship, used conversation analytical tools. He argues that asymmetry is interactionally achieved by both parties. A further study in this area by ten Have (1991), concludes his discussion by arguing that it takes specific and deliberate effort on the part of the patient to counter the interactional contingencies leading to asymmetry, and that this is rarely seen in practice.

These studies bear on the issues of non-directiveness, and the delivery of information generally. Genetic counsellors are, as far as the general public is concerned, representatives of medical authority and may find it hard to escape the weight of client expectations of medical control in such an environment. They then have to prepare the client for possibly making very serious reproductive decisions.

Further to this, clients are also likely to arrive at a genetic clinic expecting the medical professional to advise and direct the clients' course of action. This raises the issue of how clients adjust to a situation where they are being offered choices based on ambiguous information, as opposed to diagnosis followed by treatment. Using conversation analysis Greatbatch and his colleagues (in press) have shown that clients in genetic counselling settings prefer certainty to uncertainty. Clients are there to seek a diagnosis and do not respond well to uncertain conclusions.

The counsellor thus has to turn these expectations around and resist the clients' explicit or implicit demands for certainty. Given client expectations, how do counsellors present their position differently from other medical encounters, and how do clients demonstrate their adjustment or lack of, interactionally for the counsellor to assess client understanding?

On the other hand, the clients may be more knowledgable about genetic terminology than the general public if they are members of a sufferer's family. They may be in a position similar to that discussed by Macintyre and Oldman (1977), who argue that sufferers can acquire a subtly superior and special knowledge to that which is held by professionals, by virtue of experience. How is this negotiated within the interaction by both parties, and is this something the counsellor could draw upon in encouraging autonomy of decision making?

Another relevant study, by Gale and Newfield (1992), analysed a group of solution-focused marital therapy sessions. The authors aimed at understanding how the process of facilitating changes in client attitudes was interactionally achieved. The analysis showed how the therapist had to adjust and adapt his actions to fit with the husband's and wife's actions. An understanding of the clients' agendas was achieved through a detailed analysis of their communications. Through the analysis the authors also demonstrated how the therapist's model was effectively put into practice in the course of the session, and how further themes can emerge in addition to those already identified in the model.

In a more medical setting, Mallet (1990) explored verbal and non-verbal communications between nurses and post-anaesthetic patients in the recovery unit of an inner London hospital. Using conversation analysis, the findings are intended to have wider implications for intensive care, even though patients had only experienced minor dental surgery. The utterances and body movements of the nurses to engage patients in conversation were analyzed. The paper concludes that nurses are skilled communicators who take into account the patients' low level of consciousness and lack of physical orientation.

Scholz (1992) is the author one of the few papers identified and not already discussed which is specifically dedicated to the analysis of the process of genetic counselling. She argues that one of the most important tasks in genetic counselling is giving assistance in making decisions about family planning and the utilization of prenatal diagnosis. The study of genetic counselling should focus on decisions and the factors which may affect them. She criticizes current concepts of `decision' as they are applied to prenatal diagnosis, using examples from naturally occurring conversations between genetic counsellors and their clients. She argues that decision-making is a complex interactional process. It is neither directed by the counsellor nor a dialogue where topics introduced by clients are equally welcome and accepted.

Scholz takes up Wertz and Sorenson's (1986) work which showed that questions in genetic counselling were not standardized. Rather than treating this as a problem that could be solved by better training, she argues that they are being designed to elicit agreement from the patients to seeing themselves as needing to be involved in some action rather than merely in the receipt of information. Scholz continues by restating the research problems in the study of decision making in genetic counselling:

1. How is it that questions and answers are intertwined in such a way that they constitute a problem of choice, a choice among solutions or a decision sequence?

2. How is the meaning of the decision sequence made available for inspections by either participant?
3. In what ways is this sequence sensitive to the social situation of genetic counselling?

She claims that a detailed analysis of the interaction between client and counsellor demonstrates that the participants' primary concern is not a decision for or against prenatal diagnosis. They are mainly preoccupied with the counselling encounter and with working out what it is appropriate to say to each other in that context and what the other's responses might mean.

2.4 Future Research In Genetic Counselling

So far we have seen how important issues and debates are founded upon an inadequate understanding of what actually occurs in genetic counselling encounters.

Although most of the research into genetic counselling to date has provided us with some useful information, the methodologies it has most often employed can only produce partial answers. For example, it has informed us that clients often reinterpret their risks and reach subjective understandings of the information they reportedly receive. It cannot tell us why this occurs or what exactly happens in the production of two different versions of the counselling information which are then held by counsellor and client. That is, it cannot, for the most part, tell us anything about the actual process of the counselling encounter, something which needs to be understood if we are to answer questions regarding the delivery and receipt of information. After all, genetic counselling is defined as a communication process (Lindhout et al, 1991), and can only be fully understood when studied as such.

In the first section it became apparent that the major issues concerning most researchers in the area were related to, in one way or another, the effect genetic information has upon the clients of the services. It was also evident that researchers to date had failed adequately to address these issues because their research methods had provided inadequate data. One set of problems arose because of the methodologies used. Surveys and questionnaires rely on the recall abilities of the respondents and therefore, on their *post hoc* interpretation of events. A further difficulty arose when people were asked to predict their behaviour and attitudes given a hypothetical situation. All this has led to confusion about the nature of the data collected, which in turn has created difficulties when attempting to draw consistent conclusions about the major issues addressed.

Studies show that, in some circumstances, (e.g. adult onset disorders) people show a reluctance to consider the termination of pregnancies, even if showing an interest in predictive testing. In these cases, then, genetic counselling may have little influence in controlling genetic disease. However, this presumes that the reduction of affected births is a significant objective, which is itself controversial. More broadly, since research has reported how lay and professional perceptions of major issues such as risk are often different, it is difficult to assess the actual influence of genetic counselling on reproductive behaviour. It seems that in order to assess the actual effect of genetic counselling, a more appropriate method would involve a detailed analysis of the interactional processes. How exactly are decisions facilitated by the counsellor? (Gale and Newfield 1992; Scholz 1992). Conversation analytic or observational studies of medical encounters (doctor-patient communications) will help us in answering these questions.

In section two the main concerns grew out of the ambiguity surrounding the counsellors' role. If, for ethical purposes, the counsellors' role is defined as the delivery of sufficient information to enable the client to make and informed decision, it is essential that we gain a clearer understanding of the counselling encounter. For example, accepting the finding that clients all too often confuse crucial issues such as risk, then it could be argued that the client has not reached an informed decision. This situation begs the question of where ethical responsibility lies. Ethical responsibility only moves from the clinic to the client if the client fully understands the issues leading to their decision. Thus the medical profession also has an ethical responsibility to ensure that the client is in a position to take on and deal with such ethical and moral decisions.

In order to avoid such situations a full understanding of the delivery and receipt of information, and a clearer definition of the counsellor's role are called for. In the last section we have seen how these sorts of issues have been successfully addressed in comparable settings with the use of conversation analytic and ethnographic tools. There is now a growing body of knowledge in conversation analysis, from work carried out in similar settings, that could help us to gain an understanding of genetic counselling. Research using both ethnographic and conversation analytical methodologies in this area will provide us with a clearer understanding of both the counsellors' actual behaviour, and the clients' level of understanding.

Finally, the results of a study of genetic counselling situations using a combination of ethnographic and conversation analytic tools will have applications in training genetic counsellors. As Chapple and May's (1996) work suggests, the understanding of the encounter that in depth qualitative analysis would provide will also contribute to developing methods for assessing its success. In order to

disseminate good or successful genetic practice, it is necessary firstly to identify the component parts of this process. Counselling is not a unitary profession, and this raises issues of the training, orientation and qualifications of the counsellors involved in the studies presented here. However, this issue is largely sidestepped in reporting individuals attitudes to or satisfaction with counselling services. The role of the counsellor, their relationship to the clinical team, and the perception of the counsellor by the client may also be important. However, as far as CA analyses are concerned, and in terms of client uptake, what the counsellor thinks he or she is doing may be less significant than what the patient thinks is occurring. Advice, for example, only gains its advisory character if it is treated as such by the recipient. As Ahrens et al (1992) note, discussing their experiences of having a philosopher attached to their genetics unit, an observer in such a field can show how a team of geneticists is operating in practice, whatever they think they might be doing.

BIBLIOGRAPHY

Adam, S. Wiggins, S. Whyte, P. et al 1993. `Five Year Study of Prenatal Testing for Huntington's Disease: Demand, Attitudes, and Psychological Assessment'. *Journal of Medical Genetics*, vol:**30**, p549-556.

Ahrens, M. Otte, K. LeRoy, B. 1992. `The Philosopher and the Genetic Counsellor; Dialogue'. *Journal of Genetic Counselling*, vol:**1**, no.4, p310.

Atkinson, B, J. 1992. Conversation Analysis of Therapeutic Discouse: The Pursuit of a Therapeutic Agenda, Gale, J, E. *Journal of Marital and Family Therapy*, vol:**18**, no.4, p422-3.

Audrain, J., Rimer, B., Cella, D. et al. 1998. 'Genetic Counseling and Testing for Breast-Ovarian Cancer Susceptibility: What do Women Want?', *Journal of Clinical Oncology*, 16; 1: 133-138

Bleiker, E., Aaronson, N., Menko, F. et al. 1997. 'Genetic Counseling for Hereditary Cancer: A Pilot Study of Experiences of family Members. *Patient Education and Counseling*, **32**: 106-7

Bloor, M. 1976. Professional autonomy and client exclusion, in: M. Wadsworth and D. Robinson (eds) *Studies in Everyday Medical Life*, London: Martin Robertson

Borgaonkar, D, S. 1991. `Genetic Counselling to a Traditional Society' (Letter to the editor in response to Carmi, R.). *The Lancet*, vol:**337**, (March 23) p739.

Burke, B.M. 1992. 'Genetic counselor attitudes towards fetal sex identification and selective abortion', *Soc. Sci.Med.*, 34; 11: 1263-69

Carmi, R. 1991. `Genetic Counselling to a Traditional Society'. *Lancet*, vol:**337**, (2nd Feb) p306.

Chadwick, R, F. 1993. `What Counts as Success in Genetic Counselling?'. *Journal of Medical Ethics*, vol:**19**, p43-6.

Chapple, A. and May, C. 1996. 'Genetic knowledge and family relationships: two case studies',*Health and Social Care in the Community*, 4; 3: 166-171

Chapple, A., Campion, P., and May, C. (1997) 'Clinical terminology: anxiety and confusion amongst families undergoing genetic counseling', *Patient Education and Counseling*, 32: 81-91

Chase, G.A., Faden, R.R., Holtzman, N.A. et al. 1986. 'Assessment of Risk by Pregnant Women: Implications for Genetic Counseling and Education', *Social Biology*, 33; 1-2: 57-64

Clarke, A. 1991a. `Is non-directive counselling possible?'. *Lancet,* vol:**338**, (19th Oct) p998-1001.

Clarke, A. 1991b. `Non-directive Genetic Counselling'. *The Lancet*, vol:**338**, (Dec 14) p1524.

Clarke, A. 1994. *Genetic Counselling: Practice Principle.* Routledge, London.

Clarke, A., Parsons, E., Williams, A. 1996. 'Outcomes and Process in Genetic Counselling', *Clinical Genetics*, **50**; p462-469

Cull, A., Miller, H., Porterfield, T. et al. 1998. 'The use of videotaped information in cancer genetic counselling: a randomized evaluation study', *British Journal of Cancer*, **77**: p830-837

Czeizel, A., Metneki, J., Osztovics, M. 1981. 'Evaluation of information-guidance genetic counselling', *Journal of Medical Genetics*, **18**: p91-98

D'Amico, R., Jacopini, G., Vivona, G., Frontali, M. 1992. 'Reproductive choices in couples at risk for genetic disease: A quantitative and qualitative analysis', *Birth Defects: Original Article Series*, vol: **28**, no.1, p167-184. March of Dimes Birth Defects Foundation.

Decruyenaere, M. Evers-Kiebooms, G. Van den Berghe, H. 1992a. `Community Knowledge About Human Genetics'. *Birth Defects: Original Article Series*, vol:**28**, no.1, p167-184. March of Dimes Birth Defects Foundation.

Decruyenaere, M. Evers-Kiebooms, G. Van den Berghe, H. 1993. `Perception of Predictive Testing for Huntington's Disease by Young Women: Preferring Uncertainty to Certainty?'. *Journal of Medical Genetics,* vol:**30,** p557-561.

Decruyenaere, M. Evers-Kiebooms, G. Denayer, L. Van den Berghe, H. 1992b. `Cystic Fibrosis: Community knowledge and attitudes towards carrier screening and prenatal diagnosis'. *Clinical Genetics,* vol:**41,** p189-96.

Demyttenaere, K. Evers-Kiebooms, G. Decruyenaere, M. 1992. `Pitfalls in Counselling for Predictive Testing in Huntington Disease'. *Birth Defects: Original Article Series*, vol:**28**, no.1, p105-111. March of Dimes Birth Defects Foundation.

Denayer, L. De Boeck, K. Evers-Kiebooms, G. Van den Berghe, H. 1992a. `The Transfer of Information About Genetic Transmission to brothers and Sisters of Parents with a C.F.-Child'. *Birth Defects: Original Article Series*, vol:**28**, no.1, p149-158. March of Dimes Birth Defects Foundation.

Denayer, L. Evers-Kiebooms, G. De Boek, K. Van den Berghe, H. 1992b. `Reproductive Decision Making of Aunts and Uncles of a Child with Cystic Fibrosis: Genetic risk perception and attitudes towards carrier identification and prenatal diagnosis'. *American Journal of Medical Genetics,* vol:**44** p104-11.

Dingwall, R. Greatbatch, D. 1992. `Telling Bad News'. Unpublished paper presented to 3rd European Meeting on Psychosocial Aspects of Genetics, 21-23 September 1992, Nottingham.

Drew, P. and Heritage, J (eds) 1992 *Talk at Work: Interaction in Institutional Settings.* Cambridge: Cambridge University Press.

D'Ydewalle, G. And Evers-Kiebooms, G. 1987. 'Experiments on Genetic Risk Perception and Decision Making: Explorative Studies', *Birth Defects: Original Article Series*, **23**; 2: 209-225

Eggers, S. Passos-Bueno, M, R. Zatz, M. 1993. `Facioscapulohumeral Muscular Dystrophy: Aspects of Genetic Counselling, Acceptance of Preclinical Diagnosis, and Fitness'. *Journal of Medical Genetics,* vol:**30**, p589-92.

Ekwo, E.E., Seals, B.F., Kim, O.J. et al. 1985. 'Factors Influencing Maternal Estimates of Genetic Risk', *American Journal of Medical Genetics*, **20**; 491-504

Evans, D, G, R. Burnell, L, D. Hopwood, P. Howell, A. 1993. `Perception of Risk in Women with a Family History of Breast Cancer'. *British Journal of Cancer*, vol:**67**, p612-14.

Evers-Kiebooms,G., Swerts,A.,Van den Berghe, H. 1988. `Psychological Aspects of Amniocentesis: Anxiety Feelings in Three Different risk Groups'. *Clinical Genetics,* vol:**35,** p763-79.

Evers-Kiebooms, G., Swerts, A. Cassiman, J, J. Van den Berghe, H. 1989. `The Motivation of at Risk Individuals and Their Partners in Deciding For or Against Predictive Testing for Huntington's Disease'. *Clinical Genetics,* vol:**35,** p29-40.

Evers-Kiebooms, G. Denayer, L. Decruyenaere, M. Van den Berghe, H. 1993. `Community Attitudes Towards Prenatal Testing for Congenial Handicap'. *Journal of Reproductive and Infant Psychology,* vol:**11,** p21-30.

Fisher, L., Rowley, P.T., Lipkin, M. 1981. 'Genetic counseling for Beta-Thalassemia Trait Following Health Screening in a Health Maintenance Organisation: Comparison of Programmed and Conventional Counseling',*American Journal of Human Genetics*, **33**, p987-994.

Frets, P, G. Diuvenvoorden, H, J. Verhage, F. Peters-Romeyn, B, M. Niermeijer, M, F. 1991. `Analysis of Problems in Making the Reproductive Decision After Genetic Counselling'. *Journal of Medical Genetics,* vol:**28,** no.3, (March) p194-200.

Gale, J. Newfield, N. 1992. A Conversation Analysis of a Solution Focused Marital Therapy Session, *Journal of Marital and Family Therapy*, vol:**18** no.2, p153-65.

Green, M, R. Weaver, L, T. Heeley, A, F.et al 1993. `Cystic Fibrosis Identified by Neonatal Screening:Incidence, Genotype, and Early Natural History'. *Archives of Disease in Childhood*, vol:**68**, p164-7.

Hallowell, N, Murton, R., Statham, H. et al. 1997. 'Women's need for information before attending genetic counselling for familial breast or ovarian cancer: a questionnaire, interview and observational study', *BMJ*, 314: 281-283

Haug, M. Lavin, B. 1978. `Method of Payment for Medical Care and Public Attitudes Toward Physician Authority'. *Journal of Health and Social Behaviour*, vol:19, (Sept) p279-91.

Haug, M. Lavin, B. 1980. `Comment on a Comment: Soft Words for Dr. Strong'. *Journal of Health and Social Behaviour*, vol:6 p198-9.

Heath, C. (1992) The delivery and reception of diagnosis in the general practice consultation. In Drew, P. and Heritage, J. (eds) *Talk at Work* (ibid)

Hildes, E. Jacobs, H, K. Cameron, A. et al. 1993. `Impact of Genetic Counselling After Neonatal Screening for Duchenne Muscular Dystrophy'. *Journal of Medical Genetics*, vol:**30**, p670-4.

Holtzman, N.A. 1993. 'Primary Care Physicians as Providers of Frontline Genetic Services', *Fetal Diagnosis and Therapy*, 1993; 8 (suppl 1): 213-219

Houlston, R, S. Lemoine, L. McCarter, E. et al. 1992. `Screening and Genetic Counselling for Relatives of Patients with Breast Cancer in a Family Cancer Clinic'. *Journal of Medical Genetics,* vol:**29,** no.10, p691-4.

Houlston, R, S. Murday, V. Harocopos, C et al. 1990. `Screening and Genetic Counselling for Relatives of Patients with Colorectal Cancer in a Family Cancer Clinic'. *British Medical Journal*, vol:**301**, p166-8.

Huys, J. Evers-Kiebooms, G. d'Ydewalle, G. 1990. `Framing Biases in Genetic risk Perception'. *Cognitive Biases. Elsevier Science Publishers, B.V.* (North Holland).

Jarmas, A.L. 1983. 'Confessions of a Genetic Counselor', *Prenatal Diagnosis*, 3; 3: 270

Kessler, S. 1981.'Psychological aspects of genetic counseling: Analysis of a transcript', *American Journal of Medical Genetics*, 8; 2: 137-153

Kessler, S. 1992. 'Process Issues in Genetic Counselling', *Birth Defects: Original Article Series*, Vol **28**, No 1, p1-10.

Kessler, S. 1997 'Genetic Counseling is Directive? Look again'. Letter to the Editor, *American Journal of Human Genetics*, 61: 466-67

Kessler, S. and Jacopini, A.G. 1982 'Psychological aspects of genetic counseling II: Quantitative analysis of a genetic counseling transcript', *American Journal of Medical Genetics*, 12; 4: 421-35

Kessler, S., Kessler, H. and Ward, P. 1984. 'Psychological aspects of genetic counseling III. Management of shame and guilt', *American Journal of Medical Genetics,* 17; 3: 673-697

Kessler, S. and Levene, E.K. 1987. 'Psychological aspects of genetic counselling IV: The subjective assessment of probability ', *American Journal of Medical Genetics,* **28:** 361-70

Lindhout, D. Frets, P, G. Niermeijer, M, F. 1991. `Approaches to Genetic Counselling'. *Annals to the New York Academy of Sciences*, vol:**630**, p223-9.

Livingstone, J. Axton, R, A. Gilfillan, A. et al. 1994. `Antenatal Screening for Cystic Fibrosis: A trial of the couple model'. *British Medical Journal,* vol:**308,** (4th June) p159-67.

Lloyd, S., Watson, M., Waites, B. et al 1996. 'Familial breast cancer: a controlled study of risk perception, psychological morbidity and health beliefs in women attending for genetic counselling', *British Journal of Cancer*, 74: 482-87

Loader, S., Sutera, C.J., Walden, M. et al. 1991. 'Prenatal Screening Hemoglobinopathies II. Evaluation of Counseling', *American Journal of Human Genetics*, **48**: p447-451

Lynch, H, T. Watson, P. 1992. `Genetic Counselling and Hereditary Breast/Ovarian Cancer'. *Lancet,* vol:**339,** (9th May) p1181.

Macintyre S. & Oldman, P. (1977) 'Coping With Migrane in Medical Encounters' in Davis, A. & Horobin, G. (eds) *The experience of Illness and Treatment.* London, Croom Helm Ltd.

Mallett, J. 1990. `Communication Between Nurses and Post-anaesthetic Patients' *Intensive Care Nursing,* vol:**6,** no.1, (March) p45-53.

Markova, I., Forbes, C.D. and Inwood, M. 1984. 'The Consumers' Views of Genetic Counseling of Hemophilia',*Americaan Journal of Medical Genetics,* 117: 741-52

Mendez, A. and Shymansky, J.A. 1984. 'Verbal behaviours and patterns exhibited by phsicians during genetic counselling sessions', *Patient Education and Counseling*, 6; 4: 165-168

Mennie, M.E., Compton, M. E., Gilfillan, A., et al. `Prenatal Screening for Cystic Fibrosis: Psychological Effects on Carriers and Their Partners'. *Journal of Medical Genetics*, vol:**30**, p543-8.

Michie, S., Bron, F., Bobrow, M. and Marteau, T. 1997. 'Nondirectiveness in Genetic Counselling: An Empirical Study.' *American Journal of Human Genetics*, 60: 40-47

Michie, S., French, D,, Allanson, A. et al . 1997. 'Information recall in genetic counselling: a pilot study of its assessment', *Patient Education and Counseling*, 32: p93-100

Michie, S., Marteau, T.M. and Bobrow, M. 1997. 'Genetic counselling: the psychological impact of meeting patients' expectations', *Journal of Medical Genetics*, 34: 237-241

Michie, S., McDonald, V., Marteau, T.M. 1997. 'Genetic counselling: information given, recall and satisfaction', *Patient Education and Counseling*, **32**, p101-106.

Mitchell, J. Scriver, C, R. Clow, C, L. Kaplan, F. 1993. `What Young People Think and Do When the Option for Cystic Fibrosis Carrier Testing is Available'. *Journal of Medical Genetics*, vol:**30**, P538-42.

Morrison, P.J. and Nevin, N.C. (1991) Non-Directive Genetic Counselling (letter), *Lancet*, 338: p1267

Naveed, M. Phadke, S, R. Sharma, A. Agarwal, S, S. 1992. `Sociocultural Problems in Genetic Counselling'. *Journal of Medical Genetics*, vol:**29**, p140.

Nuffield Council on Bioethics (Dec 1993). *Genetic Screening: Ethical Issues, Conclusions*. Nuffield Council, London.

Parsons, E. Atkinson, P. 1992. `Lay Constructions of Genetic Risk'. *Sociology of Health Illness,* vol:**14,** no.4, p437-55.

Parsons, E, P. Clarke, A, J. 1993. `Genetic Risk: Women's Understanding of Carrier Risks in Duchenne Muscular Dystrophy'. *Journal of Medical Genetics*, vol:**30**, p562-6.

Pembrey, M. 1991. `Non-directive Genetic Counselling: Letter To The Editor'. *Lancet,* vol:**338**, p1266-7.

Raeburn, S. 1994. `Screening for Carriers of Cystic Fibrosis: Time to decide'. *British Medical Journal,* vol:**308,** (4th June) p1451-52.

Rowley, P.T., Lipkin, M., Fisher, L. 1982. 'Screening and Genetic Counselling for Beta-Thalassaemia Trait in a Population Unselected for Interest: Comparison of Three Counseling Methods', *American Journal of Human Genetics,* **36**: p677-689

Scholz, C. 1992. `On the Interactive Accomplishment of Decision in Genetic Counselling Before Prenatal Diagnosis'. *Birth Defects: Original Article Series*, vol:**28**, no.1, p47-55. March of Dimes Birth Defects Foundation.

Seidenfield, M.J., Antley, R.M. 1981. 'Genetic Counseling: A comparison of counselee's genetic knowledge before and after', *American Journal of Medical Genetics*, **10**: p107-112

Sefi, S. 1988. `Health Visitors Talking to Mothers'. *Health Visitor*, vol:**61**, (Jan) p7-10.

Seller, M, J. 1982. `Ethical Aspects of Genetic Counselling' *Journal of Medical Ethics,* vol:**8,** p185-8.

Seller, M, J. 1982. `Genetic Counselling and Reproductive Patterns'. *Midwife, Health Visitor Community Nurse*, vol:**18**, p135 passim.

Sharrock, W. (1979) 'Portraying the Professional Relationship' in Anderson, C. (ed) *Health Education in Practice*, London, Croom Helm.

Shiloh, S., Avdor, O. and Goodman, R.M. 1990. 'Satisfaction with Genetic Counseling: Dimensions and Measurement', *American Journal of Medical Genetics*, 37: 522-529

Somer, M., Mustonen, H. Norio, R. 1988. 'Evaluation of Genetic Counselling: Recall of Information, Post-Counselling Reproduction and Attitude of the Counsellees', *Clinical Genetics,* **34**: p352-365

Sorensen, J.R., Kavanagh, C.M., Mucatel, M. 1981. 'Client Learning of Risk and Diagnosis in Genetic Counseling', *Birth Defects: Original Article Series*, Vol **17**, No 11, p2115-228.
Stimson, G. And Webb, B. 1975. *Going to see the Doctor*, London: Collier MacMillan

Strong, P, M. 1980. `On Method of Payment for Medical Care and Public Attitudes Toward Physician Authority'. *Journal of Health and Social Behaviour*, 6 , p195-7.

Strong, P.M. and Davis, A.G. (1976) The management of therapeutic encounters with young children in *The Sociology of Everyday Medical Life* (eds: M. Wadsworth and D. Robinson), London: Martin Robertson,

Strong, P.M. and Davis, A.G. (1978) Who's who in paediatric encounters: Morality, expertise and Action in medical setings. In A.G. Davis (ed) Relationships between Doctors and Patients, London: Saxon House.

Super, M. 1991. `Non-directive Genetic Counselling: Letter to the editor'. *Lancet,* vol:**338**, p1266.

Suslak, C. Price, D, M. Desposito, F. 1985. `Transmitting Balanced Translocation Carrier Information Within Families: A Follow-Up Study'. *American Journal of Medical Genetics,* vol:**20**, no.2, p227-232.

Swerts, A. 1987. `Impact of Genetic Counselling and Prenatal Diagnosis for Down Syndrome and Neural Tube Defects'. *Birth Defects: Original Article Series*, vol:**23**, no.2, p61-83. March of Dimes Birth Defects Foundation.

ten Have, P. (1991) Talk and institution: a reconsideration of the asymmetry of doctor-patient interaction. In Boden, D. And Zimmerman, D. (eds) *Talk and Social Structure: Studies in Ethnomethodology and Conversation Analysis*. Cambridge: Polity Press.

Tibben, A. Vegter-V.D. Vlis, M. Niermeijer, et al. `Testing for Huntington's Disease with Support for all Parties'. *The Lanct*, (Mar 3) vol:**335**, no:8688, p553.

Tibben, A. Niermeijer, M.F., Roos, R.A. et al. 1992. `Understanding the Low Uptake of Presymptomatic DNA Testing for Huntington's Disease'. *The Lancet*, vol:**340**, (Dec 5) p1416.

Tyler, A. and Harper, P. 1983. 'Attitudes of subjects at risk and their relatives towards genetic counselling in Huntington's chorea', *Journal of Medical Genetics*, 20: 179-188

Varekamp, I. Suurmeijer, P, B, M. Rosendaal, F, R. Brocker-Vriends, A, H, J, R. 1993. `The Use of Preventive Health Care Services Carrier Testing for the Genetic Disorder Haemophilia'. *Social Science and Medicine*, vol:**37**, no:**5**, p639-48.

Van Zuuren, F.J. (1997) 'The standard of neutrality during genetic counselling: An empirical investigation', *Patient Education and Counseling*, 32: 69-79

Van Zuuren, F.J., Van Schie, E.C.M. and Van Baaren, N.K. (1997) 'Uncertainty in the information provided during genetic counseling', *Patient Education and Counseling*, 32: 129-39

Watson, E, K. Marchant, J. Bush, A. Williamson, B. 1992. `Attitudes Towards Prenatal Diagnosis and Carrier Screening for Cystic Fibrosis Among the Parents of Patients in a Paediatric Cystic Fibrosis Clinic'. *Journal of Medical Genetics*, vol:**29**, p490-1.

Watson, E, K. Mayall, E, S. Lamb, et al. 1992. `Psychological and Social Consequences of Community Carrier Screening Programme for Cystic Fibrosis'. *The Lancet*, vol:**340**, (July 25) p217-20.

Wertz, D, C., Sorenson, J, R. 1986. `Client Reactions to Genetic Counselling: Self-Reports of Influence'. *Clinical Genetics*, vol:**30**, no.6, p494-502.

Wertz, D. and Fletcher, J.C. 1988. 'Attitudes of Genetic Counselors: A Multinational Survey', *American Journal of Human Genetics*, 42: 592-600

Wertz, D.C. and Fletcher, J.C. 1998. 'Ethical and social issues in prenatal sex selection: A survey of geneticists in 37 nations', *Soc.Sci.Med*, 46; 2: 255-273

Wertz, D, C. 1992. `Ethical and Legal Implications of the New Genetics: Issues for Discussion'. *Soc. Sci. Med.* vol:**35,** no.4, p495-505.

Wertz, D.C. 1993. 'Providers' Gender and Moral Reasoning', *Fetal Diagnostic Therapy*, 8(suppl 1): 81-89

Wertz, D.C, Sorenson, J.R. and Heeren, T.C. 1986. 'Clients' Interpretations of Risks Provided in Genetic Counseling', *American Journal of Human Genetics*, 39: 253-264

Whitten, C.F., Thomas, J.F. and Nishiura, E.N. 1981, 'Sickle Cell Trait Counseling-Evaluation of Counselors and Counselees, *American Journal of Human Genetics*, 33: 802-16

Young, R.S., Jorgenson, R.J., Shaapiro, S.D. 1986. 'Efficacy of and Patient preference for Three Counseling Formats', *Journal of Cranio Facial Genetics and Developmental Biology*, **6**: p3-14

Zare, N., Sorenson, J.R., Heeren, T. 1984. 'Sex of Provider as a Variable in Effective Genetic Counseling', *Social Science and Medicine*, Vol **19**, No 7, p671-675

Other papers published by the Trent Institute for Health Services Research are listed below:-

Guidance Notes for Purchasers

96/01	Working Group on Acute Purchasing: The Use of DNase in Cystic Fibrosis (1996) by JN Payne, S Dixon, NJ Cooper and CJ McCabe.	£6.00
96/02	Working Group on Acute Purchasing: Tertiary Cardiology (1996) by J Tomlinson, J Sutton and CJ McCabe.	£6.00
96/03	Working Group on Acute Purchasing: The Use of Cochlear Implantation (1996) by Q Summerfield and J Tomlinson.	£6.00
96/04	Working Group on Acute Purchasing: Statin Therapy / HMG Co-A Reductase Inhibitor Treatment in the Prevention of Coronary Heart Disease (1996) by MD Pickin, JN Payne, IU Haq, CJ McCabe, SE Ward, PR Jackson and WW Yeo.	£6.00
97/01	Working Group on Acute Purchasing: The Clinical and Cost-effectiveness of Computed Tomography in the Management of Transient Ischaemic Attack and Stroke (1997) by A Ferguson and CJ McCabe.	£10.00
97/02	Working Group on Acute Purchasing: Prostacyclin in the Treatment of Primary Pulmonary Hypertension (1997) by TW Higenbottam, SE Ward, A Brennan, CJ McCabe, RG Richards and MD Stevenson.	£10.00
97/03	Working Group on Acute Purchasing: The Use of Riluzole in the Treatment of Amyotrophic Lateral Sclerosis (Motor Neurone Disease) (1997) by J Chilcott, P Golightly, D Jefferson, CJ McCabe and S Walters.	£10.00
97/04	Working Group on Acute Purchasing: Recombinant Factor VIII Versus Plasma Derived Factor VIII in the Management of Haemophilia A: An Examination of the Costs and Consequences (1997) by C Green and RL Akehurst.	£10.00
97/05	Working Group on Acute Purchasing: The Use of Cisplatin and Paclitaxel as a First Line Treatment in Ovarian Cancer (1997) by SM Beard, R Coleman, J Radford and J Tidy.	£10.00
97/06	Working Group on Acute Purchasing: The Use of Alpha Interferon in the Management of Chronic Myeloid Leukaemia (1997) by RG Richards and CJ McCabe.	£10.00
97/07	Working Group on Acute Purchasing: Spinal Cord Stimulation in the Management of Chronic Pain (1997) by J Tomlinson, CJ McCabe and B Collett.	£10.00
97/08	Working Group on Acute Purchasing: The Use of Growth Hormone in Adults (1997) by JN Payne and RG Richards.	£5.00
97/09	Working Group on Acute Purchasing: A Review of the Use of Donepezil in the Treatment of Alzheimer's Disease (1997) by FA Pitt, J Chilcott, P Golightly, J Sykes, M Whittingham.	£10.00

97/10	Working Group on Acute Purchasing: The Use of Bone Anchored Hearing Aids (1997) by NJ Cooper, J Tomlinson and J Sutton.	£10.00
98/01	Working Group on Acute Purchasing: A Review of the Use of Current Atypical Antipsychotics in the Treatment of Schizophrenia (1998) by S Beard, J Brewin, C Packham, P Rowlands, P Golightly.	£10.00
98/02	Working Group on Acute Purchasing: Internal Fixation of Tibial Shaft and Distal Radius Fractures in Adults (1998) by N Calvert, P Triffit, S Johnstone, RG Richards.	£10.00
98/04	Working Group on Acute Purchasing: The Effectiveness of High Dose Chemotherapy and Autologous Stem Cell Transplantation in the Treatment of Hodgkin's Disease and Non-Hodgkin's Lymphoma (1998) by S Beard, P Lorigan, A Simms, F Sampson.	£10.00
98/05	Working Group on Acute Purchasing: Angiotensin-Converting Enzyme (ACE) Inhibitors in Heart Failure: Reducing Mortality and Costs to the NHS (1998) by N Calvert, J Cornell, C Singleton.	£10.00
98/06	Working Group on Acute Purchasing The Use Of Ultrasound (Viability) Scans In Early Pregnancy Bleeding (1998) by N Calvert, C Singleton, P Tromans.	£10.00
98/08	Working Group on Acute Purchasing: The Effectiveness of High Dose Chemotherapy with Autologous Stem Cell / Bone Marrow Transplantation in the Treatment of Multiple Myeloma (1998) by S Beard, F Sampson, E Vandenberghe and F Scott.	£10.00
98/10	Working Group on Acute Purchasing: Supplementary Document: The Use of Paclitaxel in the First Line Treatment of Ovarian Cancer (1998) by S Beard, R Coleman, J Radford and J Tidy.	£10.00
98/11	Working Group on Acute Purchasing: The Use of Fluoridated School Milk in the Prevention of Dental Caries (1998) by N Calvert and N Thomas.	£10.00
99/01	Working Group on Acute Purchasing: The Role of Antileukotrienes in the Treatment of Chronic Asthma (1999) by M Stevenson, R Richards, S Beard.	£15.00
99/02	Working Group on Acute Purchasing: Hepatic Resection as a Treatment for Liver Metastases in Colorectal Cancer (1999) by S Beard, M Holmes, A Majeed, C Price.	£15.00
99/03	Working Group on Acute Purchasing: A Review of the Use of Propentofylline in the Treatment of Dementia (1999) by J Chilcott, K Perrett, P Golightly, J Sykes and M Whittingham.	£15.00
99/04	Working Group on Acute Purchasing: The Use of Routine Antenatal Anti-D Prophylaxis for Rhesus Negative Women (1999) by M Allaby, K Forman, S Touch and J Chilcott.	£15.00
99/05	Working Group on Acute Purchasing: Magnetic Resonance Imaging (MRI) in the Management of Knee Disorders (1999) by SM Beard, I Perez , S Touch and D Bickerstaff.	£15.00

99/06	Working Group on Acute Purchasing: The Effectiveness of Surgery in the Management of Epilepsy (1999) by J Chilcott, S Howell, A Kemeny, C Rittey and Richards C.	£15.00
99/07	Working Group on Acute Purchasing: Tacrolimus and Mycophenolate Mofetil as Maintenance Immunosuppressants Following Renal Transplantation (1999) by J Chilcott, M Corcoran, KM Rigg, RP Burden.	£15.00
99/08	Working Group on Acute Purchasing: The Use of Endovascular Stents for Abdominal Aortic Aneurysm (1999) by NW Calvert, M Lloyd Jones, S Thomas, RG Richards, JN Payne.	£15.00
00/01	Working Group on Acute Purchasing: The Effectiveness of Intrathecal Baclofen in the Management of Patients with Severe Spasticity (2000) by FC Sampson, SH Touch, A Hayward, G Evans, R Morton, D Playford, M Vloeburghs, A Collett, P Critchley.	£15.00

Discussion Papers

No. 1.	Patients with Minor Injuries: A Literature Review of Options for their Treatment Outside Major Accident and Emergency Departments or Occupational Health Settings (1994) by S Read.	£7.00
96/01	Working Group on Acute Purchasing: The Role of Beta Interferon in the Treatment of Multiple Sclerosis (1996) by RG Richards, CJ McCabe, NJ Cooper, SF Paisley, A Brennan and RL Akehurst.	£7.50
96/02	The Mid-level Practitioner: A Review of the Literature on Nurse Practitioner and Physician Assistant Programmes (1996) by P Watson, N Hendey, R Dingwall, E Spencer and P Wilson.	£10.00
96/03	Evaluation of two Pharmaceutical Care Programmes for People with Mental Health Problems Living in the Community (1996) by A Aldridge, R Dingwall and P Watson.	£10.00
97/01	Working Group on Primary and Community Care Purchasing : Report of the Sub-Group on the promotion of Quality in Primary Care - Effective Purchasing of Primary and Community Health Care: Promotion of Quality in the Provision of Primary Care (1997) by S Jennings and M Pringle.	£10.00
97/02	Working Group on Primary and Community Care Purchasing : Report of the Sub-Group on Information Needs for Health Needs Assessment and Resource Allocation (1997) by T Baxter, A Howe, C Kenny, D Meechan, M Pringle, P Redgrave, J Robinson and A Sims.	£10.00
98/01	Working Group on Primary and Community Care Purchasing : Hospital at Home - Lessons from Trent (1998) by I Perez, A Wilson, A Sims and R Harper.	£10.00

Copies of these documents are available from:-

Information Resources
Trent Institute for Health Services Research
Regent Court
30 Regent Street
SHEFFIELD S1 4DA
Tel 0114 222 0703
Fax 0114 272 4095
E-mail scharrlib@sheffield.ac.uk