PRACTITIONERS AND PARADIGMS OF GENETIC COUNSELLING

"SOMETIMES WE STIR UP A HORNET'S NEST"

PRACTITIONERS AND PARADIGMS OF GENETIC COUNSELLING

By

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A Thesis

Submitted to the School of Graduate Studies

in Partial Fulfillment of the Requirements

for the Degree

Master of Arts

McMaster University

January 1987

MASTER OF ARTS (1987) McMASTER UNIVERSITY
(Anthropology) Hamilton, Ontario

TITLE: "Sometimes we stir up a hornet's nest":
Practitioners and Paradigms of Genetic Counselling

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SUPERVISOR: Dr. David Counts
NUMBER OF PAGES: viii, 172

ABSTRACT

The central investigative concern of this thesis is the culture of genetic counselling in Canada--a specialized health service providing information and assistance to families at risk from genetic disorders. In particular, this study describes the variability that exists in the way in which genetic counsellors view their profession and explores the factors that contribute to this variability. The genetic counsellors' training, division of roles, view of their responsibility toward clients, and the historical and contemporary context in which they practise contribute to understanding the "individuals' versions of the culture of [genetic counselling]" (Hahn 1985:53). Three means of inquiry are utilized: extensive library research, responses from a Canada-wide questionnaire sent to genetic counsellors, and ethnographic interviews.

The history of genetic counselling has been described as a series of changes from research to medicine to psychosocial concerns. Current definitions of adequate genetic care, however, stress attention to both the medical genetic and psychosocial aspects of genetic disorders suggesting that both a medical and a psychosocial paradigm now exist in genetic counselling. I argue that the association between these two paradigms is unclear, thus producing tension and ambiguity for practitioners. Genetic counsellors divide themselves into three groups--MD/MD-PhDs who are the central caregivers, PhD geneticists who are primarily involved in research, and nonMDs/nonPhDs such as nurses, social workers and master's level genetic associates who are often doing clinic-oriented clerical jobs. Nevertheless, indicative of the uneasy relationship between the paradigms of genetic counselling, considerable debate exists in the field about who should be providing genetic counselling. A further example of this paradigmatic tension concerns the implementation of genetic counselling goals, especially for physician geneticists, the central caregivers. In particular, genetic counsellors face the problematic task of "doing something" for their clients, making them aware of their options, without influencing the client's decision-making. The contemporary context of genetic counselling is predominantly a medical one; although psychosocial concerns are acknowledged as important for adequate genetic care, the means for dealing with them are not clear. I suggest that the response to the underlying tensions in genetic counselling is ultimately an individually-constructed response, based on the individual practitioner's experience, interpretation of the facts, and notion of the boundaries of their responsibility in medicine.

ACKNOWLEDGEMENTS

I wish to thank my committee members, Dr. David Counts (Anthropology), Dr. Emoke Szathmary (Anthropology) and Dr. Ron Davidson (Pediatrics) for their ready guidance, helpful comments, and steady optimism. A special thanks to Dr. Margaret Rodman for her encouragement, especially during the proposal writing and early fieldwork. To the genetic counsellors who participated in this study I am thankful for their frankness and patience in showing me their culture.

I am grateful to my friends, Naomi Adelson and Joel Minion, who shared the trauma and hilarity of graduate school and Hamilton with me and to Bill Simeone for reminding me that everything would work out. As well, I am indebted to Fred Hemus for his wizardry with XyWrite. To my son, Anand, thank you for waiting until I handed in a first draft. And finally, I wish to thank Marjorie Mitchell and Donald Mitchell for providing me with the necessary genetic material.

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INTRODUCTION

This thesis has as its topic of investigation the practitioners of 'genetic counselling'--a specialized health service providing information and assistance to families at risk from genetic disorders. Although there is a large body of research directed towards the patient or client in genetic counselling much less attention has been paid to the perspective of individual practitioners. The meaning and experience of genetic counselling as perceived by its practitioners is explored along three avenues of inquiry in this study:

1) by examining genetic counselling's current domain of knowledge and the roles and training of its practitioners;

2) by investigating the historical and contemporary sociocultural context in which genetic counselling takes place;

3) by exploring genetic counselling professionals' views of their role and responsibility and determining their concerns as practitioners attempting to assist their clients.

Inquiry into the content, context, and individualized view of genetic counselling is central to the anthropological approach which is followed in this thesis. Anthropological approaches to medical systems focus attention on the connections between the "domain of knowledge or belief, with concomitant values and rules for behaviour [that exists in the system, and the] " . . . realm of

action in which these beliefs, values and rules are carried out" (Hahn and Kleinman 1982:5). Using an anthropological approach to genetic counsellors allows me to understand

> the explicit and implicit conceptions and ideas which motivate behaviour and which allow physicians to evaluate their own and their colleagues' actions and to make sense of their medical experience (Gaines and Hahn 1985:8).

In the specific context of genetic counselling I examine the historical process by which a medical and a psychosocial paradigm have come to influence this health service. I argue that the existence of these two paradigms has meant genetic counsellors act in an arena characterized by an accumulation of competing orientations. I suggest that the response to these underlying tensions which characterize the behaviour and ideas of genetic counsellors is ultimately an individually-constructed response based on the individual practitioner's experience, interpretation of facts, and opinion in medicine.

The remainder of this introductory chapter reviews research on genetic counselling professionals and outlines the design and theoretical basis of this particular study. Chapter One discusses genetic counselling's historical context and introduces the medical paradigm and psychosocial paradigm which characterize the field. Chapter Two describes the contemporary context of genetic counselling in Canada in reference to these two paradigms. Chapter Three utilizes questionnaire responses and interview materials to present the ethnographic description of the participants in this study and to understand their roles as genetic counselling professionals. Chapter Four deals with the current priorities and concerns of genetic counselling professionals. The goal of this chapter is to explore the approaches of practitioners dealing with genetic knowledge as individuals. Chapter Five focuses the analysis on the par-

adigmatic tensions which underlie genetic counselling and the response of individuals to these tensions. A summarizing chapter completes the thesis.

Studies of Genetic Counsellors:

The majority of publications on genetic counselling focus on issues considered to influence the efficacy of genetic counselling: methods of conveying information to the client, retention of genetic information, the impact of genetic counselling on client's reproductive behaviour and on the process of reproductive decision-making in the face of genetic risk.[1] Most of this literature is written by medical geneticists, although there is a growing number of articles written by social workers involved in genetic counselling (see, for example, Black 1982 and Mealey 1984). At least three anthropologists (Meier n.d.; Rapp 1985; 1986; Ross 1985) have initiated studies of genetic counselling that incorporate client expectations of genetic counselling and perceptions of risk. Results from these anthropological studies have not yet been published.

There are only a handful of studies aimed directly at the professionals providing genetic counselling. All of these studies have appeared since the mid-1970s and coincide with a period of change in the character of genetic counselling in North America. For the past ten to 15 years the discipline has been responding to ethical and social concerns by attempting to combine biomedical (both clinical and research) interests with attention to the psychosocial or sociomedical needs of the client. Psychosocial issues include the religious beliefs, financial constraints, emotional response, perception of genetic disorders, and reproductive goals of the client (Sorenson and Culbert 1979: 90).

A. Surveys

The implications of this client-centered, bio-psychosocial mandate for genetic counselling have led practitioners to express concern about the nature of their role in assisting individuals confronted with genetic disease (Antley 1979; Sorenson and Culbert 1979). However, only two large scale surveys of genetic counselling professionals have been published. In the first study, Sorenson and Culbert (1977:132) analyzed questionnaire responses from 496 American "genetic counselors" surveyed in 1973 "to comment on the diversity of counseling styles and to develop suggestions for conducting evaluation studies with counselling orientation as one variable."

As the first investigators to focus on practitioners of genetic counselling, Sorenson and Culbert (1977;1979) provided valuable descriptive information about who those practitioners were, the organization and setting of counselling, and types of counselling strategies employed. In their U.S. sample, 72 per cent of genetic counselors were MDs (mostly pediatricians), 11 per cent were PhDs (predominantly geneticists), eight per cent MD-PhDs, two per cent nurses, and seven per cent other (Sorenson and Culbert 1977:135). When asked to indicate their current area of "primary professional interest" MDs, PhDs, and MD-PhDs were found to be significantly "more research oriented than clinically oriented" (Sorenson and Culbert 1977:138). Furthermore, genetic counselling itself is a primary interest in "only 11% of the MDs, 6% of the MD/PhDs, and 13% of the PhDs [who are practising genetic counsellors]" (1979:88).

Sorenson and Culbert (1979) also asked participants to rank the appropriateness of certain strategies, to indicate their goals for counselling, the factors they felt were most important in counsellee decision-making, and their professional obligation to raise and discuss a variety of issues with counsellees.

A more detailed report of the results of their study will be provided in Chapters One and Four. However, at this point it is worthwhile to note the following conclusions:

> 1) "There are significant differences in the manner in which counselors are oriented in counseling" (Sorenson and Culbert 1977:150-151);
>
> 2) 75 per cent of those counsellors emphasized provision of disease risk information over discussion of psycho-social issues; 19 per cent viewed both orientations as of equal importance; and only six per cent considered psychosocial issues to be of primary importance (Sorenson and Culbert 1979:92).

Individuals' explanations for these preferences are not dealt with in the study, although these authors suggest that an orientation toward psychosocial counselling does not lend itself to easy "equation solving" and "rational decisions"; rather it "adds complex and nonquantifiable elements" to the counsellors' task (Sorenson and Culbert 1979:100).

In the late 1970s, the March of Dimes Birth Defects Foundation funded a survey of genetic counselling in the USA "focusing on three issues: delineation of client needs, client education, and the impact of counseling on reproductive intentions" (Sorenson *et al.* 1981:10). Although the agenda of this second national survey was predominantly concerned with the client, the data analyzed from pre- and post-counselling session questionnaires received from 205 genetic counsellors raise several interesting points concerning the effectiveness of counselling.

On meeting client-defined needs in genetic counselling, the March of Dimes study "revealed that, in general, while counselors do a fair job of discussing the medical questions clients bring to counseling, they do a poor job of dis-

cussing sociomedical issues and problems" (Sorenson *et al.* 1981:140). The investigators recommend all genetic counsellors receive more training in counselling methods, involve counselling professionals such as social workers and genetic associates, and use resources in the community (social service agencies, parents' groups) "to help clients cope more effectively with their sociomedical needs" (Sorenson *et al.* 1981:141). A second conclusion made in this investigation is that although most genetic counsellors in the USA accept a definition of their role which includes both the medical genetic and sociomedical (or psychosocial) concerns, they have yet to "assume the authority and responsibility for providing those who seek their services with both medical genetic and counseling expertise" (Sorenson *et al.* 1981:144).

A major weakness with this study is that it lumps together into one category the MDs, PhDs, master's level genetic associates, nurses and social workers providing this health service. Aside from a brief look at the educational background (highest degree obtained, specialization area, training in human genetics and counselling), the number of years experience, and time spent in professional activities, other possible and probable differences among these professional groups are not addressed. Calling them all "genetic counsellors" assumes a homogeneity of views among these professionals that remains untested and does not reflect the diversity of occupational labels that professionals in Canada prefer to use for themselves. A second inadequacy of this investigation is that it fails to link its conclusions to the socio-historical context in which genetic counselling is provided. Understanding the complex historical and social dynamic which has produced the culture of genetic counselling makes the views of genetic counsellors more comprehensible.

Counsellors' responses to the March of Dimes study were also utilized to investigate sex of the genetic counsellor as a variable in effective counselling. An "analysis of how providers view their counseling goals, activities, and role in counseling revealed no significant differences between the sexes " (Zare *et al.* 1984:672). Sex differences among counsellors in perceived goal accomplishment, level of satisfaction in counselling, or other counsellor-defined concerns were not investigated.

B. Genetic Counselling Professionals in Context

Only two articles were found that attempted to place the values and opinions of genetic counselling professionals within a social and historical context. Kenen (1984:541) uses "a power approach" to look at the development and establishment of the Master's level genetic counsellor as an occupation alongside medical geneticists (PhDs) and medical doctors (MDs) in the United States. Kenen's study is significant in that she draws upon the localized context of genetic counselling, the "history, power and ideology of the professionals active in the discipline and the large social context" as interactive influential sources of role definition (Kenen 1984:541). Unfortunately, Kenen's analysis assumes a very homogenous quality to each of the professional groups described; the individual professional is submerged in the group.

The second article that attempts to situate counsellor attitudes in a sociohistorical context is written by Sorenson (1979). Drawing from the history of applied human genetics in the United States, definitions of genetic counselling, studies assessing the effectiveness of genetic counselling, and his own 1973 national "attitudinal study" of genetic counsellors (Sorenson and Culbert 1977), Sorenson comments on the diversity of genetic counsellors' perspectives "toward

their professional activity" (Sorenson 1979: 283). Three themes surrounding the practitioners' interpretations of 'genetic counselling' emerge from his review: counselling as preventive medicine for the individual, as public health based preventive medicine, and as patient education. Sorenson's (1979) article is significant because it manages to draw out these common patterns in orientation without losing a sense of the "wide variability" that characterizes genetic counselling.

C. Current Research

In addition to the published studies reviewed above, state level studies of genetic counselling are being conducted in Ohio (Ross 1985: personal communication) and Indiana (Meier 1984: personal communication; Steltz-Lenarsky *et al.* 1985). The process of prenatal diagnosis from the lab, to the counselling session, to the "networks of support and criticism" that surround reproductive decision making is being studied by Dr. Rayna Rapp at a New York City clinic (1985:10). Although these investigations do include the health care professionals' perspective in their analyses, the results have not yet been published.

Subsequent to the initiation of my own research, Canadian College of Medical Genetics members have taken part in a cross-cultural survey of ethical attitudes being conducted by Dr. John Fletcher (National Institutes of Health), Dr. Dorothy Wertz and Dr. James Sorenson (both from the School of Public Health, Boston University). This "International Survey of Medical Genetics" draws upon medical geneticists and ethicists in 19 countries.

The basis of the international study is a questionnaire containing 14 "cases" each describing a situation typical of genetic counselling. Respondents

are asked to choose a "course of action" from a list which follows each case and then to explain why they have chosen that particular course of action. As the instructions to the questionnaire make clear, Fletcher and his colleagues are interested in having medical geneticists "try to express the *ethical* reasoning behind . . . [their] actions" (Fletcher *et al.*, International Survey of Medical Genetics, 1985:5). The questionnaire contains a sample case, examples of ethical reasoning, and a relatively detailed statement of the kind of answer the researchers consider appropriate to explaining why a particular course of action was taken. Questionnaire participants are urged, "please DO NOT elaborate on WHAT you would do . . . [and] do not provide only technical justification for your decisions" (Fletcher *et al.*, International Survey of Medical Genetics, 1985:6, authors' emphasis).

Certainly, these instructions are helpful for guiding respondents to answer the question that has been asked. But suggesting examples of ethical reasoning and providing, as the International Survey of Medical Geneticists does, a list of "keywords that *may* appear in ethical reasoning" (Fletcher et al., International Survey of Medical Genetics, 1985:6, authors' emphasis)[2] may bias respondents to provide the answers that are desired, too. For example, providing the list of keywords assumes that these concepts are part of an individual's notion of "ethical reasoning". Results from this "International Survey" will be available in book form as *Ethics and Human Genetics: a Cross-Cultural Perspective* (to be published by Springer Verlag in 1987).

D. Counselling Models

A second group of publications about genetic counsellors are theoretically or clinically oriented descriptions and comparisons of counselling models. These publications are mentioned briefly here to complete the picture of research done previously, but are not used in the analysis. At least four counselling models are discussed in the literature--1) the traditional patient-physician model (directive counselling) (Silverberg and Godmilow 1979:283); 2) information-giving (Hsia 1979) or non-directive educational model (Silverberg and Godmilow 1979:283); 3)psychotherapeutic (Kessler 1979); and 4) Antley's (1979b) "facilitator of counsellee decision-making model." The role of genetic counsellors as moral advisors has also been discussed (Twiss 1979). The extent to which genetic counsellors agree upon or are trained in a particular model is not known. The various counselling paradigms may be intended as models for counsellor decision-making and problem-solving (Antley 1979b), but variation in interpretation of the models in day-to-day clinical practice does not appear to have been discussed in the literature. Clearly, much study is needed to reveal 1) the factors that influence counsellor orientation, 2) the extent of individual interpretation of counselling models, and 3) the implications of orientation choice on outcome.

In summary, previous published results of studies of genetic counselling professionals have provided a basic description of sociodemographic characteristics of American genetic counsellors (MDs, PhDs, master's level genetic counsellors and others) and an awareness that there is a diversity of interpretations in counselling goals and strategies held by these professionals. At least two authors (Kenen 1984; Sorenson 1979) have begun to examine this diversity in a sociohistorical context.

Several issues remain inadequately investigated, issues to which this study responds directly.

1) With the exception of the International Survey of Medical Geneticists (Fletcher *et al.* 1985) there has been no survey of genetic counsellors outside of the USA. This research will provide a comprehensive description of the sociodemographics, goals, orientations, and concerns of genetic counselling professionals in Canada. In addition, the context in which this health service has emerged in Canada and in which it is currently provided will be examined.

2) Previous investigations have tended to lump together all health care professionals providing genetic counselling into one category: "genetic counsellors". This study will investigate differences in activities, goals, and concerns among the groups--MDs, PhDs, MD-PhDs, and others (nurses, genetic associates, and social workers) that comprise "genetic counselling professionals".

3) Although the work of Kenen (1984) and Sorenson (1979) has begun to tackle the task of contextualizing our knowledge of genetic counsellors in a framework of historical circumstance, clinical settings, and social values, the attitudes of individual genetic counsellors have been surrendered to that framework.

This thesis addresses the role and responsibilities of the genetic counselling professional as an individual by investigating the meaning practitioners attach to their work. In particular, the perspective of the central genetic counselling professional, the physician geneticist, is investigated. For an anthropologist this emphasis is on the emic view of genetic counselling. In other

words, I examine the view held by the genetic counsellor, as opposed to the etic view, in this case, the observer's or the client's view of the process.

Research Design and Methods

The research plan utilizes three primary means of data collection--library research, questionnaire responses, and taped interviews.

First, extensive library research on the stated goals and methods of genetic counselling was undertaken. Recent textbooks, articles and other educational materials which present genetic counselling objectives and methods for counsellors-in-training, general practitioners, and for other primary health care workers were investigated. Publications designed to inform the general public about the nature of genetic counselling were also used. Government publications, articles which deal with legal, public policy, and ethical concerns about genetic counselling, and responses by counsellors to these issues were also investigated.

Second, questionnaires were sent to 141 genetic counselling professionals across Canada. The population was generated by obtaining the mailing lists for the Canadian College of Medical Genetics and for *CrossOver* a publication that is aimed at the nonMD/nonPhD genetic counsellor. Questionnaire responses were received from 45 CCMG members and 31 non-MD/non-PhD health care professionals.

The questionnaire asked participants to provide basic personal sociodemographic information (age, sex, religion, education *etc.*), to indicate their regular tasks and the number of hours spent doing them, and the average number of clients they see in a week. The questionnaire also asked participants

to rank goals and responsibilities in counselling, to comment on four counselling cases, and to respond to four comments made by critics of contemporary genetic counselling.

Statistical Tests:

Questionnaire responses were tabulated for descriptive purposes, and responses were broken down by professional group (MD, PhD, MD-PHD, genetic associates, nurses and other). Mann-Whitney paired comparison tests for significance between groups were carried out wherever possible. The small sample size of each group and the nature of the questions asked precluded using more powerful statistical analyses. Results were converted to one-tailed probabilities and considered significant at $p<0.05$ unless otherwise stated. Responses to cases and comments were treated as ethnographic text and were not quantified.

Third, ethnographic interviews were conducted with five physician geneticists and six nonMD/nonPhD genetic counselling professionals. The interviews in almost all cases were approximately one hour in length, and several individuals were interviewed on more than one occasion. Interview questions were open-ended and focused primarily on the character of each individual's counselling approach, their goals and responsibilities, the meaning of genetic counselling ethics for them, and any concerns about genetic counselling that they raised.

Biomedicine and Anthropological Theory

A brief discussion of anthropological approaches to Biomedicine concludes this introductory chapter and describes the theoretical framework this

study uses to interpret the data obtained from available publications, questionnaire responses, and interviews.

Anthropological investigations of Biomedicine were preceded by the sociology of medicine which concerned itself "with such factors as the organization, role relationships, norms, values, and beliefs of medical practice as a form of human behavior" (Cockerham 1982:4). Sociologists such as Friedson (1970) concentrated on professional roles and role conflicts in medicine, while others have dealt with the powerful socialization process of medical school (Becker *et al.* 1961; Fox 1957). Anthropologists who are building upon this sociological work emphasize the familiar anthropological concerns of cultural relativism, the sociocultural production of human behaviour, and the participant's, or 'emic', perspective. Moreover they recognize that there are a multitude of factors which must be taken into account when describing the context of Biomedicine.

Considered at first as the standard against which to judge the medicine of other cultures, Biomedicine has recently been described from a culturally relativistic perspective as "the preeminent professional ethnomedicine of Western cultures" (Gaines and Hahn 1985:18). In other words, Biomedicine is simply another ethnomedicine. Characterizing the essence of an anthropological approach Gaines and Hahn point out that Biomedicine is

> a sociocultural system . . . a cultural artifact, a complex human product shaped from human and nonhuman resources constantly responding to historical circumstances which are in turn human transformations of themselves and their environments. Humans in society constantly recreate structure from structure, in a process which Giddens (1976) characterizes as evidencing a 'duality of structure' (Gaines and Hahn 1985:5).

The anthropology of Biomedicine, then, does two important things. First, it acknowledges that clinical action in response to human ill health is produced by and influenced by historical, ideological, economic, and political fac-

tors (Chrisman 1984; Doyal 1979; Frankenberg 1980; Hahn and Kleinman 1983; Young 1982). Second, it recognizes that clinical action is the product of the individual practitioner's interpretation and experience that structures, embodies, and is influenced by, these factors.

This study incorporates the theoretical orientation described above and follows the lead of Arthur Kleinman, Robert Hahn, and Atwood Gaines in approaching the study of a Biomedical specialty by investigating the following four features:

> (1) a discernible domain of knowledge or belief, with concomitant values and rules for behaviour (Hahn and Kleinman 1982:5);
>
> (2) [a] division of labor and rules of proper action and interaction within the domain, with possibly distinct roles and institutions (Gaines and Hahn 1985:5);
>
> (3) [a] social and cultural means by which the sociocultural system is reproduced and altered (Gaines and Hahn 1985:6);
>
> (4) a realm of action in which these beliefs, values and rules are carried out (Hahn and Kleinman 1982:5).

My utilization of these four features differs slightly from that of Kleinman, Hahn, and Gaines in two respects. First, in collecting the ethnographic material I focussed on the domain of knowledge that concerns the needs of clients (rather than assessing genetic knowledge), the roles (division of labour), and the social and historical context in which genetic counselling occurs in order to discover the values and rules for action which influence and guide the provision of this health service. Second, I am interested especially in the individual genetic counsellor "as a human being, thrust into a role demanding unusual adaptation" (Maretzki 1985:32). This approach is new to studies of genetic counselling and to medical anthropology (see Hahn 1985, Lock 1985, Stein 1986--three examples of studies which do not assume physicians adhere

uniformly to the same medical model). However, as Maretzki points out, the role of the individual has been a longstanding concern in anthropology:

> Anthropologists devote much attention to the individual in the social group. Thus we wonder about the way in which healers assimilate their specialist knowledge with its powerful values and meanings (Maretzki 1985:24).

The theoretical issue to be addressed here concerns the response of individuals to the changes, tensions, and ambiguities that exist within their domain of action. The assumption persists that practitioners embody the general ideology of health care relatively uniformly and with little difficulty in subordinating personal value frameworks. In this study, to paraphrase Hahn (1985:53), I am interested in "the individual's versions of the culture" of genetic counselling. In particular, the relationship between the paradigmatic structure of genetic counselling and individual practitioners' values, beliefs and motivations surrounding clinical action has not been examined prior to this research. The theoretical issue here is not simply one of revealing a gap between ideology and practice; the existence of such a gap would hardly be unexpected. However, by acknowledging the role of the individual practitioner as a key interpreter and producer of Biomedicine, this study recognizes that "medicine is not one, but many medicines" (Hahn and Kleinman 1982:10).

In short, this study describes the variability that exists in the way in which genetic counsellors view their profession and explores the factors that contribute to this variability.

CHAPTER NOTES

1 For methods of conveying information to the client see Child's (1978) review of literature published from 1952 to 1972 and, more recently, Silverberg and Godmilow (1979), and Antley (1979). In the past decade, several studies have attempted to assess the impact of genetic counselling on client's reproductive behaviour (Evers-Kieboom and van den Berghe 1979; Oetting and Steele 1982) and on the process of reproductive decision-making (Lippman-Hand and Fraser 1979a, 1979b; Wertz *et al.* 1984).

2 Keywords from the International Survey of Medical Genetics

Some of the keywords that may appear in ethical reasoning are

rights	obligation
responsibility	duty
harm	benefit
burden	choices
should be	preserve
ought to be	protect
knowledge	confidentiality
autonomy	future
truth	dishonesty
decision	relationships
	allocation of limited resources

(Fletcher *et al.* 1985:6).

CHAPTER ONE

Genetic Counselling: Historical Process and Paradigms

The intention of this chapter is to describe the historical context of genetic counselling in Canada and to outline the two paradigms which currently influence this health service. To do so I will outline the general development of medical genetics, the history of the discipline in the United States and in Canada, and review the shifting values and objectives of genetic counselling as they have been described for the United States, since comparable data do not exist for Canada.

A useful beginning to this chapter concerns the definition of the frequently and mistakenly interchanged terms human genetics, medical genetics, and clinical genetics. Childs and Davidson have recently distinguished between these three fields, as follows:

> Human genetics . . . encompasses the diversity of disciplines that contribute to knowledge and understanding of the genetic, environmental, social and familial factors that interact to produce and control human variablility. It includes anthropology, psychology, the social sciences and medicine.
>
> Medical genetics is, then, a branch of human genetics that deals with the broad application of genetics to medicine. . . .
>
> Clinical genetics . . . refers to a narrower view of medical genetics, the part devoted to the clinical encounter, including diagnosis and management (Childs and Davidson in press draft:6-7).

In order to reflect the diversity of perspectives held by the participants in this study I have not selected one of the many existing definitions of genetic counselling. Instead, I have defined genetic counselling broadly as a specialized health service providing information and assistance to families at risk of genetic disorders. The issue of defining genetic counselling is discussed later in this chapter and at several other points in the thesis. The field of human genetics emerged from a variety of areas of inquiry. Historical accounts of human genetics have generally not investigated non- or pre-Mendelian explanations for heredity. One text on human heredity mentions the ancient Greeks' knowledge of red-green colour blindness in some families and the second century A.D. Hebrews who were aware of the association of hemophilia with an individual's sex (Brennan 1985:129). Explorations of cross-cultural concepts of human heredity are noticeably absent.

Early physical anthropological research into human variability by craniometry, anthropometry, and racial typologies, plant and animal studies, and the rediscovery in 1900 of Mendel's principles of inheritance all contributed to the emerging study of human heredity (Gould 1981). Two individuals are regarded as the elders of human genetics: Sir Francis Galton for his studies of twins and his attempts to quantify human variability and Sir A.E. Garrod for his 1902 paper on 'inborn errors of metabolism' (Reed 1974:336). Galton's contributions to applied human genetics have been somewhat muddied in the public's view by his involvement in 'eugenics', a term he used in 1883 to describe his advocacy of "the regulation of marriage and family size according to hereditary endowment of parents" (Gould 1981:75). The clinical implications of Garrod's work lay unrecognized in medical genetics until 1941 when the one-gene one-enzyme mechanism was suggested (Cohen 1984:66).

In the 1950s advances in human genetics included the identification of the human diploid chromosome number (Kevles 1985:238), a chromosomal disorder (Down syndrome) by Lejuene and his colleagues in 1959 (Kevles (1985:247), and techniques for chromosomal analysis; estimates of recurrence risk for several Mendelian disorders became available as well (Fraser 1979). In the following years, the means to treat some inherited disorders and to identify affected individuals and carriers opened the doors for medical practitioners to apply genetics to clinical practice.

Emergence of Genetic Counselling in the United States: Background

In the United States, the only country for which the history of genetic counselling has been reported in detail, human genetic studies in the late 1800s and early 1900s were rapidly integrated with the currently acceptable ambitions to weed out socially unfit, diseased, or physically defective individuals or races. The American eugenics movement never reached the extremes of Nazi racism and was certainly not as strongly endorsed by government legislation, but human geneticists in America were undoubtedly influenced by the eugenics philosophy of improving the human race through regulated reproduction. In the view of one geneticist I interviewed, the tenor of early applied human genetics has left genetic counselling "still labouring under the blush of the eugenics movement."

Statements about the institutional and organizational development of genetic counselling do little more than chart the increase in the number of counselling centers. Reed (1974:335) says there were ten centers in 1951 in the USA, while Rainer (1979:295) suggests only seven in the USA and Canada in 1952. By 1968 there were 156 centers in the USA (Lynch *et al.* 1983:iii). Reed

(1974:336), referring to the 1973 "International Directory of Genetic Services", indicates there were "387 centers in the United States and a total world-wide count of 890 human genetics units." Although the numbers quoted are of varying reliability (one problem may lie in defining what a "center" is), all authors agree there has been a tremendous growth of genetic services in the last ten to fifteen years.

The institutional beginning for genetic counselling in the USA was at the Eugenics Record Office in Cold Spring Harbor, New York in 1905 under the direction of "America's leading eugenicist", C.B. Davenport (Kevles 1985:54).[1] Davenport's 'fieldworkers' were sent out to gather family histories and

> to ferret out human hereditary data by making house-to-house surveys and by scrutinizing the records of the nation's numerous prisons, hospitals, almshouses, and institutions for the mentally deficient, the deaf, the blind, and the insane (Kevles 1985:54-55).

When the Eugenics Records Office was closed in 1940, human genetics had begun to seek a new respectability separate from the excesses of eugenics (Kevles 1985:199). Kevles argues that although there was continued resistance to genetics from mainstream medicine during the first half of the century, that attitude changed during the 1950s as research turned from the genetic basis of mental deficiency and social characteristics to the genetic basis of disease. In 1957, this interest was legitimated with the establishment at John Hopkins Medical School of the first formalized training programme in clinical genetics in the United States (Kevles 1985:233).

The consultation between individuals or families and genetic specialists was not termed "genetic counselling" until well into the 20th century. "Genetic advisory services" were being provided in 1940 at the University of Michigan's Hereditary Clinic and in 1941 at the University of Minnesota's Dight

Institute (Kevles 1985:253). In 1949, dissatisfied with the eugenic undertones of labels like 'genetic hygiene', Dr. Sheldon Reed at the Dight Institute proposed the term "genetic counselling" (Reed 1974:335). Acceptable to the Institute for lack of a better suggestion, the term took hold. The extent to which the term continues to be endorsed or its meaning agreed upon by professionals in this study is discussed in a later section.

Today in the United States, there are several hundred genetic counselling centers (Lynch *et al.* 1983). Most offer a wide array of services; some offer only prenatal diagnosis, or deal only with specific disorders--Huntington's Disease, Tay-Sachs Disease, sickle cell anemia, and hemophilia in particular. Since 1980 medical genetics has been recognized as a medical specialty with an American Board of Medical Genetics and there are now numerous scientific organizations specifically for human geneticists (Kenen 1984:545; Kevles 1985:292). Medical educators, however, have been slow to provide training in clinical genetics for physicians. A recent President's Commission on Human Genetics states that

> a report on medical school curricula found that 30% of the 104 [American] medical schools studied offered no formal education in genetics. The 70% that did provide training in genetics devoted varying degrees of emphasis to the subject (President's Commission 1983:65-66).

There are specialized training programmes for medical geneticists at the level of postgraduate medical education. In addition, a two year Master's level programme in genetic counselling has been in existence at Sarah Lawrence College since 1969 (Kenen 1984:543) and similar degree programmes have sprung up at eleven other American colleges (Mertens, Hendrix and Kenkel 1986).

Emergence of Genetic Counselling in Canada: Background

Thus far, the origin and institutional development of genetic counselling have been discussed for the United States only. A comparable review of the development of genetic counselling in Canada is not possible. A search of the literature produced no material related to the history of genetic counselling in Canada. In fact, the only articles found on the history of medical genetics in Canada are both limited to a description of the career of Dr. Madge Macklin, a medical geneticist who worked at the University of Western Ontario between 1922 and 1945 (Soltan 1967; Soltan 1985). Macklin's work was primarily in the realm of medical education and it is in that context that her influence will be discussed in the next chapter.

Hoping to sketch a rough outline of the history of Canadian genetic services, I wrote to several medical geneticists who have practised for many years in Canada to ask for help and information in this matter. Unfortunately, this method failed to produce much useful material. However, I did learn that a history of medical genetics in Canada, not genetic counselling specifically, is being compiled for the Canadian College of Medical Geneticists. This collection of essays will not be available until 1987 at the earliest.

The founding of the Canadian College of Medical Geneticists is one area in the development of Canadian clinical genetics for which information is available. The expressed mandate of the College was "to establish and maintain professional standards of health care delivery in the field of medical genetics" (Miller 1975:358) but as one member described it to me, the "C.C.M.G. was founded in 1975 to try and bring order out of chaos". That order has been brought about primarily through the establishment of training protocols for geneticists (clinical geneticists, biochemical geneticists, and cytogeneticists), an

accreditation process for MD or PhD geneticists to the level of Fellow, and the evaluation and accreditation of training centers.

The Canadian College of Medical Geneticists applied to the Royal College of Physicians and Surgeons (Canada) to have medical genetics recognized as a certified medical specialty. Their bid for specialty status, had it been successful, would have had far-reaching implications for the development of medical genetics in Canada. The Royal College turned down the bid, arguing that medical genetics is not distinct enough to warrant changing its status to a free-standing medical speciality (Davidson 1986:personal communication). Thus, there is no licensing mechanism for medical geneticists in Canada. Practitioners of medical genetics who are specialists in pediatrics, for example, are recognized by provincial colleges of physicians and surgeons and claim their services from provincial health care plans under those specialties. The decision of the Royal College does not directly affect PhD geneticists since their services are not covered by health care plans, but the College's decision certainly does not increase the professional status of medical genetics. A further implication of not being a recognized medical specialty is that medical genetics remains a very low priority in medical school curricula.

In addition to its accreditation functions, the Canadian College of Medical Genetics represents the interests of its members in negotiations with provincial governments on matters of funding and the provision of genetic services (Miller 1975:358). The annual CCMG meetings provide a forum for exchange of information and maintenance of professional and personal contacts among geneticists. At present, the CCMG is restricted to medical geneticists with the MD or PhD degree although a possible affiliation with nonMD/nonPhD genetic counsellors is being investigated.

Attitudinal Studies of Genetic Counsellors

Although detailed historical accounts of the development of medical genetics and genetic counselling in Canada are not yet available, I have employed alternative means to describing the ontology of beliefs and attitudes encountered among contemporary genetic counselling professionals. First, I review the studies of value orientations found among early and contemporary American genetic counsellors in order to understand both the general historical processes which have led to contemporary genetic counselling and the ways in which genetic counsellors are characterized. Second, using Canadian data wherever possible, in the following chapter I examine the information available on the contemporary context of genetic counselling to begin to determine the forces which continue to shape genetic counsellors' orientations.

By using data from American studies I do not wish to deny the existence of differences between Canadian genetic counsellors and their counterparts to the south. I use the studies primarily because comparable Canadian data do not exist, but also because I am certain there are parallels between the two groups. Similarities in the cultures of the two countries are strong (some anthropologists speak of a North American culture) and connections between the development of medicine and genetic counselling in the United States and Canada are real. For example, the accreditation standards of the Canadian College of Medical Genetics were used as a prototype in America (Scriver *et al.* 1978:950). Many Canadian genetic counselling professionals have trained in the United States, some are of American origin, and the master's level training programme for genetic counsellors at McGill University is modelled after American ones (Marks 1984). Since the American studies reviewed in this next section provide general statements about the attitudes encountered among genetic

counsellors, rather than specific historical details, I am encouraged to use them. Influences on genetics and genetic counselling in Canada probably come from at least two other primary sources, Britain (the training ground of many Canadian geneticists) and France (particularly for Francophone geneticists in Quebec), although the information to detail these connections is lacking.

Sociologist Dr. James Sorenson has been involved in several studies of genetic counsellors in the United States (Sorenson and Culbert 1977; Sorenson and Culbert 1979) and, more recently, he is part of a research team investigating ethical attitudes of medical geneticists in 19 countries (Fletcher *et al.* 1985:personal communication). Recognizing the previously unstudied role of the genetic counsellor as an influential one, Sorenson focusses on categorizing the variety of attitudes and approaches found among genetic counselling professionals. Sorenson has finds this diversity of opinion in two primary sources--published definitions of genetic counselling and the results of his own survey of attitudes of American genetic counsellors. Briefly, Sorenson (1979) divides the types of definitions of genetic counselling into three groups:

1) preventive medicine aimed primarily at reducing the incidence of birth defects;

2) the facilitation of decision-making through educating clients about the genetics of their problem and the options available to them;

3) psychological counselling as an integral component of medical genetics.

Investigating the strategies genetic counsellors employed to see that their clients made decisions, Sorenson and Culbert (1977) sought to characterize the ways in which counsellors implement the goals of genetic counselling. In a later paper, Sorenson (1979) describes these three kinds of strategies as follows:

> 1) [About 60 per cent] held a guiding strategy towards] assuring that counseling results in disease prevention" (Sorenson 1979:282);
>
> 2) [About 24 per cent] preferred a supportive strategy . . . that permits parents in risky situations to make decisions that are both technically informed and reflective of the values and goals of the parents (Sorenson 1979:282-283);
>
> 3) [About 15 per cent] adopt an advising strategy . . . in which the counselor appears to be interested not only in disease prevention but, in addition, has an interest in both the genetic and the social implications, economic and otherwise, of reproduction among those at risk for having a genetically diseased child (Sorenson 1979:282).

Sorenson and Culbert (1977) found these differences in counsellor orientations to be significant ($p<0.05$). In examining the preferences counsellors had for goals and strategies, the researchers discussed the degree of directiveness in counselling as an important indicator of these preferences. The counsellors most likely to be directive, to tell patients what they ought to do, were those who endorsed an advising strategy and to a lesser degree those who practised preventive medicine through a guiding strategy (Sorenson and Culbert 1977:144). The one-quarter of counsellors who preferred an educative-supportive strategy "were much less willing to view their role as an advisor" and were less likely to tell patients what to do (Sorenson 1979:282).

A subsequent interpretation of their data showed that three-quarters of the counsellors they surveyed placed a priority on providing disease and risk information to their patients, 19 per cent felt disease and risk information were of equal importance to discussing psychosocial issues, while only six per cent viewed the discussion of psychosocial issues as a priority (Sorenson and Culbert (1979:92).

As mentioned above, Sorenson and his colleagues have focussed on describing and classifying the variety of opinions found among genetic counselling professionals rather than determining the context or consequences of this

variety. In a 1979 paper Sorenson does review the history of applied human genetics in the United States and several studies of the effectiveness of genetic counselling, but his work does not investigate either topic in detail. Nor does it examine the origin or impact of such conflicting and varied orientations for individual counsellors--a concern of my own study. An understanding of the context in which this diversity of professional opinions occurs is achieved in my study by examining, first, the historical changes in goals and protocols among genetic counsellors in the United States, and second, the contemporary situation of genetic counselling in Canada.

Historical Development of Genetic Counselling in North America

There have been changes in the stated goals of genetic counselling. Several authors, Epstein (1977), Kenen (1984), Kessler (1980), and Kevles (1985) in particular, describe the historical processes in the study of human genetics that have influenced contemporary genetic counselling. Although stated in different ways, each works note a change from goals of "risk and prevention to communication and counselling" (Epstein 1977:334).

Kenen (1984) and Kessler (1980) see the development of genetic counselling as a series of changes in the occupation of practitioners providing genetics to the public. As each occupational group entered genetic counselling it has left its mark in the way of establishing priorities and protocols for accomplishing its goals. PhD geneticists, the first genetic counsellors, are characterized as research-oriented individuals who emphasized statistical risks as fundamental to understanding genetic information and to achieving the goal of preventing genetic disease in the human population. Physicians, entering genetics in the 1950s, emphasized preventive medicine, a reduction in the

incidence of birth defects in families through the education of prospective parents. Most recent entrants are individuals trained in paradigms which emphasize the psychosocial issues of genetic disorders who are felt to advocate nondirective and value-free supportive counselling. This triad of occupational interests in the history of genetic counselling is described by Kessler (1980) as a series of paradigmatic shifts from eugenics to preventive medicine to psychological medicine. Kenen's (1984:547) summary of the transition in genetic counselling from "research-,. . . to clinically-,. . . to people-oriented professionals" is reproduced in Table 1.1.

Kevles (1985) discussion of the changes in orientation of genetic counsellors differs somewhat from the others. Recall from the section on the history of genetic counselling in the United States that Kevles did find strong ties between the early genetic counsellors, PhD geneticists,--and eugenics. In contrast to Kenen (1984) and Kessler (1980) who continue to associate PhD geneticists with the eugenicist goals of preventing genetic disease, Kevles is less willing to do so. Rather he is concerned with demonstrating the transition of eugenics to a "science of human heredity" (Kevles 1985:231-232).

My own survey of the numerous published attempts to define and determine the boundaries of genetic counselling suggests that most counsellors would agree that they are providing information to those who seek assistance in matters of human heredity. In addition, genetic counsellors want individuals to use that information (risks, options, treatment, *etc.*) to make decisions that are "informed", "rational" and "responsible". Finding agreement for generalizations about objectives beyond those two is much harder to do. The kind and quantity of 'information' to provide clients is widely debated. Who should counsel? How can genetic counselling best achieve its goals? Even the timing of the

information-giving process is felt to be important. What constitutes "assistance"--is it simply the presentation of genetic information alone, does it include outlining and exploring the options available to families or does it involve ongoing emotional support as well?

TABLE 1.1

Ideal Type Characteristics of Occupational Categories in Genetic Counselling

Occupation	Educational degree	Degree of claimed expertise	Status	Monetary reward	Primary goal	Organizational setting	Public perception of occupation	Predominant sex of incumbents
Physician	M.D.	High	High+	High+	Curing	Hospital	Diagnosis-remediation	Male
Biologist-geneticist	Ph.D.	High	High	High	Discovery	Academic department	Research	Male
Genetic counselor (associate)	M.Sc.	Medium	Medium	Medium	Social support	Hospital clinic	Counseling	Female

(Kenen 1984:542).

One pattern of variation in objectives exists between textbooks of clinical genetics and articles on genetic counselling. Textbooks of clinical genetics tend to focus on the medical and genetic goals and their attendant techniques of diagnosis and inheritance risk calculation. An example of this approach is the following:

> Genetic counselling is 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 and transmitting it and of the ways in which this may be prevented or ameliorated (Harper 1981:3).

Articles on "genetic counselling", on the other hand, tend to draw the reader's attention to the importance of dealing with the clients' responses to the information provided to them. The following definition, widely quoted in this type of literature, emphasizes the expectation that the genetic counsellor will provide a non-directive, value-free environment in which the client can make a decision based on medical facts, genetic contingencies, and personal goals.

> Genetic counseling is a communication process which deals with the human problems associated with the occurrence, or the risk of occurrence, of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained persons to help the individual or family (1) comprehend the medical facts, including the diagnosis, the probable course of the disorder, and the available management; (2) appreciate the way heredity contributes to the disorder, and the risk of recurrence in specified relatives; (3) understand the options for dealing with the risk of recurrence; (4) choose the course of action which seems appropriate in view of their risk and their family goals and act in accordance with that decision; and (5) make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder (Fraser 1974:637).

Table 1.2 summarizes the ways in which the goals of genetic counselling and the orientations of genetic counsellors have been classified and characterized. Although the table simplifies the issues a great deal, it is a useful means for structuring the summary of studies reviewed above. The classifications of genetic counselling professionals' orientations represented in Table 1.2 and the preceding discussion are derived from historical treatments of the field. However, the characterizations persist in the literature, in the mind of the public, and even among today's practitioners of genetic counselling. To some extent these characterizations have become stereotypes.

TABLE 1.2

Summary of Genetic Counselling Professionals' Goals and Orientations as Characterized in Publications*

Discipline of Origin, Training, & Orientation	Science	Medicine	Psychology
Practitioner:	PhD	Medical doctor	nonMDs/nonPhDs (e.g., nurses, social workers, master's level Genetic Associates)
Orientation:	Research	Clinical	People
Major Goal:	Prevention of genetic disease	Curing and prevention of disease through client education	Dealing with clients' psychosocial responses
Method of Counselling Characterized as:	Directive	Slightly less directive	Non-directive
Subject Matter Characterized as:	Value free	Value free	Value laden

*Based on the following:
Kenen (1984)
Kessler (1980)
Sorenson (1979)
Sorenson and Culbert (1977)

Recall from the work of Sorenson and Culbert (1977 and 1979) that the variety of goals embedded within definitions of genetic counselling have been categorized into three types--preventing birth defects, educating clients and facilitating their decisions, and psychologically-oriented counselling--each of which seems to conflict in some ways with the others. Recall also that three paradigmatic changes in the history of human genetics have been outlined--from eugenics, to preventive medicine to psychological medicine. I argue that in reality the historical process has been more of an accumulation of competing goals rather than a complete shift since the objectives to provide recurrence risks and to prevent genetic disorders now exist alongside those to counsel. This accumulative process can be discussed in terms of two paradigms -- a medicoscientific and a psychosocial paradigm.[2]

The Basic Paradigms

The following description of the paradigm of medicine and science is based on the writings of two distinguished medical geneticists, Charles Scriver (1977; Scriver *et al.* 1978) and Barton Childs (1977a), and two medical anthropologists (Hahn and Kleinman 1982). I have used the terms medical and medicoscientific paradigm interchangeably here. What characterizes the medical paradigm? First, the paradigm is oriented towards the discovery, treatment, cure and/or prevention of disease. This is the orientation of emphasis in medical schools and in medical genetics research programmes. These diseases are felt to be caused by phenomena that are natural and that can be determined by objective tests. Disease and its cause are viewed as entities which can be treated in isolation from their social context (Hahn and Kleinman 1982:33). Second, the paradigm has fostered the view that the responsibility of medical professionals

is "episodic" (Scriver *et al.* 1978:947) and limited "to the immediate care of patients" (Childs 1977a:7). Third, medicine has only recently begun to incorporate the psychosocial ramifications of disease and to suggest that physicians have a responsibility to deal with these aspects of patient care. Fourth, the practitioner is assumed to be able to communicate his or her knowledge in a manner that is free from value judgements.

The psychosocial paradigm has, in part, arisen in response to criticisms of the medical paradigm. A powerful image among today's health care consumers is that of the all-caring family physician who did not just treat the patient's disease, but who "really cared" about his patients and their families. Eisenberg and Kleinman (1980:3) argue that this image of the "good old days of medicine" is mythical, but the vision has become a symbol of past satisfaction with medicine in contrast to feelings of alienation and discontent with modern medicine which is perceived as impersonal and technological. Public demands for a more "holistic" and "person-oriented" approach have resulted in the entry into health care of professionals whose disciplines of orientation lie within a psychosocial paradigm (*e.g.*, social work, psychology). The first characteristic of the psychosocial paradigm is an emphasis on the emotional and social nature of and response to 'illness'. Second, it stresses that illness treatment may involve a number of visits between counsellor and client over a long period of time (Applebaum and Firestein 1983:8; Kelly 1977). Third, the treatment is noninterventionist and nondirective (*e.g.*, Rogerian therapy) and consists in large part of "listening to" and "facilitating" the clients' concerns (Applebaum and Firestein 1983:7; Dodge 1983; Rapp 1985:6-7). Although psychosocial counsellors are expected to be nonjudgmental, the topics they deal with are recognized as often emotionally charged and value laden.

Links Between the Two Paradigms

In addition to a recent generalized concern for changes in health care, the introduction of a psychosocial orientation into applied human genetics has come about for a number of reasons. First, several studies demonstrated that very little factual information was being retained by the client, that risk figures were not always a primary factor in client's decisions, and that genetic disorders can have profound and long-lasting emotional consequences (see reviews of these studies contained in Childs 1978; Evers-Kiebooms and van den Berghe 1979; Falek 1984; Lippman Hand and Fraser 1979a; Lippman Hand and Fraser 1979b). A second source of change in applied human genetics was the increased availability of safe and reliable prenatal diagnosis and therapeutic abortions. The result of these changes in genetic counselling is reflected in Fraser's definition and mandate for the field (see page 31). Items 1, 2, and 3 of the definition are familiar medical goals of "education and communication of facts [about diagnosis, risks, prognosis, and management] in the most objective fashion possible" (Murray 1976:12-13). Items 4 and 5 stem from psychosocial concerns for nondirective counselling to help the client act as he or she decides and to cope with the disorder. Genetic counselling has clearly established its goals in both the medical and psychosocial paradigms.

The scope of genetic counselling, the range of factors that are now associated with genetic conditions, and the association of a medical and a psychosocial paradigm "demands a great deal from its practitioners" (Applebaum and Firestein 1985:4). Ideally the genetic counsellor must be skilled in both 'genetics' and 'counselling' in order to respond to the genetic and social particulars of each case. In reality, "there is no one 'correct' way to handle every

situation" (Applebaum and Firestein 1983:11). Rather, as I will show, there are individualized responses to the pressures of two competing paradigms.

As a result of the accumulation of goals and orientations, genetic counselling is now felt by many observers to be ambiguously defined, poorly circumscribed, and a source of role strain for both clients and practitioners. To be sure, generalizations about exactly what genetic counselling professionals do, what they are expected to do, and what they feel comfortable doing are not easy to make. Individual counsellors--MDs, PhDs, and others--do not always fit neatly into one type of paradigm or goal orientation. As Sorenson suggests, "in the actual practice of counseling it is highly likely that any individual counselor would exhibit elements of each of these orientations" (1979:279). Each individual manifests and has been influenced by these competing paradigms and orientations. The practice of genetic counselling for any one individual is a matter of negotiated and flexible roles and responsibilities that exist within a particular structural context. It is to this context of influence that I now turn.

CHAPTER NOTES

1 There are a number of different dates cited in the literature for the establishment of the Eugenics Record Office. Reed dates the opening of the Cold Spring Harbour lab at 1904 (1979:333), although Kevles (1985:54) says the Eugenics Record Office did not open until the following year. Sorenson (1979:275) places this second event in 1910 and Fraser (1979) mentions 1915.

2 I prefer the term 'paradigm' rather than 'model', although the term Biomedical Model is often used (Engel 1977). 'Paradigm' suggests process and practice, a body of beliefs that is constantly changing shape and meaning for its practitioners. 'Model' implies a more rigid and static formulation and application of knowledge.

CHAPTER TWO

Genetic Counselling: Contemporary Context and Paradigms

The intention of this chapter is to describe the contemporary context of genetic counselling in Canada. I have organized my description in terms of the two paradigms which structure this context--the medical, or medicoscientific paradigm, and the psychosocial paradigm.

Medicoscientific Paradigm

Genetic counselling professionals are influenced by medicine and science in a number of significant ways. First, discussions of genetic matters are couched in the language of medicine and science ("proband", "patient", "autosomal dominant inheritance", "diagnosis and treatment of genetic disease"). Second, the majority of professionals providing genetic counselling are trained in either a scientific or medical context (in laboratories, medical schools, or hospitals). Third, genetic services are organized, situated and brought to the public in a medical context. Each of these links to medicine and science is discussed in detail below.

A. Medical Genetics: Current Capabilities

An adequate picture of the contemporary context of genetic counselling rests on a supporting knowledge of the content and capabilities in medical genetics. Medical and clinical genetics draw from several sub-branches of genetics: population genetics (the investigation and mathematical analysis of

genetic variation within and between populations), cytogenetics (the study of chromosomal defects), biochemical genetics (the analysis ofinborn errors of metabolism caused by enzyme defects), and molecular genetics (the study of genetic disorders at the level of DNA).

The human body probably contains more than 100,000 genes--the units of heredity that determine what we look like, and how our bodies function, and that contribute to our behaviour. These genes are arranged on 23 pairs of chromosomes in each human cell. Each gene contains the instructions for the manufacture of proteins. It is the arrangement of these genes on the chromosomes and the resulting combinations of proteins that produce and direct "the 100 trillion specialized cells that an adult human being possesses" (Baskin 1982:54). Abnormal or harmful variations in human genes and their manifestation in the individual are the fundamental interest of medical geneticists and other genetic counselling professionals.

The most recent edition of McKusick's *Mendelian Inheritance in Man* (1983)--the basic catalog of human genetic disorders--lists 3368 single gene disorders (McKusick 1983:xi). In addition, genetics plays a role in an unknown number of "multifactorial" disorders--problems caused by a combination of genetic and environmental factors. A "genetic disorder" can be caused by a non-typical arrangement of genes, a deletion of genes, or the appearance of a new and harmful gene.

Genetic disorders and diseases are classified in a variety of ways, all of which reflect a close historical and continuing link between genetics and science and medicine. (The following discussion is based on a number of textbooks of human genetics including Brewer and Sing 1983; Cohen 1984; McKusick 1983; Thompson and Thompson 1986.) McKusick's catalogue classifies

all of the single gene disorders by the way in which they are inherited at the level of the genes: "autosomal dominant", " autosomal recessive", and "X-linked" genetic disorders. Other types of genetic disorders include chromosome abnormalities (*e.g.*, Down Syndrome, also referred to as Trisomy 21; sex chromosome variations like Klinefelter's syndrome 47); the inborn errors of human metabolism which affect the production or activity of enzymes (*e.g.*, PKU, phenylketonuria, a deficiency of phenylalanine hydroxylase for which most North American newborns are routinely screened); the hemoglobin disorders (*e.g.*, the thalassemias). Each of these disorders is classified according to an abnormal structure or function at the cellular level. "Population-specific inherited disorders" (sickle cell anemia among North American blacks, for example) group those conditions which have a higher incidence in certain populations. The designation, "birth defect" or "congenital anomaly," encompasses a broad range of conditions that are often visible in the newborn and have a variety of genetic and non-genetic etiologies. In addition to the disorders listed above, neural tube defects, believed to be caused by a combination of genetic and environmental factors, or other morphological malformations like cleft palate (sometimes caused by teratogens such as infectious or chronic disease in the pregnant woman, exposure to x-rays, or chemicals or drugs--environmental factors) are all considered birth defects (Cohen 1984:99).

In addition to classifying disorders, geneticists are interested in identifying individuals in a population who are carriers of an abnormal gene or who may be at risk of a particular genetic disorder. Again, the language and context is rooted in medicine and science. "Carriers" are individuals who do not show the effect of an abnormal gene they carry (hence the term "recessive gene") but may pass it on to their offspring. Some carriers can be identified by

a variety of tests. "Genetic screening", as this mass testing is called, is usually done on newborn infants or adults in high risk populations. Newborn screening is available for a wide variety of genetic disorders like PKU (phenylketonuria) and galactosemia which can lead to severe mental retardation if left untreated (Thompson and Thompson 1986:93,95). Carrier screening is possible for several population-specific diseases including Tay-Sachs disease (Ashkenazi Jews), beta-thalassemia (populations of Mediterranean origin), and sickle-cell anemia (predominately in blacks, but also in Mediterranean peoples) (Cohen 1984:23-24;Thompson and Thompson 1986:85). Screening is offered to people in order to identify them for preventive treatment before the disease manifests itself, to make individuals aware of genetic factors in their reproductive decisions, and for scientific reasons such as determining the existence of a particular gene in a population.

Individuals who have been identified as carriers of genetic disease or who have reason to suspect they may be at risk of transmitting an inherited disorder to their children often undergo prenatal diagnosis. A 1981 article in the *American Journal of Obstetrics and Gynecology* lists 182 genetic disorders and an unspecified number of chromosomal disorders which can be diagnosed prenatally (Stephenson and Weaver 1981). Access to prenatal diagnosis is controlled by physicians and is not available to every woman who wants it. A woman may be unable to obtain prenatal diagnosis if she is under a certain age (in most provinces, 35 for amniocentesis), or if no fetal disorder is suspected. It is, for example, extremely difficult to have prenatal diagnosis done simply to determine the sex of the fetus if some X-linked disorder is not suspected.

The primary means of detecting abnormalities *in utero* is by withdrawing a sample of fluid from the amniotic sac which surrounds the develop-

ing fetus. Cells in the fluid obtained by amniocentesis are cultured and then examined for biochemical and chromosomal abnormalities. Other prenatal diagnostic techniques are ultrasound (the visualization of the fetus on a screen by moving a scanner over the woman's abdomen), fetoscopy (insertion of a fiber-optic instrument into the uterus to look directly at the fetus or to take samples of fetal skin and blood), *chorionic villus* sampling (the examination of cells from the developing placenta), and radiographic (or X-ray) examination of the fetus.

For genetic disorders that cannot be detected *in utero* or in instances when there is no pregnancy, geneticists are usually able to make a diagnosis by other means. A physical examination and a karyotype (counting and assembling photographed chromosomes into a standardized array) of the affected individual or family member may be done. The geneticist or a pathologist may examine and test the "products of conception" (an aborted embryo or fetus, the placenta and any amniotic fluid). A further source of diagnostic information is a family history or pedigree detailing the fertility and pregnancy history of individuals and the appearance of any possible genetic disorders.

The diagnosis, associated pattern of inheritance, and family history are the geneticist's tools with which to determine the statistical risk that a particular disorder will appear in a client or client's offspring. Laboratory test results are the preferred means upon which to base a diagnosis and subsequent calculation of risk figures. Every pregnancy involves a three per cent risk that some genetic abnormality will be present in the fetus (Thompson and Thompson 1986:303). The calculation of risks for Mendelian disorders is determined by straightforward principles of inheritance. (For example, the child of a normal parent and an individual with Huntington's disease, an autosomal dominant disorder, has one chance in two of having the disease.) For non-Mendelian disor-

ders, like cleft palate, there are empiric risks that are population-based and risks associated with a particular family. Other risk factors that may be revealed by taking a family history are maternal age, paternal age, consanguinity (the offspring of people who are related by blood) and exposure to teratogens or environmental substances that cause birth defects. Understanding the diagnosis, the associated risks, and their calculation are considered essential information to provide to clients.

After the disorder has been diagnosed and the risks of inheriting or passing on the condition have been calculated, the client is provided with information about the prognosis and possible treatment--"genetic management". Although geneticists are familiar with a wide range of inherited disorders, many of which can be diagnosed prenatally, and for which the risks of occurrence are calculable, the therapuetic capacity of medical genetics still lags. A vivid example of this disparity between diagnostic and therapeutic options is in Tay-Sachs disease, a genetic disorder occurring in about one in 3600 births in Ashkenazi Jewish populations (Cohen 1984:78). Tay-Sachs carriers can be detected, the disease can be diagnosed prenatally, but aside from prevention through abortion, there is no treatment--affected infants experience mental retardation, physical disabilities, seizures, and die at an early age (Cohen 1984:78). On the other hand, certain genetically-based metabolic disorders can be successfully treated (for example, the treatment of PKU by controlled diet). "Genetic management" can include referral for psychiatric counselling, but that referral signifies the end of the genetic counsellor's involvement.

B. Education and Training of Genetic Counsellors in Canada

Although attention is constantly drawn to the relevance of genetics for medicine, the undergraduate medical education of physicians includes little genetics. In Canada, an early proponent of genetic education for medical students was Dr. Madge Macklin at the University of Western Ontario at London. Her longstanding efforts to see "that medical students . . . [are] trained in the fundamentals of genetics in premedical work and in the applications of science to medicine in their clinical work" (Macklin 1938:96) have not been fully realized. A recent survey of the curricula in 127 American and 16 Canadian medical schools found only 65 schools in 1983-84 have "a separate genetics course" (Swanson 1984:39). The time devoted to genetics in these schools averaged 27 hours, and varied from five to 69 hours (Swanson 1984:39). The average of 27 hours for genetics, the lowest average of the ten preclinical disciplines surveyed, compares to an average of 233 hours for pathology, 150 hours for anatomy, and 129 hours for pharmacology. Thompson and Thompson (1986:4) maintain that although the total "time commitment is modest (about 0.5 per cent of the total hours), it should be enough to introduce students to the genetic point of view. "Students may take genetics as an elective, although the findings from the above study suggest only 2.9 per cent of nearly 6000 students (303/5981) in American medical schools did so. Medical students at one Ontario medical school rated all aspects of their genetics course (syllabus, text, lectures, seminars, and exams) at a level below "adequate"; in point of fact, they rated the course as "useless" or "needs improvement" (Arseneau *et al.* 1985:2). Genetics is not offered as a clinical clerkship at any of the 16 Canadian medical schools (The Association of Canadian Medical Colleges 1983:38).

Comprehensive training in medical genetics is provided at the graduate (*i.e.*, Master's or PhD level) or postgraduate level (*i.e.*, post-MD). For recent MD or PhD graduates this specialized training may be at a centre accredited by the Canadian College of Medical Geneticists. Physician geneticists are trained for a minimum of four years beyond their medical degree and PhD geneticists must do a minimum of two years. Successful candidates become accredited Fellows of the Canadian College of Medical Geneticists.

Many Canadian genetic counselling professionals are not Fellows of the CCMG and acquire their knowledge of medical genetics in other medically- or scientifically-oriented programmes. PhD geneticists probably have the most laboratory experience and coursework in genetics, but during their training they may have little contact with clients. Physicians who are providing genetic counselling may gain their knowledge of genetics during their hospital residency in a specialty such as pediatrics. Non-physician/nonPhD genetic counsellors achieve their genetic education in a variety of ways--as undergraduates, in formal Master's level programmes, "on the job" by attending seminars and genetics rounds at hospitals, or by reading current textbooks and medical journals. Entrants to the two-year genetic counselling diploma programme at McGill must have "University level courses in Principles of Human Genetics" and take further courses, labs, and clinics in human genetics (McGill University 1984).

With few exceptions, genetic counselling professionals who have PhDs, MDs, RNs, or Master's level degrees gain their competency in genetics in a medical-scientific context--the hospital or the lab. Not all genetic counselling professionals are physicians, but the fact that genetic counselling is practised within a semantic and institutional context that is exclusively medical reinforces its connection to medicine even for those without clinical training.

C. Available Genetic Services in Canada

> In Canada, three to five per cent of infants born each year have a significant congenital malformation, chromosome abnormality, or a clearly defined 'genetic disorder'. Hereditary disorders account for at least one-fifth of all infant deaths in the country. A major proportion (about 50 per cent) of pregnancies that miscarry spontaneously are associated with such conditions, in particular chromosomal aberrations and developmental defects.
>
> In addition to fetal loss and infant mortality, significant childhood and life-long morbidity may result from genetic disease and congenital malformation. Considerable medical and institutional costs are incurred, and psychological adjustments must be made by parents, siblings, and other family members (Rudd 1980:11-12).

These statistics speak loudly to the need for genetic counselling and screening services in Canada. Comprehensive descriptions of the range and accessibility of such services are outdated and incomplete. The following discussion of available genetic services is based primarily on annual reports and quarterly statistics I solicited from 15 major genetic counselling programmes across Canada. Reports were received from only four clinics, thus limiting the scope of this discussion. Secondary sources including a 1976 report of a "Task Force on Genetic Services" to the Ontario Council of Health (Ontario Council of Health 1976) and a survey of prenatal diagnostic centers published in 1980 (Miller 1980) were used. I have also included information obtained during conversations with genetic counselling professionals in Ontario.

Genetic counselling is provided in a variety of medical contexts to Canadians. There are fifteen major genetic counselling centers thus classified because they support a large number of specialized personnel (usually three or four medical geneticists, a research cytogenetist, genetic associates, technologists, and clerical staff), extensive diagnostic and laboratory capabilities, and they can offer the full range of genetic services to a large number of people. These

fifteen centers are situated in areas of greatest population density--in provincial capitals and major cities. Southwestern Ontario has five such centers; Quebec has three , Alberta has two; British Columbia, Saskatchewan, Manitoba, Nova Scotia, and Newfoundland each have one major centre. Prince Edward Island, New Brunswick, the Yukon and the Northwest Territories do not have major genetic counselling centers.

In addition to these large centers, genetic counselling is also offered through a number of smaller clinics or programmes that are often part of, or offered as one of, several health services at a hospital. For example, one such clinic in Ontario is offered as part of a range of services to mentally handicapped individuals and their families. This second type of genetics clinic generally has fewer personnel: perhaps one or two medical geneticists, a laboratory director with several technologists, and clerical support staff. These smaller clinics can generally offer prenatal diagnostic testing such as amniocentesis and ultrasound, but their laboratories may not be able to handle all diagnostic procedures. The number and distribution of these smaller centers is not known.

A third means of getting genetic services to the public are outreach clinics held in smaller cities and towns in outlying areas of each province. Affiliated with local Health Units, the outreach clinic is often coordinated by genetic associates, nurse coordinators, hereditary outreach nurses or social workers (sometimes called "fieldworkers"). As described in one clinic report, these individuals "arrange the clinic, assemble appropriate documents, visit families for obtaining genetic histories and provide ongoing support. They work closely with the local family doctors" (Calgary Annual Report 1983). On the clinic day, those families who have been referred will be seen at the local hospital by a medical geneticist from an urban genetics clinic. The frequency of out-

reach genetics clinics varies with regional demand, from once a month to once every three months. There are outreach programmes extending to most regions of Canada so that individuals probably have to travel no more than one day to obtain genetic consultations.

In addition to working in urban and outreach genetics clinics, geneticists may also see patients admitted to hospital. In this case, a referral from a physician or hospital social worker will bring a geneticist to a "ward consultation". Assistance for some individuals concerned about genetic matters may also be dealt with by letter or over the phone. At least one centre has a "teratogen hotline" to answer questions about possible harmful effects to a fetus from drugs, chemicals and other substances. And finally, a physician may offer advice about genetic matters as part of his or her general practice.

All major Canadian genetic centers are located in hospitals and many are either university affiliated or have their clinic premises provided by a university. Within the supporting institution, the genetics clinics may have close ties with Departments of Medical Genetics and with Departments of Pediatrics. In addition, if a genetics clinic is not able to support its own on-site laboratory, diagnostic services may be provided by the Department of Laboratory Medicine or a provincial laboratory.

Institutions play an important part in genetic services as a primary source of income for practitioners of genetic counselling. Genetic counsellors who have an MD can bill for lab work and consultation through provincial health care plans under either family practice or their specialty (pediatrics, for example). In addition to their income from health care plans, physicians are often employed by the university or hospital in which their genetic services are provided. Since the Canadian College of Medical Genetics is not a licensing

body, accredited MDs and PhDs are not able to bill as specialists in medical genetics. PhD geneticists receive their primary income from the supporting institution in which they work rather than from health care plans. As well, nonMDs and nonPhDs who are providing genetic services cannot bill for services and are paid out of genetic counselling programme budgets.

Although Canada has a fairly well developed network of genetic services, most Canadians are not aware of this health service until they are referred for tests or genetic counselling. Access to specialized genetic services is almost always by referral. Most genetic counselling clients are referred by family physicians, obstetricians or pediatricians. A smaller number of referrals come from public health nurses, social workers, and other community level workers. Some clients are referred from within hospitals and a minority are self-referrals. Individuals may be referred for a variety of reasons including the birth of a handicapped or stillborn infant, recurring miscarriages, a "positive" amniocentesis test, or a diagnosis of genetic disease in a family member.

Genetic counsellors provide information and assistance to their clients in a variety of settings - the counsellors' office, a consultation or examination room in a hospital, and in some outreach programmes in the client's home. Most counselling sessions are conducted by the geneticist, although a pre-amniocentesis or pre-ultrasound session may be done by a genetic associate or other nonMD/nonPhD. Because genetic issues are often matters of reproductive decision making, clients are usually male-female couples, or women or, less frequently, men who come on their own. At one large clinic in Alberta, 726 new patients were seen in 1983-84; 73 per cent (530) were women and 27 per cent (195) were men. If there is an affected individual (the "proband") in the family, he or she may attend and be examined by the geneticist.

The full range of current genetic diagnostic services are available in Canada, although access to those services varies regionally. Blood sample tests, karyotyping and alpha-fetoprotein level determination from amniotic fluid samples and ultrasound are widely available. Other prenatal diagnostic techniques such as chorionic villus sampling (cells are taken from the chorion, the precursor to the placenta, during the first trimester of pregnancy) have a more restricted distribution. In addition to prenatal diagnosis, cytogenetic analysis of bone marrow and skin tissue, biochemical testing for metabolic disorders, and some molecular genetic analysis is available within the Canadian genetic services system.

Demand for these services, based on clinic caseload reports, has grown steadily in Canada. For example, since 1978 the University of British Columbia clinic in Vancouver has had a fivefold increase in families seen (from 700 to 3792 in 1985). Hamilton's McMaster University-Chedoke Hospital clinic caseload has increased from 360 in 1981 to 621 in 1985. For each of the clinics that submitted annual or quarterly reports, there has been an appreciable increase in the number of prenatal diagnoses done. For example, Edmonton was doing about 50 amniocenteses in 1977 and 260 by the end of 1984. For the same period in Vancouver, the increase was from 93 to 1184, and last year (1985) this large clinic performed 1337 amniocenteses. The bulk of the increases in amniocentesis testing appears to be for "advanced maternal age". There is a well-established increase in fetal genetic disorders associated with women who are age 35 and over at the time of delivery; one clinic report states "approximately 50% of pregnant women 35 years and older now take advantage of the amniocentesis and the numbers continue to rise" (Davidson 1986:personal communication).

In addition to genetic counselling and prenatal diagnosis, Canadians also have available to them newborn screening and carrier detection for a wide array of genetic problems. All screening programmes in Canada are voluntary. A primary goal of screening programmes "is to reduce the burden and cost of genetic disease both in the individual and in society" (Clow 1982:390). From the standpoint of research and clinical medicine, this goal includes

> three major objectives [National Academy of Sciences 1975]: 1) to provide opportunities for medical intervention where treatment may neutralize the effect of the mutant gene; 2) to provide counseling about reproductive options; 3) to collect research data pertinent to public health policy and basic knowledge--*i.e.*, research and enumeration of gene frequency (Clow 1982:391).

Provincial and/or regional screening programmes operate out of clinic laboratories in Ontario, Quebec, the Atlantic region and in British Columbia. In Canada, approximately 98 per cent of newborns are routinely screened for phenylketonuria, PKU (Clow 1982:392). Screening for other metabolic disorders is available on demand at larger clinics. Carrier detection (and prenatal diagnosis) for sickle cell anemia, Tay-Sachs disease, and beta-thalassemia may be done routinely for high-risk individuals at a clinic or as part of a screening programme like the Quebec Network of Genetic Medicine (Clow 1982; Dagenais *et al.* 1985).

In addition to the standard and widely available procedures for prenatal and carrier screening described above, there are several 'high tech' specialized areas of reproductive technology and genetic therapy which bear mentioning. Reproductive technology includes alternatives for infertility or genetically risky conception such as "artificial insemination by donor", "*in vitro* fertilization", and the capacity for surgical or pharmaceutical treatment on unborn humans. "Genetic engineering" includes the technology to copy (clone)

genes, to identify, and in non-humans, repair and replace harmful genes. Although these specialized techniques are not widely available, most in fact are only experimental, and some may never be ethically permissible in humans, advances in reproductive and genetic technology are an important kind of information for the public about human genetics. With titles such as "Fetus Tests: Collision Course for America" (Dolnick 1985), magazine and newspaper articles about genetics are often couched in the language of the "Brave New World". Public knowledge of science and medicine may be initiated at school, but media coverage of exciting new discoveries is an influential means of updating that understanding. In fact for many Canadians, their only contact with issues genetic may be from media coverage.

Psychosocial Paradigm

While genetics, medicine , and science have had a strong and lengthy relationship, the links between genetics and disciplines like social work and psychology within a psychosocial paradigm are of a more recent origin and have a less well-defined shape in Canada. The fact that there is virtually no information on the counselling aspects of genetic services in Canada is in itself significant. There are no publications or studies evaluating the current contribution and capabilities of social work or psychology in Canadian genetic services. There are no descriptions or reports of the psychosocial services available to individuals faced with genetic disorders. Information about the psychosocial component of genetic counselling in Canada comes from three main sources. First, the acknowledgment that training programmes for genetic counselling professionals should include communication skills and second, the existence of nonMDs/nonPhDs such as social workers and psychologists in Cana-

dian genetic services suggests indirectly the influence of a psychosocial paradigm. The third source is the voices of genetic counselling professionals themselves.

A. Education

Training programmes accredited by the Canadian College of Medical Genetics now include competency in communication skills as well as expertise in medical genetics. Both PhD and MD Fellows should be "able to communicate genetic information (counselling) in such a manner that the individuals and couples are assisted in making decisions concerning themselves and reproduction" (CCMG brochure 1984:n.p.). The means to acquiring that communicative ability is expressed broadly for the PhD--"active participation in a genetic consultation and counselling service" and more specifically for the MD--"two full years of supervised experience in an active genetic consultation and counselling service"(CCMG brochure 1984:np). The assumption here is that successful communication skills involve an understanding of and sensitivity to both the medical genetic and psychosocial needs of patients. The existence of this goal for medical geneticists is the reflection of general concern for a more psychosocially oriented healthcare system and the recognition that, for patients, genetic disorders are principally an emotional and social experience rather than a medical state.

As well, there is now a master's level genetic counselling programme offered by McGill University. Included in the aims of the Diploma programme and supported by required courses is that students be competent in

> methods of interviewing and counselling and the dynamics of human behaviour in relation to genetic disease, [and in the] social, legal, and ethical issues in genetics (McGill University 1984).

B. NonMDs/nonPhDs in Canadian Genetic Services

Many genetics clinics now employ psychologists, nurses, master's level genetic associates and social workers --individuals from a paradigmatic orientation that stresses 'communication' and 'interpersonal dynamics' to work alongside the MD or PhD geneticists. As Rapp (1985:6-7) points out, many of these new entrants are trained in a Rogerian based model of counselling, a "therapeutic style which is non-interventionist, aimed at helping the patient make up her own mind." Not all nonMD/nonPhD genetic counsellors feel that they alone bring psychosocial skills to the field, but several suggested to me that their perspective "humanizes" or "balances" that provided by the MD- or PhD-geneticist. Furthermore, nonMD/nonPhD counsellors may not view themselves as medical or clinical workers and may try not to convey a medical image to the client.

A change in the style of genetic counselling interaction has involved a change in the semantics of that relationship. "Genetic counsellors" are now seeing "counsellees" or "clients". As well, genetic counsellors are expected to give priority to the agenda of concerns and topics the client holds. Hence, the sessions may range over a wide variety of topics--from inheritance patterns, to marriage problems, to feelings of gender inadequacy--each of which, ideally, is fully explored so that the participants may understand the meaning and implications of the issue for the client (Kelly 1977).

Although the importance of a psychosocial orientation is acknowledged in genetic counselling the extent to which it is put into practice is hard to judge. NonMD/nonPhD counsellors, many of whom are skilled in psychosocial matters, have tended to work alongside medical geneticists as part of a "team"--

an approach that is sanctioned by many observers of genetic counselling. The extent to which team members (medical geneticists, genetic associates,and social workers) have equally influential roles and responsibilities varies greatly. NonMD/nonPhDs have found themselves doing a great deal of routine prenatal diagnostic counselling, working as outreach genetic counsellors, and frequently as "clinic coordinators" responsible for the organizational and clerical needs of the clinic. Few do as much "counselling" as they might have expected or would like to do. Nevertheless their presence and participation in genetic clinics may have an impact on the other members of the team by encouraging MD and PhD practitioners to take psycho-social aspects into consideration.

This chapter has described the contemporary context of genetic counselling in Canada. In summary, the orientations and actions of genetic counselling professionals are influenced by both a medicoscientific paradigm and a psychosocial paradigm. The influence from medicine and science is seen primarily in the current content, language, and organization of genetic counselling practice. In addition, an educational background in medicine and science predominates among professionals in genetic counselling. An orientation to the psychosocial issues in genetic counselling is suggested by the recent entry of nonPhDs/nonMDs into the field, some of whom are trained in counselling skills.

Recall from the previous chapter that the influences of science, medicine, and psychosocial disciplines have accumulated in contemporary genetic counselling through historical process as a diversity of goals and orientations.

As structural influences they appear as two paradigms--medicoscientific and psychosocial. In reality the two paradigms are meshed through history and practice, the meeting of shifting priorities: pathological realities, client expectations, professional goals and societal restrictions. The patterned roles of professionals which reflect these influences extends the description of the contemporary context of genetic counselling into the next chapter.

CHAPTER THREE

Genetic Counsellors' Roles

In order to describe and understand the patterns of professional roles that characterize Canadian genetic counselling, this chapter reviews the results of a questionnaire received from 76 genetic counselling professionals across Canada. Interviews with 11 of those individuals are used as a further resource here. In addition to describing the participants and their educational backgrounds, their professional activities and the time spent on them are discussed. The chapter concludes with a discussion of the connections between genetic counselling roles, historical process, and their contemporary context. A necessary prelude to this chapter is a statement about the methodology and design of the questionnaire.

The Questionnaire Design

The intent of the questionnaire component of my investigation was threefold:

1) to provide a description of the socioeducational attributes and occupational activities of Canadian genetic counselling professionals;

2) to learn of the attitudes, concerns, and opinions held by these Canadian health care professionals about their work.

3) to compare these Canadian responses with similar work done in the United States.

Prior to this investigation there had been no survey of genetic counselling in Canada. (Coincident with this survey sent out in November of 1985, Canadian College of Medical Genetics members were asked to participate in an International Survey of Medical Geneticists being funded by the National Institutes of Health of the United States.) Initially, the questionnaire was to be sent only to medical geneticists and genetic associates in Ontario and was to be used as an adjunct to interviews with those individuals. However, the difficulties encountered in obtaining a sample of Ontario genetic counselling professionals who were interested in being interviewed on repeated occasions (four to five hours for each individual) made it prudent to make the questionnaire more detailed.

As a result the questionnaire was sent to the members of the Canadian College of Medical Genetics (CCMG) and to subscribers of *CrossOver: The Newsletter of Genetic Associates of Canada.* I felt that the use of these two mailing lists would provide a representative sample of those health care professionals who practise genetic counselling in connection with a specialized clinic or service. No list of personnel for individual Canadian genetics clinic was available as an alternate sampling device.

Two comments on the sampling of professionals are noted. First, although the survey reached all those medical geneticists who were members of the CCMG in Canada at the time of the survey, the means of sampling all others involved in genetic counselling was probably less comprehensive. Those social workers, genetic associates, nurses, members of the clergy, and others who do not subscribe to the newsletter *CrossOver* are under-represented. Also not included are medical doctors (pediatricians and general practitioners in particular) who provide 'informal genetic counselling' as part of their regular family

practice. Second, the study does not permit me to assess whether those individuals who responded to the questionnaire are representative of the total population of professionals providing genetic counselling. Third, the study does not include a control group of professionals outside of genetic counselling; hence it is not possible to determine whether the results are different from those generally encountered among health care professionals.

Two surveys of genetic counselling in the United States were used as the basis for my own questionnaire (especially Sorenson *et al.* 1981: 147-151; see copy of questionnaire in Appendix One). In addition to the section on socioeducational attributes (age, sex, marital status, children, religion, current occupation, years of practice, degrees held, and training in genetics and counselling), the questionnaire provided information on the tasks and activities (broken down by hours per week) of the respondent. Information was also obtained about the average number of clients counselled per week and the type of professionals involved in genetic counselling at the respondent's place of practice. In order to acquire data on the kinds of approaches to counselling, the questionnaire did include fixed alternative questions (Pelto 1970:105) on counsellors' goals, decision-making strategies, responsibilities in counselling, and preferred method of raising issues in counselling sessions. Four brief hypothetical counselling sessions were described in the questionnaire and respondents were asked to identify key issues in each and to make suggestions to the counsellor. The final section of the questionnaire provided those surveyed with an opportunity to respond to a selection of statements made by critics and observers of contemporary genetic counselling.

In anticipation of criticisms about using a questionnaire format to obtain information about individuals' attitudes and opinions, the following

special methodological considerations are noted. My primary concern here is to deal with the questions: "Why did I use the questionnaire format?" and "How was the choice justified given the types of data in which I was interested?" Several of the reasons for using the questionnaire are pragmatic considerations. First, the need for data on Canadian genetic counselling suggested the questionnaire as a means of reaching as large a number of individuals as possible. Second, the questionnaire format is familiar to the individuals I wished to sample and acceptable to them given the time constraints of their work. Third, I wanted to compare the data from my Canadian survey with American survey-acquired data.

The justifications for utilizing the questionnaire are twofold. First, the questionnaire was used primarily to provide basic information on the attributes and activities of Canadian genetic counselling professionals. Where it has been used to provide information on the attitudes of those professionals the questionnaire responses are discussed in conjunction with material from the interviews. The second point relates to the theoretical underpinnings of the research. It is my conviction that the statements of individuals made in publications (in this case in the medical literature), in questionnaires, or in interviews are no less valid for study than their observed actions. In choosing the questionnaire as a valid means of determining the behaviours of genetic counselling professionals I am in agreement with Pelto's statement that respondents' answers are

> assumed to be in some way an analog of ordinary behavior. The researcher assumes the habitual behavioral tendencies of subjects will be reflected in reactions to the situations structured by the anthropologist (1970:134).

Not only are respondents' answers an analog of their behaviour, they are also a form of that behaviour. Here I am dealing with the criticism that individuals respond to questionnaires as they know they should rather than as they do in practise--what people say they do *versus* what they actually do. But what people say they do, whether they say it in interviews, in medical publications, or in questionnaires, is still a representation and an interpretation of their knowledge and experience. What people say they do is still human action. Specifically what people say they do provides an indication of the boundaries within which they feel they should act. In this study I am particularly interested in the boundaries of "responsibility" that professionals define for their interaction with clients. These boundaries may not be fully congruent with the boundaries within which individuals always act but they still reflect their accumulated knowledge and experience. The questionnaire response is one form in which the experience of genetic counselling is articulated. Statements made in interviews are another, as are actions observed in counselling sessions, and even statements made by clients about what they think the genetic counsellor has told them. My goal is to examine the experience of genetic counselling from the practitioners' perspective. That perspective articulated to outsiders is an accessible form of individual experience and knowledge.

Practitioners of Genetic Counselling

The questionnaire was sent to 91 Canadian College of Medical Genetics members, to 49 subscribers of *CrossOver*, and to one individual who is not on either list but who does practise genetic counselling. Responses were received from 45 CCMG members and 31 nonMDs/nonPhDs for a total sample size of 76 and an overall response rate of 53.4 per cent. One factor which may

have decreased the response rate is the presence of the International Survey of Medical Geneticists (a 33 page survey requiring two days to complete that was sent to CCMG members just prior to receiving mine). In addition to the 76 usable questionnaires, eleven were returned unanswered and with explanations attached: on maternity or study leave (2), retired (1), no patient contact (1), on strike (1), no time (6). In total, 49.5 per cent (45/91) of CCMG members replied to the questionnaire and 63.3 per cent (32/50) of the nonMDs/nonPhDs did so.

Variations in provincial participation rates, compounded by disparities in the responses by type of genetic counselling professional in each province, inevitably made any comparisons between provinces problematic. Thus I have dealt with the responses on a national level except in Chapter Four where I am dealing with interviews done in Ontario alone. The comparative emphasis in this study was among professional groups. Regional variations in professional attitudes and counselling strategies would be an interesting topic to investigate particularly when adequate information about the history of genetic services in each region is available.

In deciding upon a method of sorting the responses into groups for description and comparison two alternatives were considered (see Figure 3.1):

Level I: The members of the CCMG are of three professional-educational degree backgrounds: MDs, PHDs, and MD-PhDs. In keeping with the split based on highest degree obtained, I could divide the remainder of the group, all those who have neither a medical nor a doctoral degree, into nurses (RNs), Genetic Associates (GAs), and others. Unfortunately, doing so meant a rather small size in each group and a rather cumbersome number of groups for comparison.

Level II: As I came to know the group better through questionnaires and interviews, I became aware that the sample has its own informal classification. From this emic perspective, individuals with either MDs and/or PhDs are seen as separate from individuals with neither degree. Individuals with a medical degree are considered distinct from those with PhDs alone. It also appears that MD-PhD geneticists have a strong professional identification with physicians, although this identification may vary among individuals and is probably influenced by the type of work (clinical or research) they do. Although nonMDs/nonPhDs view their work as different from that of physician- or PhD-genetic counsellors, they do not make within-group distinctions between nurses, social workers, master's level Genetic Associates, and any other nonMD/nonPhD. As I will discuss in more detail later in this chapter, this tripartite emic distinction has statistically significant correlates in terms of socio-demographics and professional activities (see Tables 3.2, 3.7, and 3.9). With the exception of the fact that the PhDs in my sample are predominately female, MDs, PhDs, and MD-PhDs are sociodemographically similar. Nurses, genetic associates, and others as a sociodemographic group are also similar. Although MD-PhDs do significantly more laboratory work than MDs, they are grouped with physicians since they have similar tasks. In contrast, PhDs differ from MDs and MD-PhDs in many activities. Among nurses, genetic associates and others, tasks are generally alike, although nurses are significantly more likely to be involved in outreach programmes. In short, individuals with medical degrees, PhD geneticists, and nonMD/nonPhD genetic counsellors tend to have different roles. Hence, Level II in Figure 3.1 splits the sample into those with MDs (MDs and MD-PhDs) and those with PhDs only, but groups the others into one category called nonMDs/nonPhDs. Based on the preceding discussion, I have

organized my discussion and analysis around Level II comparisons which best reflect the internal group boundaries expressed by the participants. In some cases it is instructive to move to Level I and look at variations among the educationally distinct groups.

One further issue of nomenclature requires clarification. There is some confusion within the field about the terms 'genetic counsellor' and 'Genetic Associate'. Genetic Associate refers to those individuals who have a specialized master's level degree in genetic counselling. It is comparable to the American master's level 'Genetic Counselor'. Medical geneticists, in particular, often call their nonphysician nonPhD co-workers "genetic associates." However, I use the term to refer only to those individuals who have the specialized degree because many nonMDs/nonPhDs indicated they prefer other titles (Hereditary Disease Nurse, Clinic Coordinator, social worker, *etc*).

In this thesis, I have used the term 'genetic counsellor' or 'genetic counselling professional' to refer to the total group--MDs/MD-PhDs, PhDs, and nonMDs/nonPhDs. Not all participants use this term (23 per cent or 17/74 of the questionnaire respondents do not describe their work as 'genetic counselling'), but I have chosen to retain the term because it has been widely used and provides continuity with earlier studies.

A. Social and Personal Attributes

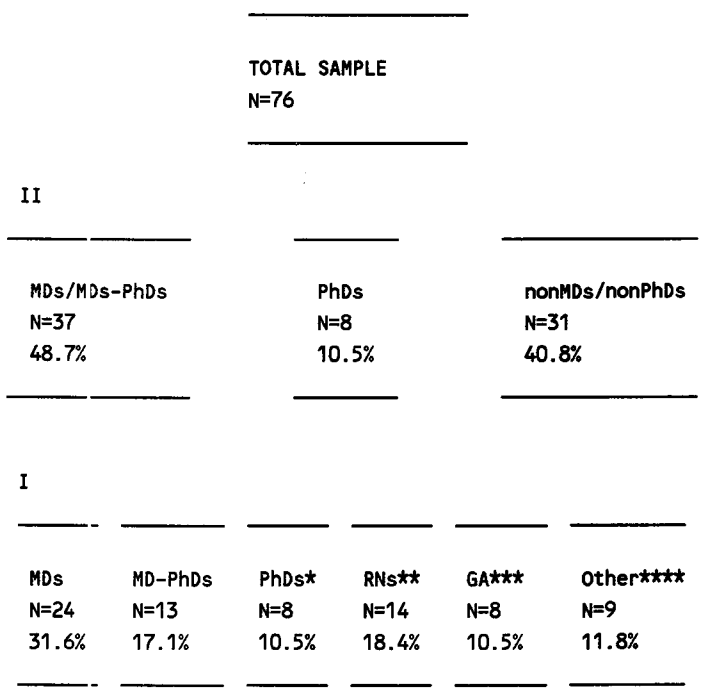
Table 3.1 (and Table 3.1a for percentages) summarizes the personal and social attributes (age, sex, marital status, presence of children, religion) for the sample of genetic counselling professionals. Most genetic counselling professionals are between 21 and 50 years of age (68.4%=52/76), female (60%=45/75), married (83.6% of 61/73) and have children (74.3%=55/74).[1] The most com-

monly selected religious affiliation was Protestant 43.4 per cent (33/76). Less than three per cent (2/76) of the sample identified themselves as Anglican, with Roman Catholicism, Judaism, atheism and 'other' comprising about equally the remaining 54 per cent (41/76). Although 60 per cent of the total group are women, 64 per cent (29/45) of those women are nurses, genetic associates and social workers. Only 32.4 per cent (12/37) of the individuals with medical degrees are women, compared to 57.1 per cent (4/7) of the PhD geneticists who responded to my questionnaire. The possibility that sampling bias resulted in the relatively high percentage of female geneticists should not be overlooked here.

Significant differences do occur between the three groups of genetic counselling professionals (see Table 3.2). MDs/MD-PhDs tend to be older ($p<0.0001$) and male ($p<0.0001$) while nonMDs/nonPhDs are younger and female.[2] PhDs are also usually older ($p<0.003$) than nonMDs/nonPhDs, but sex differences between these two were not statistically significant. Over 96.0 per cent (30/31) of the nonMDs/nonPhDs are below 50 years of age, while only 45.9 per cent (17/37) of the MDs/MD-PhDs and 62.5 per cent (5/8) of the PhDs are below 50 years of age. In fact, 67.8 per cent (21/31) of the nonMDs/nonPhDs are below 40 compared with only 21.6 per cent (8/37) of the MDs/MD-PhDs and 25 per cent (2/8) of the PhDs. Individuals with medical degrees are also significantly more likely to have children than their nonphysician/nonPhD co-workers.

FIGURE 3.1

Classification of Sample



Showing:

(a) Two Levels of Classification:
Level I - Degree based
(MDs,PHDs,MD-PhDs,RNs, GAs and Others)
Level II - Activity based and emically defined
(MDs/MD-PhDs, PhDs, and nonMDs/nonPhDs)

(b) Sample size in each category

(c) Percentage each category represents of total sample

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* Includes DSc
** Includes BScN and RN
*** Master,s level Genetic Associates
****Includes 4 BAs, 2 BSc, 2 MSW, 1 MSc in field other than genetics

TABLE 3.1

SOCIAL AND PERSONAL ATTRIBUTES

	Total Sample	MDs/ MD-PhDs	nonMDs/ nonPhDs	PhDs	MDs	MD-PhDs	RNs	GAs	Other
TOTAL NUMBER IN EACH GROUP	76	37	31	8	24	13	14	8	9
AGE RANGE									
under 20	0	0	0	0	0	0	0	0	0
21 - 30	10	0	10	0	0	0	2	5	3
31 - 40	21	8	11	2	6	2	5	3	3
41 - 50	21	9	9	3	7	2	6	0	3
51 - 60	17	14	1	2	7	7	1	0	0
over 60	7	6	0	1	4	2	0	0	0
SEX									
Male	30	25	2	3	13	12	0	1	1
Female	45	12	29	4	11	1	14	7	8
MARRIED									
Yes	61	30	23	6	20	12	12	5	6
No	12	6	7	1	3	1	1	3	3
CHILDREN									
Yes	55	36	19	6	20	10	12	2	5
No	19	7	12	1	4	2	2	6	4
RELIGION									
Protestant	33	12	18	3	9	3	10	4	4
R.Catholic	10	6	4	0	5	1	2	0	2
Jewish	8	6	2	0	4	2	0	1	1
Anglican	2	1	1	0	0	1	1	0	0
Atheist	11	5	2	4	2	3	0	2	0
Other	12	7	4	3	1	4	1	1	2

TABLE 3.1a

SOCIAL AND PERSONAL ATTRIBUTES
(percentages)*

	Total Sample	MDs/ MD-PhDs	nonMDs/ nonPhDs	PhDs	MDs	MD-PhDs	RNs	GAs	Other
TOTAL N IN EACH GROUP	75	37	31	8	24	13	14	8	9
AGE RANGE									
under 20	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0
21 - 30	13.2	0.0	32.3	0.0	0.0	0.0	14.3	62.5	33.3
31 - 40	27.6	21.6	35.5	25.0	25.0	15.4	35.7	37.5	33.3
41 - 50	27.6	24.3	29.0	37.5	29.2	15.4	42.9	0.0	33.3
51 - 60	22.4	37.8	3.2	25.0	29.2	53.8	7.1	0.0	0.0
over 60	9.2	16.2	0.0	12.5	16.7	15.4	0.0	0.0	0.0
SEX									
Male	40.0	67.6	6.5	42.9	54.2	92.3	0.0	12.5	11.1
Female	60.0	32.4	93.5	57.1	45.8	7.7	100.0	87.5	88.9
MARRIED									
Yes	83.6	88.9	76.7	85.7	87.0	92.3	92.3	62.5	66.7
No	16.4	11.1	23.3	14.3	13.0	7.7	7.7	37.5	33.3
CHILDREN									
Yes	74.3	83.3	61.3	85.7	83.3	83.3	85.7	25.0	55.6
No	25.7	16.2	38.7	14.3	16.7	16.7	14.3	75.0	44.4
RELIGION									
Protestant	43.4	32.4	58.1	37.5	37.5	23.1	71.5	50.0	44.4
R.Catholic	13.2	16.2	12.9	0.0	20.8	7.8	14.3	0.0	22.2
Jewish	10.5	16.2	6.5	0.0	16.7	15.4	0.0	12.5	11.1
Anglican	2.6	2.7	3.2	0.0	0.0	7.7	7.1	0.0	0.0
Atheist	14.5	13.5	6.5	50.0	8.3	23.1	0.0	25.0	0.0
Other	15.8	18.9	12.9	12.5	16.7	23.1	7.1	12.5	22.2

* Percentage totals may not equal 100 because of rounding.
Figures used are valid percentages, that is, the percentage of all those who answered a particular question.

TABLE 3.2

SOCIAL AND PERSONAL ATTRIBUTES,YEARS PRACTICE, AND EDUCATION: SIGNIFICANT TEST RESULTS FOR LEVEL TWO*

	MDs/MD-PHDS X PHDS	MDs/MD-PhDs X NonMDs/nonPhDs	PhDs X NonMDs/nonPhDs
Age	nss	.0001	.003
Sex	nss	.0001	nss
Married	nss	nss	nss
Children	nss	.02	nss
Religion	nss	.03	nss
YEARS PRACTICING GENETIC COUNSELLING	nss	.0001	.0001
EDUCATION:			
At least one course in human genetics	nss	nss	nss
Experience with genetics in a supervised clinic	nss	nss	nss
No training in human genetics	.002	nss	Not enough cases
At least one course in counselling	.001	.03	Not enough cases
Counselling Experience in a supervised clinic	nss	nss	nss
No training in counselling	nss	nss	nss

* Mann Whitney test results were converted to one-tailed probabilities and determined significant at $p<0.05$.

B. Practitioners' Education and Training

Information about the educational background of individuals is difficult to obtain and of restricted value given the impossibility of standardizing course or degree content. I was hesitant to compare or rank educational degrees for the same reason. The degrees held by participants were used to group individuals (see Figure 3.1, Level I) and to indicate the variety of educational backgrounds of individuals in genetic counselling. As Figure 3.1 shows, nearly one-third of this sample are MDs (31.6%=24/76) and a further 17.1 per cent of the respondents had both MD and PhD degrees. Thus almost 50 per cent of the total sample (37/76) hold medical degrees. Nearly 60 per cent of the medical degrees are in pediatrics (see Table 3.3).

TABLE 3.3

DEGREE SPECIALIZATIONS

Specialization	MD	MD-PHD	PhD
Pediatrics	18	4 -	-
Genetics	2	- 8	6
Biochemistry	-	- 2	1
Unspecified	1	9 -	-
Other	3	- 3	1
TOTAL	24	13	8

Nurses are an interesting group with respect to training. They make up a relatively large group (18.4%=14/76) in this study and bring the total percentage of individuals trained in a clinical environment to 67 per cent (51/76). I do not include them with physician geneticists, however, since their training in that environment is substantially different. The training of physician geneticists is towards the recognition and treatment of diseases, while nurses are oriented towards patient care. Furthermore, as the following section will show, the tasks of nurses in genetic counselling are quite different from those of both physician-geneticists and Phd-geneticists (see also Tables 3.8 and 3.11). And finally, several comments from nurses on the questionnaires indicate they see themselves as part of the nonMd/nonPhD group in genetic counselling.

Almost one third of the participants have a PhD (27.6%=21/76) either in combination with a medical degree (17.1%=13/76) or as their primary degree (10.5%=8/76). Two thirds of those doctoral degrees are in genetics (research area: cytogenetics, medical or biochemical genetics). A further ten percent (8/76) of the sample are Genetic Associates--individuals who have a specialized two-year Master's degree in genetic counselling.

Two questions on the survey provide some indication of the changing educational demographics of genetic counselling professionals. Participants were asked to indicate the year in which their highest degree was awarded and the number of years they had been involved in genetic counselling. Over 50 per cent of the individuals with MDs had received their degree by 1970 and almost 88 per cent had their medical degrees by 1980. A similar pattern emerges for individuals with PhDs--57.1 per cent had their degree by 1970 and 95.2 per cent by 1980. Among the nonMDs/nonPhDs, only 35.5 per cent had their degrees by 1970 and 74.2 per cent had them by 1980. But of the total sample, nearly 58 per

cent have been involved in genetic counselling for ten years or less and nearly 74 per cent for 15 years or less (Table 3.4). The difference in responses here between MDs/MD-PhDs or PhDs and nonMDs/nonPhDs is quite striking (and significant at $p<0.0001$). All nonMDs/nonPhDs in my sample have practised for 15 years or less compared with only 54 per cent (20/37) of the MDs/MD-PhDs and 62.5 per cent (5/8) of the PhDs practising for the same duration. Several interpretations are possible. First, the lower average number of years providing genetic counselling among nonMDs/nonPhDs is a reflection of their lower age distribution. Second, it reflects the historical fact that up until the early 1970s genetic counselling was provided almost exclusively by physicians and PhD researchers and often in private practice (nearly one-half of the MDs and over one-third of the PhDs indicate they have been involved in genetic counselling for over 15 years). Training programmes for genetic associates were not begun until 1971 in the USA (Rapp 1985:6) and the fall of 1985 in Canada. All Genetic Associates in my sample obtained their degrees within the last 10 years and all except one did so within the last five years. During the 1970s the increased availability of prenatal diagnosis coincided with the development of genetic counselling centers throughout Canada, and clinic co-ordinators, outreach nurses, and additional personnel stepped in to accommodate the growing demand for genetic services.

I asked respondents to indicate the type of training (course, supervised clinical training, none, or other) in human genetics and in counselling methods. Since many individuals have had more than one type of training, the figures in Table 3.5 indicate the percentage of respondents who have had, for example, at least one course in human genetics either alone or in conjunction with other training in human genetics. As a group, the majority (73.7%=56/75)

of genetic counselling professionals have had at least one course in human genetics and some clinical training in human genetics (60.5%=46/75). Only 6.6

TABLE 3.4

NUMBER OF YEARS PRACTICING GENETIC COUNSELLING

	Total Sample	MDs/ MD-PhDs	nonMDs/ nonPhDs	PhDs	MDs	MD-PhDs	RNs	GAs	Other
N	76	37	31	8	24	13	14	8	9
< one yr	2	0	2	0	0	0	2	0	0
	2.6%	0.0%	6.5%	0.0%	0.0%	0.0%	14.3%	0.0%	0.0%
1-5 yr	28	4	23	1	4	0	8	7	8
	36.8%	10.8%	74.2%	12.5%	16.7%	0.0%	57.1%	87.5%	88.9%
6-10 yr	14	7	5	2	4	3	4	1	0
	18.4%	18.9%	16.1%	25.0%	16.7%	23.1%	28.6%	12.5%	0.0%
11-15 yr	12	9	1	2	5	4	0	0	1
	15.8%	24.3%	3.2%	25.0%	20.8%	30.8%	0.0%	0.0%	11.1%
16-20 yr	12	10	0	2	6	4	0	0	0
	15.8%	27.0%	0.0%	25.0%	25.0%	30.8%	0.0%	0.0%	0.0%
21-25 yr	6	6	0	0	5	1	0	0	0
	7.9%	16.2%	0.0%	0.0%	20.8%	7.7%	0.0%	0.0%	0.0%
> 25 yr	2	1	0	1	0	1	0	0	0
	2.6%	2.7%	0.0%	12.5%	0.0%	7.7%	0.0%	0.0%	0.0%

per cent (5/76) of the group had no training in human genetics at all. As might be expected significantly more MDs/MD-PhDs than PhDs had no formal training in genetics ($p<0.002$), but there were not enough cases to test this variable between PhDs and nonMDs/nonPhDs. Other differences among MDs, PhDs, and nonMDs/nonPhDs in the type or occurrence of human genetics training were small and not statistically significant (see Tables 3.2 and 3.5).

TABLE 3.5

TRAINING IN HUMAN GENETICS AND COUNSELLING

		Total Group	MD/ MD-PhDs	PhDs	NonMDs/ nonPhDs
GENETICS	At least one course	56/75 73.7%	29/37 80.5%	6/8 85.0%	21/31 67.8%
	Experience in supervised clinic	46/75 60.5%	23/37 62.4%	5/8 62.5%	18/31 58.1%
	None	5/75 6.7%	4/37 11.1%	0/8 0.0%	1/31 3.2%
	Other	7/75 9.3%	1/37 2.8%	2/8 25.0%	4/31 12.9%
COUNSELLING	At least one course	26/75 34.2%	5/37 13.9%	0/8 0.0%	21/31 67.8%
	Experience in supervised clinic	40/75 48.6%	21/37 56.8%	4/8 50.0%	15/31 48.4%
	None	17/75 22.4%	10/37 27.8%	3/8 37.5%	4/31 12.9%
	Other	8/75 10.7%	5/37 13.9%	1/8 12.5%	2/31 6.5%

Overall, 34.2 per cent (26/75) of genetic counselling professionals indicated they had had at least one course in counselling methods and 48.6 per cent (40/75) had been trained in a supervised clinic (Table 3.5). Within the total group, however, only 13.9 per cent (5/37) of the MDs compared with 67.8 per cent (21/31) of nonMDs/nonPhDs had at least one course in counselling: a significant difference. None of the PhDs in my sample had taken courses in counselling and this lack proved to be signficant when compared with both

other groups. Over 20 per cent (17/75) of the total sample said they had no training in counselling methods. Of the MDs/MD-PhDs, 27.8 per cent (10/37) said they had no training in counselling and 37.5 per cent (3/8) of the PhDs compared with 12.9 per cent (4/31) of the nonMDs/nonPhDs gave the same response. Differences among the three groups were not statistically significant (see Table 3.2).

Although my questionnaire did not ask detailed questions on education, recall from the earlier discussion (Chapter Two) that the overall amount of course time alloted to human genetics in Canadian and American medical schools, if it is offered at all, is low (Swanson 1984:39). PhD geneticists undoubtedly have the most coursework in genetics. Graduates of specialized genetic counselling master's level programmes have "University level courses in Principles of Human Genetics and take further courses, labs, and clinics in genetics" (McGill University 1984).

Time and time again, reviews of medical school curricula have pointed to the lack of formal training in interpersonal skills (Dodge 1983) or skills in communication (Sanson-Fisher and Maguire 1980). These skills are certainly not taught in PhD programmes in genetics. The nonMDs and nonPhDs in Canada, however, are often exposed to counselling methods in their training as nurses, social workers and genetic associates (McGill University 1984). It is important to note, however, that skill in counselling was most frequently reported by all three groups as gained in the clinical context.

One aspect of a professional's preparation for genetic counselling that is not ascertained by the questionnaire is that of experience. The educational contribution of regular and repeated exposure to the wide spectrum of genetic problems, client questions and reactions, and day-to-day activity in

providing genetic counselling cannot be adequately measured by the questionnaire. The importance of experience, as it was impressed upon me in interviews, is discussed later in this chapter and in Chapter Four.

Judgements about the relative merit of each group's preparation for genetic counselling are hazardous. Given the wide variety of training backgrounds and the variety of activities done it is extremely difficult to assess with any degree of objectivity how well prepared an individual is to provide genetic counselling. Rather than attempt such assessments I asked individuals in interviews how well they felt their education and training had prepared them for their present work and I reviewed the questionnaires for statements about training for genetic counselling. It is to these two sources of information that I now turn.

Several remarks on the training of genetic counselling professionals appeared in response to the 'Cases' and 'Comments' sections of the questionnaire. Two themes were evident. First, although it was felt that professionals providing genetic counselling today are much better trained than in the past, there is still "much to learn," particularly in the area of meeting the psychosocial needs of clients. One individual commented that "few training centers actually provide instruction in techniques of counselling (even though they think they do)". A second theme was evident among genetic counsellors who place a high value on genetics in medical practice and are worried about the slight attention it receives in medical school. The inadequate genetics training in medical school was particularly worrisome to those respondents who advocate that general practitioners should be assuming more of the responsibility for "run of the mill" or "routine" genetics cases (such as advanced maternal age, or Down syndrome).

Both of these concerns also arose in the interviews I did with medical geneticists. Although they were optimistic that the training of genetic counsellors would continue to improve, the medical geneticists complained that a primary source of current problems in the discipline lay in the medical school curricula. Two issues were identified by respondents--genetics is taught in such a way that its utility is masked, and no effort is put into teaching young doctors how to communicate pertinent genetic information to their patients. One physician geneticist described the problem as a "town *versus* gown fight". In other words, medical school curricula are controlled by academics ("gown", PhDs in particular) who emphasize "gene probes, molecular genetics, and other material they consider to be really hot right now" despite the protests of physicians who want to put "genetics on the ward" ("town").

While I deal in more detail with the issue of "Who should be providing genetic counselling?" later in this chapter, it is worthwhile noting some of the sentiments expressed in interviews about the variety of training and educational backgrounds found in the field. The interview material comes from conversations with five medical geneticists (three MDs and two MD/PhDs) and six nonMDs/nonPhDs (two individuals with Master's of Social Work and four with bachelor degrees in the social sciences).

A common reaction to my question " Did your training prepare you for the sorts of things you are encountering in your work?" was an emphatic "no!" In discussing the matter further, individuals described the genetics component of their work (diagnosis, calculating and conveying recurrence risks, and treatment) as "straightforward" or "the easiest part of counselling". The "professionally challenging part" of their work is felt to be in coping with the range of attitudes and reactions clients bring to the session. In general, expertise and

confidence in interacting with clients is described as the product of experience. Not surprisingly, physicians and those with medical training tended to emphasize the value of that training as a necessary stepping stone to gaining communication skills through experience. In response to my question, one medical geneticist replied at first that his training had been "completely inadequate." He then went on to say:

> I was not prepared for all the questions that patients want answered. The only part of my training that I think prepared me was the fact that when I did my pediatrics I had to go out and practice pediatrics in small communities and learn how practices work with families. If I had stayed in the university hospital I was in I wouldn't have any idea how the world functioned. My PhD prepared me for nothing. You do most of the hot shot tech stuff, but in terms of relating with people my university training was completely inadequate and the only thing that prepared me was the fact that I did a fair amount of non-major hospital-based practice. And I sort of picked up, I think, how to relate to people most of the time.

Given the recent entry of non-physicians and non-PhD researchers into genetics, many of those now practising have little or no formal training in human genetics or counselling for genetic disorders. While this lack of formal training is not true of master's level Genetic Associates it is true for the nonMD/nonPhDs I interviewed. The nonMD/nonPhDs I interviewed emphasized the benefits of the "social work" or "psychosocial" perspective they bring to their job, rather than the genetics component. One woman described her job this way:

> I don't know that we always need to know it [the genetics in that detail]. We've got access to specialists. [What genetics do you need to know?] The very basic genetics stuff, so that you can understand what they're talking about. But you also have to have a good grasp of how families work and what their needs are.

A common theme among the nonMDs/nonPhDs is that the specialized skills (*i.e.*, the knowledge of genetics) can be picked up on the job or left to the

medical geneticists. The desired sensitivity and ability to communicate with people (counselling skills) are not peculiar to genetic counselling but are felt to have more to do with "knowing how to work with families".

Several of the medical geneticists felt the social workers, nurses and Genetic Associates who work with them are probably better at understanding and "getting involved with the patients" because of their training. However most study participants, including the nonphysicians and nonPhDs, maintain that the nonMDs/nonPHDs should not assume all the responsibility for genetic counselling since they lack the expertise in genetics and in medicine. Interestingly, those master's level Genetic Associates who are trained in both genetics and counselling but are doing clerical work in genetics clinics are felt by medical geneticists to be "overqualified" and "probably unhappy" with their circumscribed role. The nonMDs and nonPhDs I interviewed did not express these sentiments, but it is unlikely they would have done so in only the one interview. (This issue is discussed later in the chapter.)

While nonMDs and nonPhDs are felt to be well-suited to share some of the patient care with physicians, individuals who have only a PhD are considered ill-prepared by some of their colleagues to deal with patients. Although the laboratory competence and research contributions of the PhD geneticist are not disputed, the emphasis their training places on the genetics is felt by others to handicap them.

> I think the science gets in the way for the PhD. They have two mistresses, one is science and the other is science [laughter]. I mean they're constantly hung up on the scientific aspect. If it's not scientific . . . it is not qvaluable if you can't prove it. Dammit all! A lot of the stuff we do is not valid by that description. If I had to justify everything I do on the basis of these data bases I'd go crazy (an MD geneticist).

> I used to think I could do clinical genetics with a PhD. After I went and did my medical training I realized I didn't know what I was talking about. I was wrong (an MD-PhD geneticist).

Although I do not have interview data from PhD geneticists, their comments on the questionnaires suggest they are concerned about the lack of genetic knowledge among their colleagues. Not surprisingly, they do not share the sentiment of some of their co-workers that "the science gets in the way." Indeed, the diagnosis made by their physician colleagues, especially in the case of non-Mendelian or cytogenetic disorders, may depend on the identification of the genetic disorder by the PhD geneticist. Furthermore the contribution of the PhD geneticist is not just case-related but also concerns research that increases the repertoire of known genetic anomalies.

In sum, the majority of individuals providing genetic counselling have been trained in a medical paradigm and clinical environment. The recent entry of non-physicians and non-PhD geneticists into the field has strengthened the ties of genetic counselling to a psycho-social paradigm and introduced the concern that practitioners have communication and interpersonal skills--counselling skills. Training in human genetics is likely to be from coursework and experience in a supervised clinic. There was considerable concern expressed by individuals from all degree categories about the fact that the clinical application of genetics is not enhanced in medical school curricula. In comparison to genetics, which is considered the "easy part" of their work, counselling is regarded as much more challenging. The training of nonMDs and non-PhDs includes counselling methods more often than does the the training of physicians and PhD geneticists, but nearly one-quarter of the total group indicate they have had no training in counselling. Experience in counselling and clinical contact with patients are valued as the best ways to acquire communica-

tion and interpersonal skills. MDs/MD-PhDs and nonMDs/nonPhDs interviewed felt their training was an asset for the work they were now doing, but both of these groups were in agreement that PhD training was a disadvantage for genetic counselling. The paradigmatic orientation of this doctoral training, perceived as one toward "numbers" and "high tech research", is felt to prevent PhD geneticists from developing the skills in interacting with clients that MDs and nonMDs/nonPhDs value. The perceived translation of the PhD researcher's scientific objectivity in the lab into impersonality and insensitivity in dealing with clients works with the persistent association between PhD geneticists and eugenics to contribute to occupational competition in genetic counselling.

From the previous discussion of emically-derived information on the educational and training background among genetic counselling professionals, a separation of roles within the field begins to appear. The variety of educational and work experiences is large, although all individuals are involved in "genetic counselling". Further insight into these patterns of roles and responsibilities subsumed under the label of "genetic counselling" is available from questionnaire data on tasks and from interview materials.

C. Tasks and Time

Respondents were asked to indicate the number of hours they spend on a particular activity (seeing patients, lab and research, teaching, clerical and administrative etc) in an average week. The results demonstrate considerable variation in both total number of hours involved in genetic counselling and in the breakdown of hours for each type of activity.

The total number of hours involved in all genetic counselling related activities (seeing clients, clerical and/or administrative duties, laboratory work,

meetings, and other) varies a great deal from individual to individual (see Table 3.6). Of the 70 respondents who indicated their total hours, the range was from on to 75 hours and the mean was 25.8+/-17.6 hours per week. The least hours spent in genetic counselling related activities are represented by PhDs (mean of 7.3+/-5.9 hours per week, N=8), the most by Genetic Associates (mean of 36.3+/-14.1 hours per week, N=8) respectively. The mode for all professional groups combined was 15 hours per week. There were statistically significant differences among the genetic counselling professionals in terms of the total number of hours per week spent in all genetic counselling related activities (see Table 3.7). The amount of time spent by PhD geneticists in all counselling related activities is significantly smaller than that for all those individuals with a medical degree ($p<0.001$) and nonMDs/nonPhDs ($p<0.0001$). Differences between MDs/MD-PhDs and nonMDs/nonPhDs were not statistically significant.

Comparing the number of clients seen by MDs or by PhDs to non-MDs/nonPhDs or among any of the degree-based groups of Level I does not show statistical significance (see Table 3.7 and 3.10). Of the 70 respondents who indicated they have regular client contact, the group mean is 5.0+/-4.3 (N=70); MDs see the most clients per week (6.0+/-4.1, N=21) and PhDs see the fewest (3.3+/-2.4, N=6). Whereas the total sample (N=70) spends about five hours each week seeing clients, PhDs spend an average of one hour per week seeing clients, MDs spend about five and one-half hours, and Genetic Associates indicate they spend nearly seven hours a week seeing clients. These differences are significant between PhDs and MDs ($p<0.0007$) and between PhDs and GAs ($p<0.0007$) although not between MDs and GAs. Looking at the number of clients seen per week and the amount of time spent with them it would seem

MDs spend about one hour with each case, PhDs less than an hour, and Genetic Associates slightly more than one hour.

Although my questionnaire did not detect statistically significant differences in the number of clients seen by the MDs/MD-PhDS, PhDs, and non-MDs/nonPhDs, it is clear from interviews and comments on the questionnaire that differences in terms of the *type* of contact with clients do exist. "Seeing patients" for nonMDs/nonPhDs is often limited to administrative or clerical "intake" duties, pre-test information sessions, collecting pedigrees, and obtaining blood samples. MDs on the other hand describe their interaction with patients in terms of detailed "genetic counselling" sessions involving a a wide range of topics--collecting information, making a diagnosis, providing an estimate of reproductive risks, recommending treatment where applicable, and outlining client reproductive options. Unfortunately, I do not have interview-based data to comment on the notion of "seeing patients" held by PhD geneticists.

As might be expected, MDs/MD-PhDs and PhDs each spend significantly more hours each week involved in research than do other professionals (see Tables 3.7 and 3.10). The reverse is true of clerical and administrative activities--nonMDs/nonPhDs spend an average of 10.8+/-9.9 hours per week doing these tasks compared with 6.2+/-5.9 hours per week indicated by those with MDs and 0.5+/-0.5 by PhDs. Looking at the breakdown of hours more closely (see Table 3.6) it is clear that PhDs and MD-PhDs are doing significantly more research than any of the other groups. Nurses and "Others" appear to be doing the majority of the clerical and administrative work while PhDs, in particular, do significantly less.

TABLE 3.6

MEAN NUMBER OF CLIENTS AND HOURS PER WEEK BY GROUP

	Total Sample	MDs/MD-PhDs	nonMDs/nonPhDs	PhDs	MDs	MD-PhDs	RNs	GAs	Other
N=	70	33	31	6	21	12	14	8	9
Number of Clients	5.0+/-4.3	5.5+/-4.3	4.9+/-4.5	3.3+/-2.4	6.0+/-4.1	4.5+/-4.7	4.7+/-4.2	5.8+/-5.3	4.4+/-4.9
Total Hours in Genetic Counselling	25.8+/-17.6	28.7+/-20.3	27.5+/-13.1	5.3+/-5.9	28.3+/-15.7	29.4+/-28.3	22.5+/-8.3	36.3+/-14.1	27.0+/-15.1
Hours Seeing Patients	5.2+/-5.3	6.2+/-5.2	5.2+/-5.5	1.0+/-1.4	5.6+/-4.2	6.3+/-7.1	5.9+/-5.1	6.8+/-7.4	2.8+/-3.7
Clerical & Admin. Hours	7.5+/-8.2	6.2+/-5.9	10/8+/-9.9	0.5+/-0.5	5.4+/-5.0	6.8+/-7.4	10.2+/-6.1	7.1+/-5.9	15.0+/-15.3
Lab Hours	3.4+/-8.9	6.3+/-12.2	0.1+/-0.4	3.6+/-5.7	6.3+/-13.0	5.6+/-9.9	0.8+/-0.3	0.0+/-0.0	0.2+/-0.0
Meeting Hours	1.7+/-1.7	1.7+/-1.5	1.6+/-1.9	1.6+/-1.6	1.9+/-1.5	1.1+/-1.6	0.5+/-1.2	3.0+/-2.4	2.0+/-1.6
Total Non Counselling Hours**	21.9+/-17.9	29.6+/-17.4	8.7+/-9.1	39.9+/-16.7	24.9+/-17.0	37.4+/-16.7	9.1+/-5.4	4.6+/-4.4	11.9+/-14.5
Research Hours	9.7+/-13.3	11.8+/-14.6	3.3+/-6.7	25.5+/-12.3	7.3+/-11.0	18.8+/-16.8	2.8+/-2.7	3.1+/-3.4	4.2+/-11.9
Teaching Hours	3.0+/-3.7	4.4+/-4.1	0.9+/-1.6	5.6+/-3.9	4.0+/-3.3	4.3+/-5.5	1.2+/-1.7	0.3+/-0.5	1.1+/-2.0

* Includes the number of hours in activities (seeing patients, clerical and administrative duties, laboratory work, meeting with counselling staff, and other) related to genetic counselling.

** Includes the number of hours in activities (research, practise, teaching, administration, and other) not related to genetic counselling.

TABLE 3.7

MEAN NUMBER OF CLIENTS AND HOURS PER WEEK: SIGNIFICANT TEST RESULTS FOR LEVEL TWO*

	MDs/MD-PHDS X PHDS	MDs/MD-PhDs X NonMDs/nonPhDs	PhDs X NonMDs/nonPhDs
CLIENTS PER WEEK	nss	nss	nss
HOURS: Total Hours in Genetic Counselling	.001	nss	.0001
Hours Seeing Patients	.001	nss	.004
Clerical & Admin Hours	.002	.02	.0001
Lab Hours	nss	.001	.02
Meeting Hours	nss	nss	nss
Total Non-Counselling Hours	nss	.0001	.0001
Research Hours	.006	.0005	.0001
Teaching Hours	nss	.0001	.0004

* Mann Whitney test results were converted to one-tailed probabilities and determined significant at $p<0.05$.

MDs/MD-PhDs indicated they spend an average of 29.6+/-17.4 hours per week in research, clinical practice, teaching and administrative activities not related to client contact. Individuals with PhDs spend nearly 40 hours a week at these sorts of activities. Genetic Associates, RNs, Social Workers and other nonMD/nonPhDs on the other hand, spend much less time (8.7+/-9.1 hours/week) at activities outside of counselling: significantly less ($p<0.0001$) than MD/MD-PhDs and PhDs.

Participants were asked to indicate their regular activities from a number of administrative, counselling, diagnostic, and patient management tasks. There is a fair amount of uniformity within the group for counselling-related tasks such as amniocentesis counselling, counselling for emotional problems and financial difficulties and genetic counselling (see Table 3.8). This uniformity was not supported in interviews when tasks and responsibilities of clinic members was discussed. In my opinion, the questionnaire did not detect differences in counselling-related tasks for two reasons. First, the terms used, "amniocentesis counselling" or "genetic counselling" are too broad to be useful. Second, failure to indicate these tasks as part of their routine might suggest the respondent's unwillingness to 'counsel' and to deal with clients' psychosocial aspects--an attitude that is clearly contradictory to the expressed goals of genetic counselling.

In terms of counselling for family problems, MDs/MD-PhDs are significantly more likely to indicate they do this in comparison with both PhDs and nonMDs/nonPhDs (see Table 3.9). NonMDs/nonPhDs are more likely to indicate follow-up to counselling as part of their regular routine in comparison to the other two groups. While all nonMDs/nonPhDs obtain a family social history from their clients, in comparison, only 72.2 per cent (26/26) of individuals

with MDs and 37.5 per cent (3/8) of the PhDs do so (both significant at $p<0.01$). Each of the administrative tasks--scheduling appointments, obtaining medical records, assigning patients to counsellors, and coordinating clinic activities--is more often performed by a nonMD/nonPhD (all differences here are significant at $p<0.01$).

Of the diagnosis-related activities, nonMDs/nonPhDs do not do physical examinations or make diagnoses although most of the MDs/MD-PhDs do ($p<0.0001$). PhDs do not do physical examinations and 62.5 per cent (5/8) do not make diagnoses. Both PhDs and nonMDs/nonPhDs are less likely than individuals with medical degrees to take a medical history from their clients. None of this sample perform amniocentesis--a task that is usually left to registered technologists and obstetricians. Taking pedigrees (the schematic representations of families used in genetics to chart the occurrence of a genetic disorders) and medical histories, and referring for diagnostic tests are part of the regular routines for most genetic counselling professionals.

Patient management tasks such as providing medical treatment and information for longterm management and referring to specialists are done primarily by MDs. Other management tasks such as referring clients to support groups or outside agencies are most often done by MDs and by non-MDs/nonPhDs.

TABLE 3.8

TASKS BY GROUP

		Total Sample	MDs/ MD-PhDs	nonMDs/ nonPhDs	PhDs	MDs	MD-PhDs	RNs	GAs	Other
N =		75	37	31	8	23	13	14	8	9
ADMINISTRATIVE										
Scheduling appointments	No	65.3%(49)	91.7%(33)	29.0% (9)	87.5% (7)	91.3%(21)	92.3%(12)	21.4% (3)	50.0% (4)	22.2% (2)
	Yes	34.7%(26)	8.3% (3)	71.0%(22)	12.5% (1)	8.7% (2)	7.7% (1)	78.6%(11)	50.0% (4)	77.8% (7)
Obtaining records	No	46.7%(35)	75.0%(27)	6.5% (2)	75.0% (6)	78.3%(18)	69.2% (9)	0.0% (0)	12.5% (1)	11.1% (1)
	Yes	53.3%(40)	25.0% (9)	93.5%(29)	25.0% (2)	21.7% (5)	30.8% (4)	100.0%(14)	87.5% (7)	88.9% (8)
Assigning clients to counsellors	No	72.0%(54)	86.1%(31)	51.6%(16)	87.5% (7)	82.6%(19)	92.3%(12)	42.9% (6)	37.5% (3)	77.8% (7)
	Yes	28.0%(21)	13.9% (5)	48.4%(15)	12.5% (1)	17.4% (4)	7.7% (1)	57.1% (8)	62.5% (5)	22.2% (2)
Coordinate clinic	No	57.3%(43)	69.4%(25)	35.5%(11)	87.5% (7)	69.6%(16)	69.2% (9)	35.7% (5)	37.5% (3)	33.3% (3)
	Yes	42.7%(32)	30.6%(11)	64.5%(20)	12.5% (1)	30.4% (7)	30.8% (4)	64.3% (9)	62.5% (5)	66.7% (6)
COUNSELLING										
Obtain family social history	No	20.0%(15)	27.8%(10)	0.0% (0)	62.5% (5)	26.1% (6)	30.8% (4)	0.0% (0)	0.0% (0)	0.0% (0)
	Yes	80.0%(60)	72.2%(26)	100.0%(31)	37.5% (3)	73.9%(17)	69.2% (9)	100.0%(14)	100.0% (8)	100.0% (9)
Genetic counselling	No	20.0%(15)	8.3% (3)	29.0% (9)	37.5% (3)	8.7% (2)	7.7% (1)	28.6% (4)	12.5% (1)	44.4% (4)
	Yes	80.0%(60)	91.75(33)	71.0%(22)	62.5% (5)	91.3%(21)	92.3%(12)	71.4%(10)	87.5% (7)	55.6% (5)
Amniocentesis counselling	No	32.0%(24)	33.3%(12)	25.8% (8)	50.0% (4)	34.8% (8)	30.8% (4)	14.3% (2)	25.0% (2)	44.4% (4)
	Yes	68.0%(51)	66.7%(24)	74.2%(23)	50.0% (4)	65.2%(15)	69.2% (9)	85.7%(12)	75.0% (6)	55.6% (5)
Counselling for family problems	No	58.7%(44)	37.8%(14)	71.0%(22)	100.0% (8)	43.5%(10)	30.8% (4)	71.4%(10)	62.5% (5)	77.8% (7)
	Yes	41.3%(31)	59.5%(22)	29.0% (9)	0.0% (0)	56.5%(13)	69.2% (9)	28.6% (4)	37.5% (3)	22.2% (2)
Counselling for emotional problems	No	58.7%(44)	45.9%(17)	64.5%(20)	87.5% (7)	52.2%(12)	38.5% (5)	57.1% (8)	62.5% (5)	77.8% (7)
	Yes	41.3%(31)	51.4%(19)	35.5%(11)	12.5% (1)	47.8%(11)	61.5% (8)	42.9% (6)	37.5% (3)	22.2% (2)
Suggest resources for financial problems	No	62.7%(47)	61.1%(22)	54.8%(17)	100.0% (8)	65.2%(15)	53.8% (7)	28.6% (4)	75.0% (6)	77.8% (7)
	Yes	37.3%(28)	38.9%(14)	45.2%(14)	0.0% (0)	34.8% (8)	46.2% (6)	71.4%(10)	25.0% (2)	22.2% (2)
Refer to counselling specialists	No	37.3%(28)	27.8%(10)	38.7%(12)	75.0% (6)	21.7% (5)	38.5% (5)	35.7% (5)	37.5% (3)	44.4% (4)
	Yes	62.7%(47)	72.2%(26)	61.3%(19)	25.0% (2)	78.3%(18)	61.5% (8)	64.3% (9)	62.5% (5)	55.6% (5)
Follow-up to counselling	No	29.3%(22)	41.7%(15)	6.5% (2)	62.5% (5)	34.8% (8)	53.8% (7)	7.1% (1)	0.0% (0)	11.1% (1)
	Yes	70.7%(53)	58.3%(21)	93.5%(29)	37.5% (3)	65.2%(15)	46.2% (6)	92.9%(13)	100.0% (8)	88.9% (8)
DIAGNOSTIC										
Take pedigree	No	14.7%(11)	19.4% (7)	3.2% (1)	37.5% (3)	21.7% (5)	15.4% (2)	0.0% (0)	0.0% (0)	11.1% (1)
	Yes	85.3%(64)	80.6%(29)	96.8%(30)	62/5% (5)	78.3%(18)	84.6%(11)	100.0%(14)	100.0% (8)	89.9% (8)
Take medical history	No	25.3%(19)	11.1% (4)	32.3%(10)	62.5% (5)	8.7% (2)	15.4% (2)	21.4% (3)	50.0% (4)	33.3% (3)
	Yes	74.7%(56)	88.9%(32)	67.7%(21)	37.5% (3)	91.3%(21)	84.6%(11)	78.6%(11)	50.0% (4)	66.7% (6)
Do physical examination	No	58.7%(44)	13.9% (5)	100.0%(31)	100.0% (8)	13.0% (3)	15.4% (2)	100.0%(14)	100.0% (8)	100.0% (9)
	Yes	41.3%(31)	86.1%(31)	0.0% (0)	0.0% (0)	87.0%(20)	84.6%(11)	0.0% (0)	0.0% (0)	0.0% (0)
Refer for diagnostic tests	No	30.7%(23)	13.9% (5)	48.4%(15)	37.5% (3)	8.7% (2)	23.1% (3)	50.0% (7)	25.0% (2)	66.7% (6)
	Yes	69.3%(52)	86.1%(31)	51.6%(16)	62.5% (5)	91.3%(21)	76.9%(10)	50.0% (7)	75.0% (6)	33.3% (3)
Laboratory work	No	64.0%(48)	47.2%(17)	90.3%(28)	37.5% (3)	60.9%(14)	23.1% (3)	92.9%(13)	100.0% (8)	77.8% (7)
	Yes	36.0%(27)	52.8%(19)	9.7% (3)	62.5% (5)	39.1% (9)	76.9%(10)	7.1% (1)	0.0% (0)	22.2% (2)
Make diagnosis	No	53.3%(40)	11.1% (4)	100.0%(31)	62.5% (5)	8.7% (2)	15.4% (2)	100.0%(14)	100.0% (8)	100.0% (9)
	Yes	46.7%(35)	88.9%(32)	0.0% (0)	37.5% (3)	91.3%(21)	84.6%(11)	0.0% (0)	0.0% (0)	0.0% (0)
Perform amniocentesis	No	100.0%(75)	100.0%(36)	100.0%(31)	100.0% (8)	100.0%(23)	100.0%(13)	100.0%(14)	100.0% (8)	100.0% (9)
	Yes	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)	0.0%(0)	0.0% (0)	0.0% (0)	0.0% (0)	0.0% (0)
Participate in genetic screening	No	62.7%(47)	61.1%(22)	64.5%(20)	62.5% (5)	65.2%(15)	53.8% (7)	50.0% (7)	87.5% (7)	66.7% (6)
	Yes	37.3%(28)	38.9%(14)	35.5%(11)	37.5% (3)	34.8% (3)	46.2% (6)	50.0% (7)	12.5% (1)	33.3 (3)
PATIENT MANAGEMENT										
Provide medical treatment	No	76.0%(57)	50.0%(18)	100.0%(31)	100.0% (8)	52.2%(12)	46.2% (6)	100.0%(14)	100.0% (8)	100.0% (9)
	Yes	24.0%(18)	50.0%(18)	0.0% (0)	0.0% (0)	47.8%(11)	53.8% (7)	0.0% (0)	0.0% (0)	0.0% (0)
Provide information for longterm management	No	44.0%(33)	19.4% (7)	64.5%(20)	75.0% (6)	8.7% (2)	38.5% (5)	64.3% (9)	62.5% (5)	66.7% (6)
	Yes	56.0%(42)	80.6%(29)	35.5%(11)	25.0% (2)	91.3%(21)	61.5% (8)	35.7% (5)	37.5% (3)	33.3% (3)
Refer to parents group	No	25.3%(19)	22.2% (8)	19.4% (6)	62.5% (5)	13.0% (3)	38.5% (5)	7.1% (1)	25.0% (2)	33.3% (3)
	Yes	74.7%(56)	77.8%(28)	80.6%(25)	37.5% (3)	87.0%(20)	61.5% (8)	92.9%(13)	75.0% (6)	66.7% (6)
Refer to social or educational agency	No	26.7%(20)	16.7% (6)	25.8% (8)	75.0% (6)	13.0% (3)	23.1% (3)	14.3% (2)	37.5% (3)	33.3% (3)
	Yes	73.7%(55)	83.3%(30)	74.2%(23)	25.0% (2)	87.0%(20)	76.9%(10)	85.7%(12)	62.5% (5)	66.7% (6)
Refer to specialists	No	30.7%(23)	11.1% (4)	45.2%(14)	62.5% (5)	8.7% (2)	15.4% (2)	50.0% (7)	37.5% (3)	44.4% (4)
	Yes	69.3%(52)	88.9%(32)	54.8%(17)	37.5% (3)	91.3%(21)	84.6%(11)	50.0% (7)	62.5% (5)	55.6% (5)
Management follow-up	No	26.7%(20)	25.0% (9)	19.4% (6)	62.5% (5)	21.7% (5)	30.8% (4)	7.1% (1)	37.5% (3)	22.2% (2)
	Yes	73.3%(55)	75.0%(27)	80.6%(25)	37.5% (3)	78.3%(18)	69.2% (9)	92.9%(13)	62.5% (5)	77.8% (7)
OTHER										
Participate in outreach programme	No	44.0%(33)	44.4%(16)	38.7%(12)	62.5% (5)	34.8% (8)	61.5% (8)	0.0% (0)	87.5% (7)	55.6% (5)
	Yes	56.0%(42)	55.6%(20)	61.3%(19)	37.5% (3)	65.2%(15)	38.5% (5)	100.0%(13)	12.5% (1)	44.4% (4)
Provide education for professionals	No	17.3%(13)	11.1% (4)	29.0% (9)	0.0% (0)	8.7% (2)	15.4% (2)	14.3% (2)	37.5% (3)	44.4% (4)
	Yes	82.7%(62)	88.9%(32)	71.0%(22)	100.0% (8)	91.3%(21)	84.6%(11)	85.7%(12)	62.5% (5)	55.6% (5)
Provide education for the public	No	18.7%(14)	22.2% (8)	12.9% (4)	25.0% (2)	21.7% (5)	23.1% (3)	7.1% (1)	0.0% (0)	33.3% (3)
	Yes	81.3%(61)	77.8%(28)	87.1%(27)	75.0% (6)	78.3%(18)	76.9%(10)	92.9%(13)	100.0% (8)	66.7% (6)
Participate in parent/patient self-help group	No	50.7%(38)	41.7%(15)	58.1%(18)	62.5% (5)	30.4% (7)	61.5% (8)	42.9% (6)	62.5% (5)	77.8% (7)
	Yes	49.3%(37)	58.3%(21)	41.9%(13)	37.5% (3)	69.6%(16)	38.5% (5)	57.1% (8)	37.5% (3)	22.2% (2)

TABLE 3.9

TASKS: SIGNIFICANT TEST RESULTS FOR LEVEL TWO*

	MDs/MD-PHDS X PHDS	MDs/MD-PhDs X NonMDs/nonPhDs	PhDs X NonMDs/nonPhDs
ADMINISTRATIVE			
Scheduling Appointments	nss	.0001	.004
Obtaining Records	nss	.0001	.001
Assigning clients to counsellors	nss	.001	nss
Coordinate clinic	nss	.003	.01
COUNSELLING			
Obtain a family social history	nss	.008	.002
Genetic counselling	nss	.01	nss
Amniocentesis Counselling	nss	nss	nss
Counselling for family problems	.003	.004	nss
Counselling for emotional problems	.04	nss	nss
Suggest resources for financial problems	.05	nss	.03
Refer to counselling specialists	.02	nss	nss
Follow-up to counselling	nss	.0005	.006
DIAGNOSTIC			
Take pedigree	nss	.02	nss
Take medical history	.01	.01	nss
Do physical examination	.0001	.0001	nss

TABLE 3.9 continued

	MDs/MD-PHDS X PHDS	MDs/MD-PhDs X NonMDs/nonPhDs	PhDs X NonMDs/nonPhDs
Refer for diagnostic tests	nss	.001	nss
Laboratory work	nss	.0001	.01
Make diagnosis	.01	.0001	nss
Perform amniocentesis	nss	nss	nss
Participate in genetic screening	nss	nss	nss
PATIENT MANAGEMENT: Provide medical treatment	.01	.0001	nss
Provide information for long-term management	.006	.0001	nss
Refer to parents group	.04	nss	.03
Refer to social or educational agency	.004	nss	.02
Refer to specialists	.01	.0009	nss
Management follow-up	nss	nss	.03
OTHER: Participate in outreach programme	nss	nss	nss
Provide education for professionals	nss	.03	nss
Provide education for the public	nss	nss	nss
Participate in parent/patient self-help group	nss	nss	nss

*Mann-Whitney test results were converted to one-tailed probabilities and determined significant at $p<0.05$.

Activities like genetic screening, outreach clinics, education, promotion, and participation in genetic support or self-help groups were also investigated. Overall, 37.3 per cent (28/75) of the sample indicated they participated in genetic screening programmes. There are no significant differences between groups in this respect. Participation in outreach programmes which bring specialized genetic services to smaller communities is part of the regular routine for 56 per cent (42/75) of the respondents. All the nurses (N=14) and 65.2 per cent (15/23) of the MDs compared with 37.5 per cent (3/8) of the PhDs, 38.8 per cent of the MDs-PhDs, and 12.5 per cent (1/8) of the genetic associates participate in outreach programmes (see Table 3.11).

Providing education for other health care professionals is part of the occupational routine for most genetic counselling professionals as is public education. Although nonMDs and nonPhDs indicated they are involved in educational activities, between-group variation exists in the average reported number of hours teaching (Table 3.6). Whereas MDs/MD-PhDs teach an average of 4.4+/-4.1 hours each week and PhDs 5.6+/-3.9 hours, nonMDs and nonPhDs spend less than one hour each week (0.9+/-1.6 hours/week) teaching ($p<0.0005$). Both MDs/MD-PhDs and PhDs report involvement in providing education for professionals more often than nonMDs/nonPhDs. The questionnaire did not ask respondents to specify where they are teaching, but interview materials suggests physicians and in particular, PhDs, are often teaching university level courses. Genetics associates and nurses are more likely to do lectures (once or twice a month) to public health nurses, high schools, parents' groups, *etc.*

From this discussion of activities a clearer picture of the pattern of roles that exists among genetic counsellors emerges. There are a number of routine or standard tasks, such as taking pedigrees or medical histories, that are

performed by most genetic counselling professionals. In terms of their tasks, non physicians/nonPhDs as a group are quite homogeneous in on respect: they assume the bulk of the responsibility for the day-to-day running of the genetics clinic. In general, however, the role of the nonphysician/nonPhD is quite varied. NonMD/nonPhD involvement in aspects of genetic counselling other than clerical and administrative ones seems to vary from clinic to clinic. In many places nonphysicians/nonPhDs are doing only clinic-oriented activities such as clerical and administrative work. Contact with clients by non-MDs/nonPhDs often takes the form of counselling and follow-up to counselling by telephone and letter. When nonMDs/nonPhDs do see clients, possibly in sessions prior to prenatal diagnostic testing, they are not involved in treatment- or diagnosis-related activities. The total number of hours nonphysicians and non-PhDs indicate they spend in genetic counselling is not significantly different from that indicated by MDs/MD-PhDs. Part of the reason for this is that although nonMDs/nonPhDs are often part time workers at the clinic, all their activities are directed towards "genetic counselling". MDs/MD-PhDs (and PhDs) on the other hand, may be providing genetic counselling in addition to working in other areas of medical genetics. Although there are very clear and statistically significant differences between individuals with MDs and non-MDs/nonPhDs in virtually all types of tasks, they do see about the same number of clients each week and indicated total number of hours in genetic counselling are similar. I have argued that nonMDs/nonPhDs and MDs/MD-PhDs see clients for different reasons, but both use "genetic counselling" to describe their involvement.

TABLE 3.10

SOCIODEMOGRAPHICS, YEARS IN PRACTISE , NUMBER OF CLIENTS, HOURS: SIGNIFICANT TEST RESULTS FOR LEVEL ONE

VARIABLE	MD x PhD	MD x MD-PhD	MD x RN	MD x GA	MD x OTHER	PhD x MD-PhD	PhD x RN	PhD x GA	PhD x OTHER	MD-PhD x RN	MD-PhD x GA	MD-PhD x OTHER	RN x GA	RN x OTHER	GA x OTHER
SOCIO-DEMOGRAPHICS															
Age	nss	nss	0.007	0.0001	0.002	nss	nss	0.0005	0.01	nss	nss	nss	nss	nss	nss
Sex	nss	0.03	0.003	0.05	0.03	0.04	nss	nss	nss	0.0001	0.0006	0.0004	nss	nss	nss
Marriage	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss
Children	nss	nss	nss	0.007	nss	nss	nss	0.03	nss	nss	0.02	nss	0.009	nss	nss
Religion	nss	nss	nss	nss	nss	nss	nss	nss	nss	0.01	nss	nss	nss	nss	nss
YEARS PRACTISE IN GENETIC COUNSELLING	nss	nss	0.0001	0.0002	0.0002	nss	0.0008	0.002	0.003	0.0001	0.0001	0.0001	nss	nss	nss
NUMBER OF CLIENTS	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss
HOURS															
Total hours in Genetic Counselling	0.0001	nss	nss	nss	nss	nss	0.0001	0.0001	0.003	nss	nss	nss	0.02	nss	nss
Hours seeing patients	0.0007	nss	nss	nss	0.02	0.02	0.0009	0.007	nss	nss	nss	nss	nss	nss	nss
Clerical & Administration	0.001	nss	0.008	nss	nss	nss	0.0001	0.0003	0.001	nss	nss	nss	nss	nss	nss
Laboratory Hours	nss	nss	nss	nss	nss	nss	0.04	nss	nss	nss	nss	nss	nss	nss	nss
Meeting Hours	nss	nss	0.0006	nss	nss	nss	nss	nss	nss	nss	nss	nss	0.01	0.01	nss
Total Non-Counselling Hours	0.02	0.03	0.001	0.003	0.01	nss	0.0001	0.0001	0.0003	0.0001	0.0001	0.001	nss	nss	nss
Research Hours	0.0005	0.01	nss	nss	0.005	nss	0.0001	0.0003	0.002	0.001	0.005	0.0006	nss	nss	nss
Teaching Hours	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss

TABLE 3.11

TASKS: SIGNIFICANT TEST RESULTS FOR LEVEL ONE

VARIABLE	MD x PhD	MD x MD-PhD	MD x RN	MD x GA	MD x OTHER	PhD x MD-PhD	PhD x RN	PhD x GA	PhD x OTHER	MD-PhD x RN	MD-PhD x GA	MD-PhD x OTHER	RN x GA	RN x OTHER	GA x OTHER
ADMINISTRATIVE															
Scheduling Appointments	nss	nss	0.0001	0.03	0.0009	nss	0.005	nss	0.01	0.0005	nss	0.002	nss	nss	nss
Obtain records	nss	nss	0.0001	0.002	0.001	nss	0.001	0.02	0.01	0.0009	0.01	0.01	nss	nss	nss
Assign clients to counsellors	nss	nss	0.03	0.03	nss	nss	nss	nss	nss	0.02	0.01	nss	nss	nss	nss
Coordinate clinic	nss	nss	0.04	nss	nss	nss	0.03	nss	nss	nss	nss	nss	nss	nss	nss
COUNSELLING															
Obtain a family social history	nss	nss	nss	nss	nss	nss	0.008	0.02	0.01	nss	nss	nss	nss	nss	nss
Genetic counselling	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss
Amnio-centesis counselling	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss
Counsel-ling for family problems	0.009	nss	nss	nss	nss	0.004	nss	nss	nss	0.04	nss	0.04	nss	nss	nss
Counselling for emotional problems	nss	nss	nss	nss	nss	0.03	nss	nss	nss	nss	nss	nss	nss	nss	nss
Suggest resources financial problems	nss	nss	0.03	nss	nss	0.05	0.003	nss	nss	nss	nss	nss	0.04	0.03	nss
Refer to counselling specialists	0.01	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss
Follow-up to counselling	nss	nss	nss	nss	nss	nss	0.02	0.02	0.04	0.02	0.02	0.05	nss	nss	nss
DIAGNOSTIC															
Take pedigree	nss	nss	nss	nss	nss	nss	nss	nss	nss	ns	nss	nss	nss	nss	nss
Take medical history	0.01	nss	nss	0.03	nss	0.04	nss	nss	nss	nss	nss	nss	nss	nss	nss

TABLE 3.11 (continued)

VARIABLE	MD x PhD	MD x MD-PhD	MD x RN	MD x GA	MD x OTHER	PhD x MD-PhD	PhD x RN	PhD x GA	PhD x OTHER	MD-PhD x RN	MD-PhD x GA	MD-PhD x OTHER	RN x GA	RN x OTHER	GA x OTHER
DIAGNOSTIC															
Do physical examination	0.0001	nss	0.0001	0.0001	0.0001	0.0003	nss	nss	nss	0.0001	0.0003	0.0002	nss	nss	nss
Refer for diagnostic test	nss	nss	0.02	nss	0.005	nss	nss	nss	nss	nss	nss	0.05	nss	nss	nss
Laboratory tests	nss	0.03	nss	nss	nss	nss	0.02	0.02	nss	0.006	0.001	0.02	nss	nss	nss
Make diagnosis	0.01	nss	0.0001	0.0001	0.0001	0.04	nss	nss	nss	0.0001	0.0003	0.0002	nss	nss	nss
Perform amniocentesis	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss
Participate in genetic screening	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss
PATIENT MANAGEMENT															
Provide medical treatment	0.02	nss	0.008	0.02	0.02	0.02	nss	nss	nss	0.008	0.02	0.02	nss	nss	nss
Provide information for long-term management	0.002	nss	0.002	0.01	0.005	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss
Refer to parents' group	0.02	nss	nss	nss	nss	nss	0.02	nss	nss	nss	nss	nss	nss	nss	nss
Refer to social or educational agency	0.004	nss	nss	nss	nss	0.03	0.01	nss	nss	nss	nss	nss	nss	nss	nss
Refer to specialists	0.01	nss	0.02	nss	nss	0.04	nss	nss	nss	nss	nss	nss	nss	nss	nss
Management follow-up	0.05	nss	nss	nss	nss	nss	0.02	nss	nss	nss	nss	nss	nss	nss	nss
OTHER															
Participate in outreach programme	nss	nss	0.04	0.01	nss	nss	0.008	nss	nss	0.003	nss	nss	0.0001	0.01	nss
Provide education for professionals	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss
Provide education for the public	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss
Participate in parent/ patient self-help group	nss	nss	nss	nss	0.02	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss

There is a clear division between the types of activities done by MDs/MD-PhDs and by PhDs. Physicians see the most clients per week and do almost all of the diagnosis-, treatment-, and management-related tasks. In addition to their medical tasks, MDs do indicate they provide "counselling". Physicians are frequently involved in research, teaching, and outside practise.

Genetic counselling professionals who hold only a PhD are clearly research-oriented, although they do provide case-related expertise. They have the fewest number of clients per week and are more likely than the other professionals to be involved in non-medical or non-counselling related tasks. A primary orientation of PhD geneticists is in research and laboratories.

Who should be providing genetic counselling?

The previous discussion of roles in genetic counselling has been based primarily on questionnaire- and interview-based information about education and activities. Additional information comes from the comments made by genetic counselling professionals about their roles and from an examination of the links between these roles and the context in which they exist.

The question of who should be providing genetic counselling has been widely discussed in the literature. The predominate opinion found in these writings is that genetic counselling should be provided by a team of experts.

> The personnel of the group should include an M.D. trained in genetics to take responsibility for the medical acts performed by the group and, depending on the size of the group, a number of others with M.D. or Ph.D. degrees, or both, and training in the techniques of genetic counseling. There must be a cytogeneticist available. A variety of auxiliary personnel such as public health nurses, social workers, or genetic associates can provided valuable information in interviewing, searching files and literature sources, collating information, and following up families (Fraser 1974:652).

The team approach is presented as an excellent way to provide the multiple components of genetic care--continuing research, accurate medical genetic information, attention to the clients' pyschosocial needs, and efficiently run clinics. In general, the team approach does exist in Canadian genetic counselling. However, a team of individuals involved in the care of each case may not always be feasible, especially at smaller centers with limited resources and personnel. As well, genetic issues for clients are not always easily partitioned for each expert to deal with separate issues. Although genetic counselling professionals may speak of their work as composed of two parts--the genetics and the counselling, in practise the two are intertwined. Most importantly from this discussion of roles it will be seen that the "team" is really composed of the physician geneticist involved in client contact, the PhD researcher as a behind-the-scenes expert in genetics, and nonMDs/nonPhDs fulfilling a variety of jobs, primarily clinic-oriented and clerical and administrative support for physician geneticists.

Among the individuals I spoke with, the question of who should be providing genetic counselling concerned two issues: the roles of physicians and non-physicians and the involvement of referring physicians in genetic care. The first issue, as it was explained to me by a medical geneticist, is essentially a question of to what extent genetic counselling belongs within the practise of medicine. Raising the question implies that the needs of people facing genetic disorders, particularly their emotional and psychosocial needs, might be better served by professionals whose fields of orientation are nonmedical, for example, psychology or social work. However, when I included the comment "Genetic counselling should not be within the practise of medicine" on the questionnaire, the reaction was overwhelmingly a negative one. For the entire sample, 91.5 per

cent (65/71) disagreed with this statement. Similarly high percentages of MDs/MD-PhDs (94.5%=34/37), PhDs (66.7%=4/6) and nonMD/nonPhDs (93.1%=27/29) did the same (the differences are not statistically significant). The comments which accompanied these responses repeatedly stressed that the medical content and information, particularly the diagnosis and treatment, were integral to good genetic counselling and a major influence on people's response to coping with genetic disorders. Several people expressed shock at the statement and wondered what the alternative would be. Clearly the sentiment is to continue to provide genetic counselling within the practise and paradigm of medicine. Furthermore, there is support among those sampled for physician-geneticists to continue to be the primary caregivers in genetic counselling. (Although I do not have interview data with PhD geneticists, their questionnaire responses do support this view.)

This study did find widespread support for a medically-oriented genetic counselling but the roles of non-physicians remain at issue. Specifically, the non-physicians around whom the debate centers are the nurses, social workers and master's level genetic associates and other nonMD/nonPhDs collectively known as "genetic associates". One geneticist described this issue as "the million dollar question" and another described it as "a source of never-ending debate".

At one level, the role of the nonMD/nonPhD is ill-defined because they do not share the same depth of historical involvement in genetic counselling as do physicians and PhD geneticists. Recall from Chapter One that genetic counselling has strong historical ties to clinical medicine and to the science of human genetics. Although physician geneticists have in some sense usurped the clinical role of PhD-geneticists, PhD-geneticists have found a well-supported

role as researchers and scientific advisors. For nonMD/nonPhDs, on the other hand, the ties of genetic counselling to their disciplines of orientation such as psychology and social work are much more recent and less well established. Indeed nonMDs/nonPhDs did not enter genetic counselling until it had become firmly "medicalized" (Kenen 1984). As the discussion of educational backgrounds and division of activities made clear, the implications of this late entry into the field has meant that nonMDs/nonPhDs although they may be trained in the client-oriented skills of nursing and social work, spend most of their time doing clerical tasks. The one area where some nonMDs/nonPhDs have established a nonclerical role in genetic counselling is that of prenatal diagnosis, specifically amniocentesis or advanced maternal age counselling. The entry of nonMDs/nonPhDs into amniocentesis counselling has occurred as an increased demand for the test has encouraged its "routinization" (Kenen 1984;Rapp 1985). The more routine a test or procedure the more likely a non-physician is to be doing it.

> Yeah, that's [amniocentesis counselling] a nice little package of the whole genetic-services deal that is a contained area. There are a few danger signals where the genetics associate can readily see where they can turn for help. And the ground rules are fairly straightforward [MD/PhD geneticist].

> It [the entry of genetic associates into amniocentesis counselling] is probably the result of two things. First the amniocentesis load has grown and people need somebody to talk to. Secondly, most of it is of a fairly routine nature and there are a lot of similarities between cases. . . . It seemed like an area which the doctors, the medical geneticists who were basically pediatricians, didn't need to get involved with. There was no reason for them to run up there and tell people the same thing that we could tell them. Unless there is some very unusual medical condition involved, and most of the amnios are done for the same basic reason [advanced maternal age], it's easy to learn the ramifications of that and how to communicate them [Clinic coordinator].

But even in amniocentesis counselling, the role of the nonMD/nonPhD is often limited to providing pre-procedure information, accompanying the woman during the test, and notifying clients of the test results only if no abnormalities are found. Involvement in further discussions with couples whose test results are "positive" is circumscribed by the feeling that this task is best left to the physician-geneticist. Among individuals who strongly advocate this position, the importance of having a physician involved as the primary care giver in genetics was even described as a matter of rights and legalities. Several people felt "a patient has a right to see an MD," and one physician-geneticist said "I take legal responsibility for what I say, [but] the nurses [who also participate in client care] do not." Importantly, several non-physicians reported they would not feel comfortable bearing responsibility for the diagnosis and treatment related aspects of genetic care or for genetic management in general. Indeed, for them to do so would be unethical and technically illegal since legal control over the diagnosis and treatment of disease is held by physicians.

Kenen (1984) writes that nonMD/nonPhD genetic counsellors in the United States have established a counselling-oriented role for themselves, involved in direct client care, primarily by lobbying for professional recognition. Although the field of genetic counselling in the United States has been "medicalized" and "the permanent linkage of genetics and medicine" achieved, nonphysician genetic counsellors have successfully gained professional recognition and autonomy (Kenen 1984:543). Recognition has been possible by establishing both a professional organization (the National Society of Genetic Counselors) and certification examinations through the American Board of Medical Genetics (Kenen 1984:547). Although there is an association of genetic

associates in at least one Canadian province it does not have much political power or influence among MDs and PhDs geneticists who dominate the field. There are no nationally recognized certification procedures for nonMD/nonPhD genetic counsellors in Canada. Recently the Canadian College of Medical Geneticists asked the Ontario Association of Genetic Associates to prepare a position paper on their desired affiliation with the CCMG. In comparison to their colleagues in the United States, Canadian nonMD/nonPhDs do not have a formally recognized or well-defined role in the client care aspect of genetic counselling.

Part of the reason genetic associates, trained for involvement in client care, do not fulfill that role is related to historical process and circumscribed occupational power. At another level the question of roles touches at the heart of concerns about how best to provide comprehensive genetic care, that is, care that addresses both the medical/genetic and psychosocial needs of people facing genetic problems. Geneticists may acknowledge that the genetics of a case is often relatively straightforward and could be done by a nonMD/nonPhD. Furthermore, they may feel that the communication skills of the genetic associates, social workers and nurses they work with are superior to their own. But nonMD/nonPhDs do not have a substantial role in the client care aspect of genetic counselling. It is widely agreed that, in general, the needs of people facing genetic problems are best met by physician geneticists who are trained in the necessary clinical skills and who acquire counselling skills through experience.

A paradox exists here. On the one hand, although the importance of the contribution to coordinating clinic activities was not overlooked by MD geneticists, several were concerned that nonMDs/nonPhDs in genetic counselling

were "overqualified" for a clerical role. Furthermore, nonMDs/nonPhDs in genetic counselling are perceived by many of their physician colleagues to be without a well-defined role. On the other hand, on an individual basis, non-MDs/nonPhDs in this study do not appear to feel they lack a well-defined role. The comments of nonMDs/nonPhDs in interviews and on questionnaires did not express dissatisfaction with their work. In fact, many of them supported the role of the physician as primary care-giver in all aspects of genetic counselling. Why then was the issue of roles an important one among the physician geneticists I interviewed?

First, the concern of MD geneticists may reflect an awareness of the expanded role of American Genetic Counselors rather than a real threat to the position of professional dominance now held by physicians in Canadian genetic counselling. A second consideration, and one that must be acknowledged, is that this study did not include indepth interviews with a large number of nonMD/nonPhDs. If Canadian nonMD/nonPhD genetic counselling professionals are dissatisfied with their role, they did not express it in the questionnaires or interviews I did, nor do they express it in publications as do their American counterparts. In my opinion, a third factor is involved here. The "medicalization" of genetic counselling has not ceased in deference to a more "psychosocially-oriented" paradigm. Although the entry of nonMD/nonPhDs into genetic counselling may be one factor that has drawn attention to the emotional needs of clients, responsibility for those needs are now subsumed within a medical paradigm. The psychosocial aspects of genetic disease are not thought of as a separate field of expertise requiring the attention of nonMDs/nonPhDs. The physician geneticist is expected to provide comprehensive genetic care--to diagnosis genetic disease, to provide up-to-date genetic information, to discuss

options for treatment and/or management, and to be responsive to the whole spectrum of psychosocial needs that may accompany genetic disease. In principle, genetic care is the responsibility of team members whose roles are defined by the separate interests of science, medicine, and psychosocial issues. In practice, genetic care is a medical process.

Further support for the continued medicalization of genetic counselling concerns the role of the referring physician. In the literature advocating a team approach to genetic counselling, it is a physician-geneticist or a non-physician such as a Genetic Associate or social worker who is best prepared to deal over the long term with the psychosocial component of client care. But among those individuals that I interviewed, it was the referring physician--ideally the family physician who should fulfill this role. In general, the geneticists interviewed described themselves as "consultants" bringing specialized diagnostic and treatment related skills to a case, but preferring to leave both the psychosocial and longterm management to the referring doctor. One physician geneticist suggested this consultative arrangement was best because "it maintains his [the referring physician's] primacy in the relationship with the patient". The importance of leaving intact this relationship between family doctor and patient is predicated on the belief that family physicians have established a good communicative relationship with their patients. Most of the people I interviewed felt confident in leaving the responsibility for in-depth counselling with the family physician. Several, however, did express concern about the competence of general practitioners in both genetics and in communication skills.

> The family physician may be the one that does the best counselling of the lot. He may not have the expertise to know what the issues are, in other words what the diagnosis is, or the risk figures, or the options available, but once he does know that, if he is a traditional family doctor, he knows how best by far to do the real counselling. [MD/PhD geneticist]

The previous sections described a sample of the professionals providing counselling for genetic disorders in Canada and discussed the kinds of activities they are involved in. Genetic counsellors describe their work as composed of two interrelated parts--the genetics and the counselling. The fact that the genetics is considered the easy part and the counselling more difficult both reflects their preparation for genetic counselling and influences their ideas about role division. Current emphasis is on providing both medically and psychosocially oriented genetic services within a medical context (see Chapter Two). The desire to deal with both the medical and psychosocial aspects of genetic care has brought with it a debate of who is best suited to provide such care. This issue is perceived primarily as being an issue of the roles of physician versus nonphysician geneticists.

The division of roles among genetic counselling professionals that emerges from discussions of their educational preparation and an analysis of their activities is one that is clearly linked to the historical development of the field and to the contemporary structure of genetic counselling in Canada. The professional dominance of MD geneticists is directly tied to both a long and publicly supported connection between medicine and genetics and to the current context of genetic counselling. The transformation of human genetics from a discipline of scientific interest alone to a subject of clinical interest with a medically oriented content, setting, and training both produces and supports a central role for physician geneticists. With little disagreement, professionals at all levels of genetic counselling support the central role of the physician geneticist.

PhD geneticists, although they have a longer history of involvement in genetics, are doing little genetic counselling. In the opinion of many of their physician and nonMD/nonPhD colleagues, the research oriented activities and training of the PhDs hinder their ability to develop good communication skills with clients. But the strong association in Canadian society between the potential of science and the improvements in human health care have helped PhD geneticists to weather the medicalization of genetic counselling and to cultivate an accepted and essential role in research.

Nonphysician/nonPhDs who often have a background in communication skills and who have entered genetic counselling relatively recently are felt to be capable of some patient care. They do not, however, assume much responsibility for client care in genetic counselling. NonMDs/nonPhDs are doing the bulk of the clerical work associated with genetic services. Additional responsibility and further involvement in client contact appears to be made on a clinic to clinic basis. The counselling or psychosocial orientation nonMDs/nonPhDs may bring to the field is not seen as an area of expertise separate from the current medical structure and context. Rather the counselling component of genetic care is subsumed within the medical paradigm such that physician geneticists and referring family doctors are perceived as the professionals best able to deal with their clients psychosocial needs.

This chapter has investigated the roles which characterize this sample of 76 Canadian genetic counselling professionals. From a variety of personal and educational backgrounds, genetic counselling professionals divide themselves into three groups: those with MDs, those with PhDs only, and nonMDs/nonPhDs. The nature of each group's roles was determined from questionnaire- and interview-based data about genetic counsellors' educational

preparation and daily activities. The differing roles of physician geneticists, PhD geneticists, and those with other degrees separate the professional community along lines that reflect the historical basis and the contemporary structure of genetic counselling described in Chapters One and Two. The central role in genetic counselling is that of the physician geneticist. The PhD-geneticist has a clearly defined role, outside of client care, in research. The nonMD/nonPhDs, the most recent entrants to genetic counselling, have as yet a rather ambiguous role and status.

In the next chapter I turn from the context, the underlying paradigms and roles of genetic counselling, to the individual genetic counsellor. I examine the meaning of genetic counselling as it was voiced to me through the goals and agendas genetic counsellors set for themselves and their notions of 'responsibility' in meeting those agendas.

CHAPTER NOTES

1 All percentages used are "valid percentages"; that is, the percentage of all those who answered a particular question (Norusis 1983: 21)

2 Results from Mann-Whitney paired comparison tests were converted to one-tailed probabilities and determined significant at $p<0.05$.

CHAPTER FOUR

Individual Boundaries of Responsibility

Thus far I have described the history and contemporary context of genetic counselling in Canada, focussing on the paradigms which underlie that context and the roles which have arisen through historical process. Context, roles, and as will be discussed in this chapter, agendas for counselling all provide the genetic counselling professional with a sense of his or her boundaries of responsibility in interacting with clients. Four cases are employed in this chapter to illustrate the individualized nature of these boundaries.

Responsibility is a concept that is woven into everything genetic counselling professionals do--their research and their interaction with colleagues, with clients, and with other healthcare professionals. The issue of responsibility in genetic counselling was suggested to me by several medical geneticists as an area demanding investigation; the issue is also implicit in much of the literature on genetic counselling. While I would hesitate to call it an emic concept, responsibility is used here as it has been defined and described by genetic counselling professionals and as it appears implicitly in their statements. Responsibility includes and is shaped by professionally sanctioned notions of ethics and generally held ideas about morals. Responsibility, as I will show, is also tied to personal feelings about client needs and counsellor limitations. The following two sections discuss the goals and protocols of genetic counselling in order to outline what genetic counselling professionals view as important in

their interaction with clients and to convey a sense of the meaning genetic counselling has for these individuals. From this, an understanding of individually-constructed notions of responsibility and approaches to genetic counselling is possible.

Goals

In the following section I briefly describe the results of the questionnaire sections that asked respondents to indicate the importance of four goals of genetic counselling. These findings are compared to an attitudinal survey of American genetic counsellors referred to in Chapter Two (Sorenson *et al.* 1981). Following this comparison, the problems of asking questions about individually-held goals of genetic counselling are discussed.

For the group as a whole, "providing medical and genetic information" and "reducing patient anxiety" were of primary importance (see Table 4.1). "Preventing disease or abnormality" was also of importance, but reducing "the number of carriers of genetic disorders in the population" was of less or no importance. Comparison with the study by Sorenson and colleagues (1981) shows that the response patterns for the first three goals were similar for the American group.

The published reports from the American study do not indicate whether between-group differences in goals were investigated. The findings from my study showed minimal and non-statistically significant between-group differences in the goals to provide medical and genetic information, reduce patient anxiety, or prevent disease or abnormality. However, for 58.3 per cent (21/36 of MD/MD-PhDs and 75 per cent (6/8) of those with PhDs only, the goal

TABLE 4.1

GOALS AND DECISION-MAKING STRATEGIES

	Total	MD/MD-PhD	NonMD/NonPhD	PhD	MD	MD-PhD	RN	GA	OTHER
N =	76	37	31	8	24	13	14	8	9
GOALS:									
The prevention of disease or abnormality									
High importance	40.8%(31)	43.2%(16)	41.9%(13)	25.0%(2)	37.5%(9)	53.8%(7)	50.0%(7)	12.5%(1)	55.6%(5)
Moderate importance	53.9%(41)	51.3%(19)	54.8%(17)	62.5%(5)	58.3%(14)	38.5%(5)	50.0%(7)	75.0%(6)	44.4%(4)
Low importance	5.3%(4)	5.4%(2)	3.2%(1)	12.5%(1)	4.2%(1)	7.7%(1)	0.0%(0)	12.5%(1)	0.0%(0)
The removal or lessening of patient anxiety									
High importance	85.5%(65)	89.2%(33)	83.9%(26)	75/0%(6)	91.7%(22)	84.6%(11)	92.9%(13)	87.5%(7)	66.7%(6)
Moderate importance	14.5%(11)	10.8%(4)	16.1%(5)	25.0%(2)	8.3%(2)	15.4%(2)	7.1%(1)	12.5%(1)	33.3%(3)
Low importance	0.0%(0)	0.0%(0)	0.0%(0)	0.0%(0)	0.0%(0)	0.0%(0)	0.0%(0)	0.0%(0)	0.0%(0)
A reduction in the number of carriers of genetic disorders in the population									
High importance	13.3%(10)	5.6%(2)	22.6%(7)	12.5%(1)	4.3%(1)	7.7%(1)	42.9%(6)	0.0%(0)	11.1%(1)
Moderate importance	40.0%(30)	36.1%(13)	51.6%(16)	12.5%(1)	30.4%(7)	46.2%(6)	57.1%(8)	37.5%(3)	55.6%(5)
Low importance	46.7%(35)	58.3%(21)	25.8%(8)	75.0%(6)	65.2%(15)	46.2%(6)	0.0%(0)	62.5%(5)	33.3%(3)
Provide clients with medical and genetic information									
High importance	98.7%(75)	97.3%(36)	100.0%(31)	100.0%(8)	100.0%(24)	92.3%(12)	100.0%(14)	100.0%(8)	100.0%(9)
Moderate importance	1.3%(1)	2.7%(1)	0.0%(0)	0.0%(0)	0.0%(0)	7.7%(1)	0.0%(0)	0.0%(0)	0.0%(0)
Low importance	0.0%(0)	0.0%(0)	0.0%(0)	0.0%(0)	0.0%(0)	0.0%(0)	0.0%(0)	0.0%(0)	0.0%(0)
DECISION-MAKING STRATEGIES:									
Tell clients that decisions are theirs alone and refuse to make any decisions for them									
Appropriate	90.5%(67)	83.3%(30)	96.8%(30)	100.0%(8)	91.3%(21)	69.2%(9)	92.9%(13)	100.0%(8)	100.0%(9)
Inappropriate	9.5%(7)	16.7%(6)	3.2%(1)	0.0%(0)	8.7%(2)	30.8%(4)	7.1%(1)	0.0%(0)	0.0%(0)
You will support any decisions that clients make									
Appropriate	97.4%(74)	94.6%(35)	100.0%(31)	100.0%(8)	95.8%(23)	92.3%(12)	100.0%(14)	100.0%(8)	100.0%(9)
Inappropriate	2.6%(2)	5.4%(2)	0.0%(0)	0.0%(0)	4.2%(1)	7.7%(1)	0.0%(0)	0.0%(0)	0.0%(0)
Inform clients what other people in their situation have done									
Appropriate	66.7%(50)	88.9%(32)	45.2%(14)	50.0%(4)	87.0%(20)	92.3%(12)	28.6%(4)	50.0%(4)	66.7%(6)
Inappropriate	33.3%(25)	11.1%(4)	54.8%(17)	50.0%(4)	13.0%(3)	7.7%(1)	71.4%(10)	50.0%(4)	33.3%(3)
Inform clients what you would do if you were in their situation									
Appropriate	24.0%(18)	38.9%(14)	6.5%(2)	25.0%(2)	30.4%(7)	53.8%(7)	7.1%(1)	0.0%(0)	11.1%(1)
Inappropriate	76.0%(57)	61.1%(22)	93.5%(29)	75.0%(6)	69.6%(16)	46.2%(6)	92.9%(13)	100.0%(8)	88.9%(8)
Advise clients what they ought to do									
Appropriate	8.2%(6)	14.3%(5)	3.3%(1)	0.0%(0)	17.4%(4)	8.3%(1)	7.7%(1)	0.0%(0)	0.0%(0)
Inappropriate	91.8%(67)	85.7%(30)	96.7%(29)	100.0%(8)	82.6%(19)	91.7%(11)	92.3%(12)	100.0%(8)	100.0%(9)

to "reduce the number of carriers" was of low importance. Only 25.8 per cent(8/31) of the remaining professionals held the same opinion. The differences here between MD/MD-PhDs or PhDs and nonMD/nonPhDs are statistically significant (see Table 4.2). Interestingly, this goal was of moderate-to-high importance among the RNs, but only of low-to-moderate importance for each of the other groups. In comparing RNs to each of the other groups (MDs, PhDs, MD-PhDs, GAs, and Others) the significance levels for this difference range from $p<0.05$ to $p<0.0001$ (see Table 4.3).

Recall that the literature has characterized PhDs, MDs, and nonMD/nonPhD genetic counsellors as having different priorities: PhDs are characterized as oriented towards the prevention of genetic disease, MDs toward educating patients about genetic disease, and nonMDs/nonPhDs toward the psychosocial attributes of genetic disease. Hence, one might expect to see an association between PhDs and the goal of reducing the number of carriers in the population, MDs and the provision of medical and genetic information, and non-MDs/nonPhDs and reducing patient anxiety. A clear triadic distinction in goals such as those listed was not detected by my survey. This lack of degree-based differences may have been due to the small size of the sample (too small to pick up the differences). It might also be the nature of responses to questionnaires; that is, respondents are aware of societal expectations that they be non

-interventionist and non-directive, and hence, have answered the questions accordingly. This second problem, however, applies to both my study and the American one. It is my opinion that the degree-based distinctions in priorities specified in the literature simply do not reflect reality. As the above results suggest, an association between certain goals and educational degrees is not supported in the questionnaires. Also, I did not encounter it in the interviews.

Once I had done the interviews, the ineffectiveness of my question, "What are your goals in genetic counselling?" became very apparent. I asked the question because I wanted to know what priorities were held by individuals providing genetic counselling. Questions about goals did not lead to answers about priorities but to answers about problems in putting those priorities into practice.

The standard answer to my queries about an individual's goals was "to provide accurate information in as objective a manner as possible." But my question was met several times by a laugh and a shrug ("I don't think any of us know what the goals [of genetic counselling] are"). Frequently, the respondent would have a clear statement of what goals he or she did not endorse ("I'm certainly not trying to reduce the load of genetic disease"; "it isn't to stamp out genetic disease"). The more fruitful approach to understanding what priorities genetic counsellors hold was simply to talk about *what* they were doing and *how* they went about doing it.

TABLE 4.2

GOALS AND DECISION-MAKING STRATEGIES:
SIGNIFICANT TEST RESULTS FOR LEVEL TWO

	MD/MD-PhD X PhD	MD/MD-PhD X NonMD/NonPhD	PhD X NonMD/NonPhD
GOALS:			
The prevention of disease or abnormality	nss	nss	nss
The removal or lessening of patient anxiety	nss	nss	nss
A reduction in the number of carriers of genetic disorders in the population	nss	.002	.02
Provide clients with medical and genetic information	nss	nss	nss
DECISION-MAKING STRATEGIES:			
Tell clients that decisions are theirs alone and refuse to make any decisions for them	nss	.04	nss
You will support any decisions that clients make	nss	nss	nss
Inform clients what most other people in their situation have done	.045	.0001	nss
Inform clients what you would do if you were in their situation	nss	.001	nss
Advise clients what they ought to do	nss	nss	nss

* Mann-Whitney test results were converted to one-tailed probabilities and determined significant at $p<0.05$.

TABLE 4.3

GOALS AND DECISION-MAKING STRATEGIES: SIGNIFICANT TEST RESULTS FOR LEVEL ONE

VARIABLE	MD x PhD	MD x MD-PhD	MD x RN	MD x GA	MD x OTHER	PhD x MD-PhD	PhD x RN	PhD x GA	PhD x OTHER	MD-PhD x RN	MD-PhD x GA	MD-PhD x OTHER	RN x GA	RN x OTHER	GA x OTHER
GOALS:															
The prevention of disease or abnormality	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	0.05	nss	0.05
The removal or lessening of patient anxiety	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss
A reduction in the number of carriers of genetic disorders in the population	nss	nss	0.0001	nss	nss	nss	0.002	nss	nss	0.003	nss	nss	0.0008	0.02	nss
Provide clients with medical and genetic information	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss
DECISION-MAKING STRATEGIES:															
Tell clients that decisions are theirs alone and refuse to make any decisions for them	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss
You will support any decisions that clients make	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss
Inform clients what most other people in their situation have done	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss
Inform clients what you would do if you were in their situation	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss
Advise clients what they ought to do	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss	nss

Agendas in Genetic Counselling

This section, a description of genetic counselling session agendas, is based on material collected from questionnaires and interviews. The agendas outlined here are constructed from individuals' responses to my question, "What do you do here?" and are not the protocols suggested in publications (see, for example, Kelly's (1977) four stages of genetic counselling, Silverberg and Godmilow's (1979) six stage process, or Cohen (1984)). Furthermore, this section reflects primarily the agendas of the central caregivers in genetic counselling, the physician geneticists. Discussion of the way in which genetic counsellors outline their interaction with clients provides a clear indication of the sorts of things they feel are relevant and important to that interaction.

The majority of participants responded to my question, "What is genetic counselling?" by describing the process they go through with their clients rather than outlining a set of goals. As mentioned above, my questions about individually held goals were also answered in this way. The language of description and the emphasized aspects of the process varied from individual to individual, but there were common patterns throughout.

Each genetic counselling professional described his or her work as one of collecting, interpreting, and giving information--primarily genetic and medical information. Information collection included taking a patient's pedigree, asking about medical history, obtaining birth or medical records, test results, and pathology lab reports, arranging for additional tests, and in some cases, doing a physical examination. Genetic counselling professionals are in agreement that this information is essential to their work. It is at the heart of what they are trained to do.

In making a diagnosis, genetic counsellors tended to agree that the most valuable kinds of information come from test results (*e.g.*, a chromosomal translocation apparent on a karyotype) and from the pedigree (discovering that the client's problem is found also in relatives). Sometimes the diagnosis is made on the basis of a karyotype or other test result even before the geneticist has met with the client. One geneticist said he sometimes asks people to "go away and come back" if he needs time to work on his diagnosis. Diagnoses may be made in consultation with other clinic or team members in a "case conference" or weekly clinic meetings.

Methods of collecting and assessing the genetic and medical information vary little. Genetic counsellors tend to ask similar questions of each client, and some mentioned they have a "mental checklist" of questions to go through. Obtaining a pedigree and family medical history are done in much the same way as a physician takes a medical history, by asking a series of specific, usually close-ended questions. For example, a geneticist might ask: Did you notice anything unusual about the pregnancy before your miscarriage? Did you take any drugs during your pregnancy? Did your mother or your sisters ever miscarry? Recommending certain diagnostic tests, for example a karyotype where trisomy 21 (Down syndrome) is suspected, or constructing a pedigree for each client, is not only routine but their omission would be viewed as poor practice.

In the third step of the counselling process, providing information for the client, there is more variation of protocol. There are really two parts to this third stage--first, presenting the medical and genetic facts to the clients, and second, discussing the implications of those facts with the client. Discussing the diagnosis is done by explaining the genetic basis, the mode of

inheritance, and the manifestation of the disorder in affected individuals. Although these explanations are considered fundamental to any understanding of the situation by the client, some genetic counsellors questioned how much detail to include. The concern here is that the information be "pitched at a level the client could understand." The concern with explaining the medical implications of a genetic disorder is to avoid "scaring someone off" by being directive.

In addition to describing the disorder, the risk of passing the condition on to offspring is calculated for the client and often explained with the aid of charts, diagrams, or analogies (drawing cards from a deck, flipping a coin, the chance of rain on a given day). Putting the risk of genetic disease in some context is considered important since "numbers alone are not very meaningful" for clients. Several people said it was important to provide the risk figures but felt they may not really be that important to clients.

> For a lot of people the numbers are not very meaningful. . . . I don't tend to be overly concerned with them remembering the actual numbers. If they remember that it is an inherited condition and there is some risk for themselves or for their children that is strong enough to motivate them to come back [if they want more information], then I think they've got the basic message. And whether that is one in ten or one in 362. . . . [The counsellor stops talking and shrugs] (nonMD/nonPhD genetic counsellor).

> I don't think people really want counselling about risks, I think they want information about the medical impact of the disease (MD/PhD geneticist).

> I downplay the statistics. I don't harp on it. I emphasize the management aspect (MD geneticist).

Discussion of the implications of the genetic disorder for the client in terms of options for treatment or management is the second aspect of this third stage. Discussion of client options is considered an important part of the protocol although the manner and extent to which they are discussed may vary.

For example, in one of the questionnaire cases, involving a couple who have learned that their unborn child has Down Syndrome, the counsellor "asks the couple if they have ever thought about raising a mentally handicapped child; . . . [he] gives them a pamphlet from a local Down Syndrome parental support group and suggests they contact the group." Many of the responses to this case labelled the counsellor as "directive" for failing to present all the options--in this case, therapeutic abortion and adoption, as well as keeping the child--to the couple. The importance of presenting all the options available was stressed repeatedly in interviews and questionnaire responses; it is a widely agreed upon protocol.

Several individuals suggested the best way to discuss the implications and the means for clients to deal with genetic disease was by trying out alternative "scenarios". Others suggested they would ask clients what they wanted to do and then discuss only that option with them. Two individuals stressed the importance of establishing exactly what their clients' concerns are.

> I'm tending now to start [the session] by saying "why are you here?" I initially started off saying this is what I'll do for you and I ended up answering questions people weren't asking (MD/PhD geneticist).

In publications, the purpose of these discussions is described as allowing clients to deal with the disorder in a way that is relevant in terms of their family goals (for example, Fraser *et al.* (1974:637) or Silverberg and Godmilow (1979:281)). In interviews and questionnaire responses this goal was more often described as having clients arrive at a decision or plan that is "rational", "intelligent", "sensible", or "reasonable". Rational decisions are also understood to be informed decisions--ones that utilize the information provided to the client by the counsellor or that are made after this information has been provided. If

a client's initial reaction is to do something like the father in questionnaire Case Four (see Appendix One) who refrained from telling his daughters they might be at risk for polycystic kidney disease or the woman in Case Three who does not want to hear about her unborn child's neural tube defect, this reaction may be interpreted as "irrational". An irrational decision is one that is made before the client has heard all the information and options the counsellor determines are important. Irrational decisions are associated with strong emotions. An emotional reaction is not conducive to utilizing the information provided by the genetic counsellor and will hinder a "sensible" decision. If a client reacts very emotionally to the news of a genetic disease, the counsellor may suggest he or she return in a few days to discuss the client's options. That clients need time to get over the shock of the diagnosis "so they can handle the information and come to terms with the facts" is widely agreed upon among genetic counselling professionals.

Recall that the American studies investigated the strategies of decision-making endorsed by genetic counsellors. Counsellors' opinions on the appropriateness of degrees of involvement in client decision-making is not easy to determine accurately from a questionnaire (see Table 4.1). Predictably perhaps, the large majority of respondents indicated it was appropriate for genetic counsellors to "tell clients their decisions are theirs alone and refuse to make any decisions for them" (90.5% = 67/74), "to support clients' decisions but not make any for them" (97.4% = 74/76), and that it is inappropriate "to advise clients what they ought to do" (91.8% = 67/73). Clearly the group recognizes the concern that they be non-directive and let clients make decisions on their own. As with goals indicated by genetic counsellors, comparison with Sorenson *et al.* (1981) shows a pattern of whole group response that is similar for both

American and Canadian samples. (Sorenson and colleagues' 1981 publication does not list between-group differences in decision-making strategies.)

Two types of involvement in client decisionmaking that did show differences between professional groups concerned the use of examples based either on the counsellor's or other clients' experiences. MDs/MD-PhDs were signficantly more likely than nonMD/nonPhDs to indicate that it was appropriate for genetic counsellors to "tell clients what other clients in their situation had done" (see Table 4.2). As well 36.4 per cent (14/36) of the MDs/MD-PhDs felt it was appropriate for counsellors to "tell clients what they would do if they were in their situation" while only 6.5 per cent (2/31) of the nonMDs and nonPhDs find it appropriate to respond personally. These results suggest a generally held reluctance to include oneself or one's own values in the client's decision-making process and an understanding that knowing what other people in similar situations have done may be helpful to their own clients. The differences in this response between MDs/MD-PhDs and nonMDs/nonPhDs may reflect a difference in ideas about being non-directive. Whereas MDs/MD-PhDs may view non-directiveness as simply avoiding telling people what to do, non-MDs/nonPhDs may prefer to let clients come up with decisions on their own.

The importance of discussing more than the genetic and medical facts with clients was emphasized repeatedly. The following quotation suggests additional information the counsellor should include in the session.

> As a prelude to genetic counselling it is important to divine a couple's educational and social background, their religious attitudes and, if possible, something of their marital relationships if information is to be presented effectively and sensitively. (Emery 1984:5)

That there are no guidelines for "divining" or utilizing this psychosocial information is no doubt connected to the sentiment expressed by genetic counsellors that their work was "more than counselling about risks--it's tougher than that." As the following quotations make clear, counsellors do acknowledge the importance of the psychosocial issues for clients. But the majority of genetic counselling professionals I interviewed did not feel comfortable discussing the social realities of genetic disorders with clients. As one geneticist put it, he prefers to discuss the "problems that are medically meaningful."

> You see, clients don't need continual genetic counselling they need continual pyschotherapy or psychosocial counselling (MD geneticist).

> Fifty percent of the people I see . . . really just want to talk. They don't necessarily want to talk about genetics, they want to talk about the impact the disease had one their family, and the fact their marriage is breaking up, and their child needs a lot of attention and how do they get the child to daycare, and this sort of thing. . . . They just really want to talk about the impact the disease has on their family (MD-PhD geneticist).

Several participants stressed the need for clients to develop "an understanding of what the diagnosis and risks mean to them" and to "deal with the burden of the genetic disease" through discussions with the counsellor. Exactly how the counsellor assists the client in this process is not clear although client social, cultural, and emotional factors were noted by some study participants as important to the process of dealing with the genetic disease. One interviewee suggested the following:

> Let them [clients] develop their own perception [of the risks] by indicating if the child is affected the likelihood is such and such, the life expectancy, what physical disabilities the child may have. In other words as clear and comprehensive a picture of what an affected child would be like. It is important always to do that because it is one of the elements in parental decisions as to what they should do; not just the statistical risks but their perception of what the burden of having an affected child is (MD geneticist).

Many different topics may be discussed during this part of the session: the client may wish to express anxieties about raising a handicapped child, feeling of inadequacy in their personal relationships, conflicting emotions about having an abortion, or despair at the thought of having to try again to have a normal child. In order to determine how they are included in the sessions, I asked questionnaire participants to indicate for a number of issues whether they prefer to allow clients to raise the issue, to create an opportunity for clients to bring it up or to confront clients with the issue. For eight of the nine issues listed the preferred way of including the issue in discussion was to create an opportunity for the client to bring it up (see Table 4.4). When the issue is telling family members who may be at risk for the genetic disorder, the group preferred to confront the clients directly. In interviews it was clear to me that telling at-risk family members is one topic where genetic counselling professionals feel particularly motivated to encourage their clients in how to proceed. In general, however, confronting clients was the least preferable method especially for discussing the clients' religious views, the economic cost of the disorder and the effect of the disorder on the clients' sex life. Simply waiting for the client to bring up issues was also not a widely endorsed method. These patterns of raising issues are not statistically different among MDs/MD-PhDs, PhDs, and nonMDs/nonPhDs or any of the Level One groups.[1]

The point at which the genetic counsellors' role is completed varies from case to case. Most sessions with a genetic counselling professional are a one-time occurrence for a client and last between one and one and one-half hours. After one and one-half hours "everyone is talked out." Very rarely do clients return for further consultation; although they are encouraged to do so "if they have more questions" the responsibility to return is left up to the client.

Some professionals said they are satisfied with a session once the client "has made a decision," "has no more questions," or "when the information has been understood." Others pointed out that cases involving high-risk families or disorders where treatment- or diagnosis-related developments are occuring rapidly are never really closed. After the session is over, the genetic counselling professional usually sends a letter outlining his or her findings and recommendations to the referring physician; infrequently, the client receives a copy or a similar letter. The letter in effect transfers responsibility for further client care, particularly for further discussion of psychosocial matters, to the family physician. As mentioned in Chapter Three, counsellors expressed little or no hesitation in doing this. The assumption that a family doctor knows the client best and the desire to preserve the consultative relationship overrides the worries genetic counselling professionals may have about the referring doctor's ability to deal with genetic diagnoses.

Follow-up to counselling, additional contact with the client sometime after the primary visit, is a particularly interesting topic. Although the importance of detailed follow-up to counselling was noted repeatedly in publications, in interviews and in the comments accompanying each hypothetical case on the questionnaire, it is done infrequently. For this reason I have not included follow-up as a fourth stage in the protocol of counselling. Follow-up to see how clients were coping with the diagnosis and decision-making is almost never done. Some programmes have a routine six-month and/or eighteen-month follow-up to check clients' "comprehension and retention of the facts." One geneticist I interviewed said the follow-up he does "tends to be project-oriented rather than patient-oriented." Several nonMDs/nonPhDs said they contact clients who have come for prenatal diagnostic testing by phone or mail to obtain statistics on the

TABLE 4.4

PREFERRED METHOD OF RAISING ISSUES

	Total Sample		MDs/ MD-PhDs		NonMDs/ nonPhDs		PhDs	
EFFECT OF GENETIC DISORDER ON CLIENT'S FAMILY LIFE:								
Allow client to raise issue	6.8%	(5)	14.3%	(5)	0.0%	(0)	0.0%	(0)
Create opportunity for client to raise issue	79.5%	(58)	71.4%	(25)	83.3%	(25)	100.0%	(8)
Confront client with issue	13.7%	(10)	14.3%	(5)	16.7%	(5)	0.0%	(0)
Refer to other professional	0.0%	(0)	0.0%	(0)	0.0%	(0)	0.0%	(0)
EFFECT OF GENETIC DISORDER ON CLIENT'S SEXUAL LIFE:								
Allow client to raise issue	24.7%	(18)	22.2%	(8)	20.7%	(6)	50.0%	(4)
Create opportunity for client to raise issue	61.6%	(45)	58.3%	(21)	68.9%	(20)	50.0%	(4)
Confront client with issue	9.6%	(7)	16.7%	(6)	3.4%	(1)	0.0%	(0)
Refer to other professional	4.1%	(3)	2.8%	(1)	6.9%	(2)	0.0%	(0)
GENETIC DISORDER AS A REASON FOR LIMITING FAMILY SIZE:								
Allow client to raise issue	12.5%	(9)	11.4%	(4)	10.0%	(3)	28.6%	(2)
Create opportunity for client to raise issue	69.4%	(50)	65.7%	(23)	76.7%	(23)	57.1%	(4)
Confront client with issue	16.7%	(12)	22.9%	(8)	10.0%	(3)	14.3%	(1)
Refer to other professional	1.4%	(1)	0.0%	(0)	3.3%	(1)	0.0%	(0)
METHODS OF CONTRACEPTION:								
Allow client to raise issue	11.1%	(8)	11.1%	(4)	14.3%	(4)	0.0%	(0)
Create opportunity for client to raise issue	54.2%	(39)	44.4%	(16)	60.7%	(17)	75.0%	(6)
Confront client with issue	27.8%	(20)	41.7%	(15)	14.3%	(4)	12.5%	(1)
Refer to other professional	7.0%	(5)	2.8%	(1)	10.7%	(3)	12.5%	(1)
ALTERNATE FORMS OF PARENTHOOD (INCL ADOPTION & ARTIFICIAL INSEMINATION):								
Allow client to raise issue	4.2%	(3)	5.7%	(2)	3.4%	(1)	0.0%	(0)
Create opportunity for client to raise issue	52.8%	(38)	45.7%	(16)	58.6%	(17)	62.5%	(5)
Confront client with issue	41.7%	(30)	48.6%	(17)	34.5%	(10)	37.5%	(3)
Refer to other professional	1.4%	(1)	0.0%	(0)	3.4%	(1)	0.0%	(0)
PROVIDING INFORMATION FOR FAMILY MEMBERS WHO MAY BE AT RISK:								
Allow client to raise issue	4.1%	(3)	5.9%	(2)	3.2%	(1)	0.0%	(0)
Create opportunity for client to raise issue	32.9%	(24)	26.5%	(9)	38.7%	(12)	62.5%	(5)
Confront client with issue	61.6%	(45)	67.6%	(23)	54.8%	(17)	37.5%	(3)
Refer to other professional	1.4%	(1)	0.0%	(0)	3.2%	(1)	0.0%	(0)
CLIENT'S RELIGIOUS VIEWS:								
Allow client to raise issue	23.9%	(17)	30.3%	(10)	16.7%	(5)	25.0%	(2)
Create opportunity for client to raise issue	60.6%	(43)	48.5%	(16)	70.0%	(21)	75.0%	(4)
Confront client with issue	8.5%	(6)	15.2%	(5)	3.3%	(1)	0.0%	(0)
Refer to other professional	7.0%	(5)	6.1%	(2)	10.0%	(3)	0.0%	(0)
ECONOMIC COST OF GENETIC DISORDER TO CLIENT:								
Allow client to raise issue	32.4%	(24)	28.6%	(10)	29.0%	(9)	62.5%	(5)
Create opportunity for client to raise issue	47.3%	(35)	45.7%	(16)	51.6%	(16)	37.5%	(3)
Confront client with issue	12.2%	(9)	17.1%	(6)	9.7%	(3)	0.0%	(0)
Refer to other professional	8.2%	(6)	8.6%	(3)	9.7%	(3)	0.0%	(0)
SOCIAL STIGMA OF DISORDER:								
Allow client to raise issue	21.6%	(16)	22.2%	(8)	20.0%	(6)	25.0%	(2)
Create opportunity for client to raise issue	56.8%	(42)	52.8%	(19)	63.3%	(19)	50.0%	(4)
Confront client with issue	21.6%	(16)	25.0%	(9)	16.7%	(5)	25.0%	(2)
Refer to other professional	0.0%	(0)	0.0%	(0)	0.0%	(0)	0.0%	(0)

MANN WHITNEY PAIRED COMPARISON TESTS FOR SIGNIFICANCE:
LEVEL II AND LEVEL I: no statistically significant differences

outcome of pregnancy for clinic records. Although they were not providing follow-up, genetic counselling professionals said they made sure their clients knew they were "welcome to call or come back if they had questions." The obstacles cited by study participants to more comprehensive follow-up were related to administrative or systemic problems such as "lack of funding" and "lack of personnel." Several of the individuals interviewed said they would like to see more comprehensive follow-up done.

Within the variety of individually described genetic counselling protocols, several items emerge in a pattern of counsellor-defined priorities and concerns. Information about the genetic basis and medical implications of the disorder, risks, and the psychosocial aspects of genetic disease and client decision-making are topics that were described in similarly patterned ways throughout the study. Not only do they provide genetic counsellors with a means of organizing and structuring their interaction with clients, but also these issues represent the complex and shifting boundaries of professional "responsibility" to the client. The following description provides both a summary of this section on counsellor-defined priorities and an introduction to the individually constructed notions of responsibility discussed in this chapter.

1. The value of genetic and medical information. Lab tests and pedigrees are considered valuable because they enable the counsellor to be make an accurate diagnosis and to calculate risk figures. The diagnosis and the risk figures are then given to the client and take on another value. These facts will help a client cope with the problem. Information about the range of management options available to the client is valued because it is felt to ensure unbiased or "nondirective" counselling.

2. The amount of genetic and medical information. The ideal is that clients be given only as much genetic information "as they need to know", because too much genetic detail is felt to be confusing. Clients are felt to have the right to know "everything" about the medical implications and the management or treatment options which surround their genetic problem. Curtailing discussion of the options for clients is felt to be "directive counselling."

3. The function of genetic and medical information. Genetic and medical facts have a variety of functions as perceived by the professional. In some cases, as with information about a prenatal diagnostic test, the information is expected to provide clients with an idea of what the test might show and a "realistic expectation" of what genetic counselling can accomplish. Medical and genetic information has the power to help a client deal with his or her emotional reaction to the genetic problem and prepares the client to understand more information so that he or she may make an informed decision.

4. Risks. Most genetic counselling professionals felt recurrence and occurrence risks could be comprehended by their clients when two criteria are met: the risks should be placed in context and the client must be "emotionally ready" to make sense of them. Detailed discussion of numbers and statistics was felt to be useless if the client is emotionally upset about the diagnosis itself. Risks are most effective when they are put in context, *i.e.*, in "the spectrum of lifetime risks" or in comparison to the three per cent "baseline" risk of a birth defect being present at birth (Thompson and Thompson 1986:303). (A low risk, one that is lower than ten per cent, is felt to be reassuring for clients.)

5. Psychosocial aspects and client response to genetic disease. As a group, genetic counselling professionals prefer to let clients raise issues for discussion by creating an opportunity for them to do so. At least initiating discussion of psychosocial aspects of a disorder or the client's response to genetic and medical information is considered by some participants to be their responsibility. Dealing with clients' emotional reactions is felt to be the most challenging part of counselling. "Eliciting" and "facing" these reactions is valued by counsellors because clients are perceived to need this opportunity to be emotional and to "talk" as part of the process they must go through to "face the facts" and take on the responsibility for dealing with the genetic disorder that confronts them. Although professionals recognize the importance of discussing the psychosocial and day-to-day implications of genetic disorders, the extent to which these matters are dealt with in detail may be circumscribed by individual willingness to do so and by the fact that most of their interactions with clients occur in a single hour-long session. In most cases, genetic counselling professionals assume clients who need further discussion will do so with their family physician.

6. Client decision-making. A central goal of genetic counselling is that clients make a decision. This decision has particular characteristics: it should be "rational", that is, not made as part of an emotional reaction, and it should be "informed" or made on the basis of the information clients received through genetic counselling. In fact, as Kenen (1984:546) has pointed out the expectation that clients will make informed decisions is "translated into no-risk-taking in cases with high recurrence risks and high burden defects." In other words, clients will choose to use whatever

means are available to them to minimize their risks of producing a child with a genetic disease.

Counsellors maintain that client decisions take time, require "support", and the opportunity to "talk things through." Genetic counselling professionals value the ability to be nondirective and to let clients arrive at a decision on their own.

a) Time is considered a valuable and powerful component in genetic counselling. Time functions, like information, to allow the client "to hear the facts", and to understand medical and genetic information, to pass through the emotional part of counselling for genetic disorders, and to arrive at a "reasonable" decision.

b) "Support", a term that was not clearly defined by genetic counsellors in either the questionnaires or the interviews, is felt to be particularly important for the woman going through prenatal diagnosis. Support for this woman from her spouse or "partner" is essential if the diagnosis is positive and is felt to be a key element in her decision-making ability. Long-term support for dealing with psychosocial concerns is the responsibility of the referring family physician.

The six points listed above outline common patterns of counsellor-defined priorities in genetic counselling. It is clear that counsellors' priorities have arisen from the two paradigmatic influences--medicoscientific and psychosocial--which guide genetic counselling. It is now possible to look at how these priorities (subsumed into the two paradigmatic-related categories of medical genetic information and psychosocial aspects) reflect the field's historical basis, contemporary structure, and professionally defined roles. Importantly, it is also possible to show that these priorities are connected to individually shaped

notions of "responsibility" in interacting with clients.

1. Genetic and Medical Information. Providing this information is standard protocol accepted by all the genetic counselling professionals in this study. It is recognized as the basis of their involvement with clients. Given that the historical focus of medical genetics has been to produce information about the diagnosis and treatment of genetic diseases the emphasis placed on providing this type of information to the client is understandable. Particular emphasis has been placed on determining a client's risk of inheriting or passing on a genetic disorder. The assumption here is that clients will respond to a "high risk" (the boundaries of which are not defined) by minimizing their risks, either by not reproducing or by availing themselves of prenatal diagnosis. However, there is increasing recognition that these risk figures are not very meaningful to clients and often not the basis of their decisions (Lippman-Hand and Fraser 1979a). Although genetic counselling professionals try to place risk figures "in context", the goal of doing so is often only to help people remember the number. Determining whether that number is important to clients, how clients conceptualize their risks, and what factors other than risks predominate in client decision-making is not clearly within the responsibility of the genetic counselling professional.

Furthermore, the present structure of genetic counselling in Canada continues to encourage a paradigmatic orientation in genetic counselling based on the production and dissemination of information about the genetic basis and medical implications of genetic disease. This information is considered to be made up of value free facts and there are relatively clear, easily learned protocols for presenting these facts; in

short, genetic and medical information is assumed to be nondirective. With little variation among individuals, outlining the genetic basis, medical implications, and client options is clearly felt by genetic counselling professionals to be a major part of their responsibility

2. Psychosocial Aspects. Although genetic counselling professionals may feel they should initiate discussion of client responses to genetic disease and certain psychosocial issues, they do not all feel a responsibility to discuss these matters either in detail or over the long term. There is very little historical support to encourage counsellors to explore the meaning genetic disease has for clients. Ties between a psychosocial paradigm which supports this "counselling" or "communication-oriented" care and the contemporary structure of genetic counselling are weak. Individuals trained in the psychosocial aspects of genetic counselling often have a peripheral role in client care, training in counselling and communication oriented medical practice is minimal, and the traditional encounter of patient with specialist--a single brief "consultation"--limits the potential for developing stronger ties. Practitioners are aware that emotional, social, and cultural factors are relevant to client decision-making and coping (Applebaum and Firestein 1983:6; Murray 1984). However, there is no clearly defined protocol for eliciting and responding to these factors and for assisting clients to deal with the genetic and medical "facts" in a way that is relevant and meaningful to the client. Furthermore, genetic counselling professionals are concerned that these factors and the range of psycho-social issues which may arise in their work are value-laden. A primary concern of counsellors in dealing with the psychosocial aspects of their work is to remain nondirective: "giving advice without

telling people what to do." Rather than professionally sanctioned protocols, there are individual motivations about the degree to which each genetic counsellor's responsibility includes the discussion of psychosocial issues.

Individual Approaches to Genetic Counselling: Four Case Studies

Individual motivations and characteristics among those genetic counselling professionals I interviewed were not difficult to determine. Although here I have cited from only four individuals, interviewees in most cases had very clear ideas about how their approach to genetic counselling differed from that of their colleagues; in several cases, individuals could describe the essence of this difference in one word. Among all individuals interviewed, discussions about their personal approach to genetic counselling inevitably concerned, as did questions about personal goals, the emphases each placed on either the medical/genetic or the psychosocial component of genetic disorders. This duality and interplay of emphases, described from several perspectives throughout this thesis (paradigmatic, organizational, educational and counsellors descriptions of their agendas) has a heightened importance for genetic counselling professionals in their interactions with clients. Individual approaches to the psychosocial aspects of genetic conditions determine the boundaries of that counsellor's responsibility to the client.

In order to look at individual motivations among genetic counselling professionals I have included comments from four individuals who talked about dealing with the psychosocial component of their work. In selecting these passages I have tried to draw attention to the issue at hand and at the same time

allow each individual's style to come through their own words. The names that have been used are fictitious.

A. Dr. Smith

Dr.Smith is a physician who describes himself as an "expert in medical genetics." He has been doing "genetic assessments" for over 15 years and sees one or two clients each week. Dr. Smith views himself as a "consultant to the family doctor"; he sees clients in "a one-shot deal" and prefers to leave both the long-term medical and the "psychotherapeutic management of genetic disorders" up to the client's family doctor.

Several years ago, Dr. Smith became concerned about "patient disinterest" and inattentiveness during his counselling sessions. After discussing a particularly disturbing case with a colleague, Dr. Smith began to feel that he was not able to deal with what he calls the "psychotherapeutic" or the "emotional" aspect of his work. "I could ask the questions but I couldn't provide clients with answers."

Now, "as part of the procedure", clients who are referred to Dr. Smith also meet with a hospital social worker prior to their appointment. Although he notes that most people don't understand why they have to see the social worker and some protest that they don't need a social worker, Dr. Smith is pleased with the arrangement.

> Some people question seeing [a social worker], but they are calmed by him if he does his job. Although it [seeing both a social worker and a geneticist] makes the experience somewhat fragmented for them, I think it is beneficial for the patient because she is less distacted when she sees me. She is paying attention to me. . . . Patients used to just stare out the window while you talked to them, but now they sit forward and pay attention.

Involving the social worker relieves Dr. Smith as he describes it,

> of having to deal with touchy situations. Now when I sense the patient is sensitive about something I am free to back off and then ask [the social worker] about it later.

For Dr. Smith the boundaries of his responsibility and his skills in genetic counselling are clear. Because patients come to him with "medical problems," he feels they should see a physician such as himself. However, for Dr. Smith, "the social worker's part is the therapeutic aspect" and he argues that, along with other geneticists, he does not have the skills to deal with the psychosocial concomitants of genetic disease.

> Everyone has their own philosophy about how to counsel. . . . Everybody is a counsellor, but not everyone is able to deal with the wide variety and the extremes of reactions. [I mean] everyone can give advice to a friend, but not everyone can do it in such a way that it helps that person.

B. Dr. Jones

Dr. Jones is a physician who works in a regional genetics clinic as a "staff geneticist." He sees six to seven families each week of which about 80 per cent of the cases do not return ("they don't need to") after the initial hour long visit. His ability to provide good genetic care, according to Dr. Jones, is tied up with his commitment to "the pastoral aspect of being a family doctor to patients." Pastoral care, for Dr. Jones, means the doctor is

> someone they can come to and get answers not just to the genetic side of things but how to deal with it. I guess it's the idea of a family physician being concerned about the impact of this particular birth defect upon the family,and its repercussions on the family, in an ongoing way than a one-time only visit. It's a more complete use of my skills that I've been trained to use going to medical school.

Eager to help me understand the concept of pastoral care, Dr. Jones quoted from a letter written by one of his colleages Commenting "now that's a definition of pastoral care," Dr. Jones read:

> 'I think most patients never confront the fact that they expect more from us than simply the technical exercises of diagnosing and treating illnesses. They expect care, emotional support, sympathy, loyalty, sacrifice, respect, and trust.'

For Dr. Jones, the key to his ability to provide pastoral care is that he strives for the "art of medicine" or what he sees as "putting spirtuality into practice." In genetic counselling specifically this means becoming involved in his clients' emotional reactions.

> There is a debate going on about whether the psychological support of patients is part of genetic counselling or not. I happen to think it is. I have been rewarded by my efforts for taking on the psychological support.

Not only does he treasure the ability to practise the "art of medicine" but Dr. Jones feels it is a skill nonphysicians do not possess.

> I am practising medicine. That's different from what the genetic associate does. They can certainly have as much, even more, pastoral involvement with the couple that has a problem, but I think I can get into the situation deeper than they can. There is an additional problem with the PhD . . . it must be terribly frustrating for them not to get terribly pastoral or terribly into the family. They can just give the textbook explanation of what this is that the patient can really read about. . . . There is the science of medicine that we all share, but there is the art of medicine which I don't think is present for the others.

C. Dr. Newton

Dr. Newton describes himself as a "clinical geneticist . . . who dabbles in everything." He sees 15 clients each week and has been involved in genetic counselling for 11 to 15 years. On the one hand, Dr. Newton limits his responsibility to his patients in terms of providing genetic and medical information.

> I think my responsibility is to provide the most accurate medical diagnosis I can or the mostaccurate risks of recurrence that I can. And let them know if I think there is anything extraneous which may modify those risks that I've given them. I think that's the limit of my job.

But he also feels, as does Dr. Jones, that the practice of medicine involves "something more" than simply providing facts. As the second passage indicates, this ability to provide something more comes directly from being a doctor.

> I think what I do is the practice of medicine. I think many geneticists don't do the practice of medicine. There is a difference. I think that genetics is part of medicine and it should be practised in the same fashion. . . . [Patients] are really in here to talk about the impact of their problems and their lives and they want a little genetic information. There are two classes of medical or clinical geneticists: those who are interested in practising medicine and those who are interested in providing numbers.
>
> It sounds terrible, but I think you actually have to go through the medical indoctrination. . . . It's very easy to say 'your child has hydrocephalus and you have a one per cent chance of having that recur.' You say it with a different light after you've had to take care of a child with hydrocephalus and know all the complications that can go on. I used to think non-medical people could do counselling. I realized they can't do it because I changed so much having seen the disease [hydrocephalus] in its natural progress. You can only pick that up by dealing with it on a day-to-day basis.

His experience as a practising physician has convinced Dr. Newton that "there is no such thing as a non-directive counsellor." As he says,

> if the family wants to know and they ask me, I'll tell them. I am a directive counsellor because my experience in . . . medicine showed me that people really do want good advice as to what to do. . . . I think you have to be directive and just try to rein in your enthusiasm. . . . It's very difficult to rein yourself back. I'm entitled to be directive, but I hope I'm pretty honest and make my biases known.
>
> I remember once in the last two years bringing up 'Do you really think you ought to have children?' Shouldn't do that, but I did. Tsk, tsk, I'm turning in to a eugenicist. But this was a couple who had a lot of problems and I wasn't quite sure they understood

> what I was saying. And I felt, 'when you look at all your problems, do you really think you ought to bring a child into this situation.' I brought that up. I felt guilty doing that. I felt I had stepped over what my boundary had been.

D. Ms. Anderson

Ms. Anderson came to genetic counselling three years ago with a background in social work and now sees about two clients each week. Although she has access to a physician for assistance in matters of diagnosis and treatment, the majority of her contact with clients is on her own. Ms. Anderson views herself as an "information-giver".

> To give people just as much information as I possibly can about what they are coming to us about. The condition, the risks, the cause, whatever, and also to refer them to the appropriate community resources and to provide some support for them, and to be completely nonjudgmental. If you see yourself as an information giver I think you have to be nonjudgmental. . . . My position is to support her in whatever she decides and just give her as much information as possible so that she can make an informed decision.

For Ms. Anderson her commitment to her role as an information-giver stems from her desire to remain objective and supportive in her interaction with clients. It is also influenced by her own experience receiving genetic counselling in 1975 and in 1979.

> I was very aware of the change in the four years that it occured. I was given a lot more information. . . . I learned a lot more about the condition in my second session than I did in the first. . . . So I left feeling better about it.

Ms. Anderson is very clear about the nature of her role. But she also shares the concern that care for families facing genetic diseases, that genetic counselling include more than just the provision of information. It is care that comes from listening, talking, and communicating with clients. For Ms. Anderson, however, this additional care comes from social work, and is not based in medicine.

> I also think there should be some kind of social work input into counselling because I really think that sometimes we stir up a hornet's nest. That's not dealt with. No matter how much you explain that no-one is at fault, that you can't choose your genes, that's not the way people feel. That's very hard to deal with. I'd like them to have someone that they could call up a few weeks down the road and say, 'you know, I really just want to talk about what this means to my family and what this means to me.'

Individual Boundaries of Responsibility

To conclude this chapter I want to examine the connections between these individuals' approaches to genetic counselling and his or her ideas about responsibility in interacting with clients. Where an individual feels comfortable situating the boundaries of his responsibility to deal with the psychosocial issues of genetic disease forms the basis of individual responses to the paradigmatic pressures that exist in genetic counselling. As such these boundaries are dependent upon a variety of factors discussed in this thesis--the counsellor's education and experience, their tasks and role, and the particular genetic, medical, and social nature of each case. Other factors not investigated in this study, but suggested by Lock (1985:125-126), could include the counsellor's age and sex, personality, speciality within medical genetics, proximity to other healthcare facilities, professional literature, and the mass media. But thinking about how each individual defines their responsibilities led me to consider an additional infuential factor.

Each individual's sense of responsibility is linked to their view of the boundaries of "medicine". Medicine is no longer, if it ever was, clearly bounded. Conservative opinion that medicine is restricted to the diagnosis and treatment of diseases of pathologic origin assumed doctors would leave the treatment of nonpathological human ailments to others. But as the definition of dis-

ease has widened to include "illnesses" with a predominantly social origin (*anorexia nervosa*, for example) practitioners of medicine are now urged to deal with the psychosocial concomitants of pathologic disease and the psychosocial ailments of their patients. Genetic disease is a case in point here since the pathological realities are often not treatable, the client is often not physically ill, and the psychosocial adjuncts may strike at one of our most deeply held and cherished values--the ability and freedom to reproduce "well".

The response of medical professionals to social pressure "to treat the patient" and not just the disease has been mixed. Some physicians continue to practice as before, but may acknowledge concerns for psychosocial care by referring patients onto social workers, psychiatrists, or community resources. Others have opened their doors to whatever needs their clients bring, perhaps arguing that medical doctors have always done so. Both alternatives exist in genetic counselling.

The response of nonmedical professionals to the changing nature of medicine is important here too. While medicine has altered its boundaries, fields like psychology, social work, and even social sciences like anthropology and sociology have sent their disciples out to encourage these changes and carve a niche for themselves in medical matters. Nonphysicians who work "within" medicine may actually view their contribution to medicine as one which overlaps or is separate from, complements or conflicts with, is equal to or secondary to medicine. Their position in relation to medicine and hence to medical practitioners will influence what they accept as their responsibilities. An important point must be noted here. For nonphysicians in genetic counselling, particularly for nonMD/nonPhDs, the opportunity to express their role as complementary

and equal to medicine is often restricted by the paradigmatic and occupational domination of MDs.

Attaching boundaries to medicine and defining responsibilities in accordance with those boundaries is not easy to do. The boundaries are never immobile, they are continually influenced by both social and individual priorities. As the boundaries of medicine have shifted so do the responsibilities of individuals. Although Dr. Newton may define the limits of his responsibility to providing accurate diagnostic and risk information, medicine for him must do more than just provide facts and numbers. For Dr. Jones the boundaries of his medicine are permeable to the psychosocial needs of his patients. For Dr. Smith the boundaries are much more narrowly drawn to exclude the psychosocial or "sensitive" topics. Ms. Anderson confines her responsibilities to information-giver, secure in the assumption that this will prevent her from becoming directive or judgemental. Recognizing her contribution as one distinct from medicine, the lack of support for in-depth psychosocial counselling means she can only wish for more input of a social work nature in genetic counselling.

Summary

Acceptance that genetic counselling is based on the provision of objective genetic and medical facts is widespread. Genetic counselling professionals also feel that clients must deal with the psychosocial expressions of these pathological realities. But does dealing with those expressions fall within the professional's domain of responsibility? Does it fall within the realm of medicine as "medicine" is defined by each person? Can the psychosocial response be separated from its genetic basis and medical implications so that a team of specialists may each contribute their skills to good genetic care? As I have tried

to show, resolving these questions which concern the psychosocial concomitants of genetic disease is dealt with by each professional on an intensely personal level. Within the concensus that good genetic care involves both medical genetic and psychosocial care exist individual patterns for acting upon that concensus. As the boundaries of responsibility are drawn in dealing with the psychosocial aspects of genetic disease they enclose individually constructed and relevant characteristics of the people providing genetic counselling. The motivation and support for being more than an "information-giver" comes primarily from the individual, secondarily from the contemporary organization of genetic counselling, and only minimally from historical tradition in the field.

CHAPTER NOTES

1 The issues I listed in the questionnaire were based on those used by Sorenson and Culbert (1977). However, our results are not directly comparable. Sorenson and Culbert (1977:147) asked respondents to indicate whether each issue should be included in genetic counselling and whether they felt a professional obligation to raise the issue. Although most counsellors indicated the issues were important for genetic counselling, a professional obligation to raise four of the issues was not widely endorsed. Limitation of family size because of genetic disease, client's methods of contraception, effect of genetic disorder on client's sex life, and the economic burden of genetic disease on society although they are important for inclusion in genetic counselling raising them was not considered a professional obligation (Sorenson and Culbert 1977:149-150).

CHAPTER FIVE

Genetics and Counselling: An Unresolved Tension

This final chapter draws together the major points of this study by focussing my analysis on the paradigmatic tensions which underlie genetic counselling and the impact of these tensions on individual practitioners.

As suggested in the introduction to this thesis, an anthropological approach to Biomedicine is based on understanding 1) the historical and social context of this system of healing, 2) the ideas, motivations, and behaviours of its practitioners, and 3) the meaning Biomedicine has for those practitioners. Genetic counselling, then, like any other sociocultural phenomenon, is a dynamic compilation of ideas, actions, and meaning produced and embodied by individuals in a particular social and historical context.

From an historical perspective, I have argued that there has been an accumulation of competing goals in genetic counselling. Since its inception, the science of human genetics has been shaped by the context in which it occurs, the nature of its discoveries, and by the individuals involved. Knowledge, context, and individuals interact to produce priorities within the discipline. For genetic counselling professionals these priorities are translated into behaviour.

As discussed in Chapter One, the goals of genetic counselling and priorities of early genetic counsellors have been associated with a triad of interests--science and the goal of reducing the incidence of genetic disorders, medicine and disease prevention achieved by educating individuals, and psychology and the goal of helping individuals to deal with their emotional and

social responses to genetic disorders. The disciplines of orientation summarized here are also associated with certain modes of counselling: science and directive counselling, medicine and less directive, more educationally-oriented counselling, psychology and supportive or non-directive counselling. Whether or not these associations were ever quite so clear-cut is unlikely. For the contemporary genetic counsellor, however, they linger as systemic influences and extremes within which he knows he must act.

In their contemporary manifestations, the emphases which exist in genetic counselling do not separate neatly into the three disciplines of orientation--science, medicine, and psychology--but are more appropriately described in terms of two paradigms whose interconnections are unresolved: a medical or medicoscientific paradigm and a psychosocial paradigm. These paradigms are not viewed as entirely separate or opposed, one positive and helpful and the other negative and irritating to the participants in this study. However, the paradigmatic dichotomy which I have outlined does exist in their specialized culture: genetics is "easy" while counselling is "challenging".

Existing within the medical paradigm is a lingering "blush", as one geneticist suggested, of eugenics. Clearly, few genetic counselling professionals are in fact interested in eugenics. Nevertheless, the memory of eugenics has been translated onto an expanding reproductive technology against which our social and moral priorities strain. The status of this technology is ambiguous: on the one hand, prenatal diagnosis allows us to choose our children; on the other hand, that choice may be a painful dilemma (Rothman as cited in Canadian Broadcasting Corporation transcript 1986:18). Science is at once treasured and feared for its magical powers.

Also part of the medical paradigm are the familiar and respected goals of diagnosing, curing and preventing disease. Translated into a modern genetic context, the scope and meaning of these goals is altered. Diagnosis comes to include prenatal diagnosis or diagnosis in terms of risks. Treatment is limited and rarely curative. The goal of preventing disease, in its genetic form, becomes a goal of making clients aware of their means of avoiding genetic disease--prenatal diagnosis, therapeutic abortions, not reproducing. The practitioner's primary role is no longer "intervention" in the conventional prescriptive or curative sense, but becomes the provision of information to allow clients to make informed decisions.

The psychosocial paradigm is of recent origin in medical genetics and is still of uncertain status in relation to the medical paradigm. The psychosocial orientation demands attention be given to the emotional, psychological, and social concomitants of genetic conditions. It also suggests that clients' agendas be given priority even if that agenda is "irrational" or does not include an "informed decision". Although genetic counsellors are respectful of the need for clients to respond emotionally to genetic disorders and to explore the social meaning of the event, the realm of psychosocial issues is ambiguous and value-laden for many practitioners. The absence of clear-cut protocols for implementing or measuring the achievement of psychosocial goals creates further tension and resistance to this paradigm.

The context in which these paradigmatic tensions exist is critical to understanding their impact. The context for genetic counselling is almost exclusively a medical and scientific one. As such, it exerts a powerful and controlling influence over the practice of genetic counselling. The setting, the training of a majority of genetic counselling professionals, and the language of genetic

counselling sessions exemplify this medical orientation. Clients, usually referred by a physician, come to a hospital, clinic, or doctor's office for a genetic counselling appointment. They are likely to see a physician geneticist although a nonMD/nonPhD such as a nurse or a genetic associate may attend to administrative matters, collect the pedigree and then provide the client with some basic preliminary information about prenatal tests. The language and interaction of the meeting is medical: the client's family and medical history is taken, diagnostic tests results are discussed, a diagnosis of genetic disease is made, options for treatment, if any, are discussed, as well as options for management. In this context, management may refer to "managing the situation"--what avenues are open to the client for responding to the diagnosis of a genetic condition in themselves, their children, or their families?

The contemporary context strongly reinforces the provision of medical and genetic information. While it acknowledges the importance of psychosocial issues, the context includes only tenuous support for this paradigm through training in counselling and employment of psychosocially-oriented personnel. The medical paradigm and profession and genetic counselling have a relatively long and close association. The recent involvement of a psychosocial paradigm has created tension within genetic counselling. The impact of this tension was investigated, first , by looking at the roles of genetic counselling professionals, and second, by examining individually-held notions about responsibility in genetic counselling.

The description of roles found among genetic counselling professionals in this study is derived from questionnaires from 76 individuals across Canada and interviews with 11 people in Ontario (five MD geneticists and six nonMD/nonPhDs). Genetic counselling professionals divide themselves into

three groups: MD geneticists, geneticists who have only a PhD, and those who are neither physicians nor PhDs (nonMDs/nonPhDs). Physician geneticists have the central role in genetic counselling. In comparison to others, they see the most clients per week and do almost all of the diagnosis, treatment, and management related tasks. The routine of PhD geneticists includes few medical or counselling related tasks, but emphasizes their role in research. Nurses, genetic associates, social workers, and other nonMDs/nonPhDs are frequently involved with the day-to-day running of genetics clinics. Their involvement in client care is generally limited to information sessions for prenatal diagnosis and telephone or letter follow-up to counselling.

Who among these professionals should be providing genetic counselling? That the question is being asked by genetic counsellors reflects tension within the field between the two paradigms. Since the early 1970s, a number of nonMD/nonPhD genetic counsellors whose disciplines of orientation lie within the psychosocial paradigm have entered the discipline. In conjunction with a publicly expressed desire for a more psychosocially-oriented health care, their entry focussed attention on the capacity of MD and PhD geneticists to provide that care. The desire to deal with both the medical and psychosocial aspects of genetic care has brought with it a debate about who is best suited to provide such care. There is widespread agreement that the primary caregiver should continue to be a physician geneticist. A second and a well-defined role is ascribed to the PhD geneticist; although his science background and expertise in the laboratory are felt by his colleagues to impede his ability to relate to patients, his role in research is valued. The role of the nonMD/nonPhD in genetic counselling is more difficult to characterize and is a source of tension within the discipline. Although none of the nonMDs/nonPhDs in this study

expressed discontent with their role, they are perceived by their colleagues to be "overqualified" for clinic-oriented tasks and to be without a well-defined or satisfying role.

In part, the concern surrounding the role of nonMDs/nonPhDs is indicative of a perceived threat to the occupational dominance of medical practitioners in genetic counselling. In my opinion, tension between a medical and a psychosocial paradigm is another equally important factor. Turning attention to the psychosocial aspects of genetic disorders has not created a separate and complementary role for individuals who are skilled in or oriented toward 'counselling'. The counselling component of genetic care has been subsumed within the medical paradigm such that the responsibility for client's psychosocial needs are perceived to lie with the physician geneticist (or the referring family physician). If medicine is broadening its boundaries, is there then a need for non-physicians trained in psychosocial skills in genetic counselling? In short, although its emphases have changed in response to social priorities, genetic counselling continues to be a medical process dominated by medical practitioners.

How have the accumulation of orientations and the resulting paradigmatic tensions affected the individuals who are providing genetic counselling to clients? In particular, how have these tensions affected the dominant caregivers, the physician geneticists? In terms of content, there is agreement among all groups that the basis of genetic counselling should continue to be the provision of medical and genetic information. This is the "easy" part of genetic counselling: medical and genetic information is well-defined, relatively straightforward and understandable. With room for little individual variation in determining or applying it, medical and genetic information is valued for its objectivity both for the genetic counsellor and for the client. Provision of

information is felt to allow the genetic counsellor to remain non-directive and to help the client to reach a successful outcome of counselling. For genetic counselling professionals, this outcome is an "informed decision"--a decision that is unimpeded by emotionality and irrationality and is reached after clients "have come to terms with the [genetic and medical] facts".

Where do the psychosocial aspects of genetic counselling fit? Acknowledged by genetic counsellors as important to their clients, but perceived as value-laden and ambiguous, dealing with the psychosocial aspects of genetic disease is felt to be the "challenging" part of their work. Most counsellors do feel an obligation to initiate discussion of the client's emotional response (is it anger? guilt? fear?) and to raise some of the "sensitive" issues (the social stigma of disability, for example), but there is little historical or contemporary support for counsellors to explore in depth the social meaning that genetic disorders have for their clients.

The way in which individual genetic counsellors relate these two parts of the process--the "genetics" and the "counselling"--lies at the heart of their responses to the paradigmatic tension that exists within genetic counselling. Hahn and Kleinman (1982) and Hahn (1985) have discussed the process in which the patient's emotions and behaviour are "medicalized", that is "medically transformed and medically used" (Hahn 1985:91). Hahn elaborates in the following way.

> 'Listening to the patient' and 'taking the patient's history' are not efforts to sympathetically comprehend the patient's life world and inner meanings, fears, and desires, but rather to diagnose disease conceived of by criteria independent of their personal features (Hahn 1985:91).

In the context of genetic counselling, I would agree that the psychosocial aspects of genetic conditions similarly become medicalized.

Genetic counsellors do acknowledge the need for clients to be emotional and to talk but that need is often transformed into the means by which the counsellor's goal of an informed decision can be reached. Although clients may be encouraged to voice their concerns and anxieties about a genetic disorder, those feelings are not valid in their own right, but are viewed as obstacles to achieving the medical goal of an informed decision. The psychosocial aspects of genetic conditions are turned into a variable affecting prognosis, not disease prognosis in this case, but certainly the outcome of counselling. The social is subverted and ultimately converted to the medical.

I suggest that this process of medicalization is a powerful coping mechanism and tool for genetic counselling professionals. In effect, it allows them to feel more comfortable dealing with the psychosocial issues by evading them and denying their integrity and meaning for the client. Furthermore, for physician geneticists, in particular, medicalization validates their central role in genetic care; making psychosocial issues medical issues eliminates the need for genetic counsellors who specialize in the psychosocial aspects of genetic disorders.

I also suggest that medicalization inadequately describes the way in which genetic counsellors connect the components of their work. The process of medicalization as Hahn and Kleinman (1982:52-53) have described it does not take into account individually constructed ideas about the boundaries of medicine. The extent to which genetic counsellors include the psychosocial aspects of genetic conditions within their sphere of responsibility to their clients is dependent upon many factors--predominant paradigmatic influences, the particulars of each case, counsellor's education and routine tasks, for example. But responsibility in interacting with clients is ultimately dependent upon the

boundaries each genetic counselling professional attaches to "medicine". For some physician geneticists, the responsibility to deal with psychosocial issues is too great, too ambiguous, or too far outside the realm of what they have been trained to do. For others, it becomes part of the "art of medicine". Although I do not have enough data to explore in detail how nonMDs/nonPhDs situate their responsibilities in relation to "medicine", I suggest they may choose to view their contribution as one which overlaps or is separate from, complements or conflicts with, is equal to or secondary to, medicine.

Drawing the boundaries of their responsibility is a response to ambiguities and tensions genetic counsellors encounter in their interaction with clients. Based on my analysis I suggest that there are two culturally defined principles which exert a considerable influence on the action of genetic counselling professionals in responding to these ambiguities and ultimately to the underlying paradigmatic tensions. Guiding the action of genetic counselling professionals and motivating the medicalization of their work are two conflicting principles:

> 1) activism--the medical preference for "doing something" rather than "doing nothing" (Hahn and Kleinman 1982:44). Activism is reflected and expressed in the predominant concern among genetic counsellors to "help people". For most genetic counsellors this concern entails providing information about the genetic facts and medical implications of the genetic condition, diagnosing or labelling the condition, allaying fears, putting risks into perspective, and referring clients for further help when necessary. The goal of activism in genetic counselling is that clients will make an informed decision, one that is based on the information they have received during genetic counselling.

2) non-directive counselling--of paramount concern to most genetic counsellors is that they assist their clients in a manner that is value-free and allows clients to choose their own course of action. For most counsellors, abiding by this principle of non-directive counselling means providing only genetic and medical information (objective facts) and steering away from the social, emotional, and psychological concomitants of genetic conditions.

The first principle, activism, as Hahn and Kleinman (1982) have argued is a central element in the behaviour of Biomedical practitioners. Intervention, "doing something", is what medical doctors are trained to do. In the majority of cases, the patient goes to the doctor for help and thus empowers the doctor to intervene and to tell the patient what to do. But this interaction based on activism, an interaction that is familiar to both patient and doctor, conflicts with the second principle of genetic counselling--being non-directive. Non-directive counselling, a fundamental tenet of the psychosocial paradigm, arose, in part, in response to concerns that medical doctors were intervening and telling patients what to do.

The tension between "doing something" and remaining "non-directive" is particularly acute in genetic counselling. Although physician geneticists may give specific and directive advice concerning the medical care of individuals with genetic diseases, they cannot do so when the social concomitants of those diseases are encountered. For a number of reasons our cultural expectations about reproductive behaviour and medicine make this situation one of extreme ethical complexity and cultural sensitivity.

First, reproductive behaviour is a private matter, a concern of individual couples and of families. Any suggestion of interference by non-family

members or the state is threatening. Second, an inability to reproduce or an inability to reproduce a normal, healthy child may be perceived by parents as a direct reflection of their social worth and as a blemish on their self-image.

Culture-bound ideas about medicine also play a role here. First of all, we assume that medicine and science can cure our ills. Second, we assume that physicians act in our best interests. Third, we assume that medicine and science as they affect us through physicians are not influenced by the values of that physician. Thus, we have come to expect directive advice in medical matters, and we rarely equate that advice with the physician's morality.

Cultural expectations about reproduction and medicine meet in the sensitive arena of genetic counselling. People who come for genetic counselling have concerns and questions about their reproductive future; they may also have the expectation that the genetic counsellor will "prescribe" the solution for them, much as a physician would. But clients may also fear that they will be told how to manage their reproductive futures. Ultimately, the reproductive issue is one of choosing our children; a complex question that we tend to see as an issue of morality and ethics. Directive assistance in genetic counselling is not acceptable because it implies that the genetic counsellor's morality will influence the client's decision. Furthermore, non-directive genetic counselling means that clients must take on a responsibility for their decisions to which they are unaccustomed in medical matters.

For the individual genetic counsellor, there is no clear mandate for action which resolves the tensions created by these paradigms. Indeed such guidelines may be both impractical and impossible. The individual genetic counsellor must deal with shifting priorities--the scientific capacity to prevent disease, the client's right to know of this capacity, and the client's right to

reject it--and a complex mandate--to provide comprehensive genetic care that addresses both the clients' medical genetic and psychosocial concerns in a non-directive way. It is in this arena of contradictory and shifting expectations that genetic counsellors walk their tightrope.

Genetic counsellors may respond to the ambiguities of the social meaning of genetics by enhancing the medical component and converting psychosocial issues into medical ones--Hahn and Kleinman's "medicalization" that reduces the subjectivity of those issues. But they do so in ways which are congruent with their definitions of medicine. As a result, the culture of genetic counselling is comprised of individually constructed boundaries for medicine, boundaries which concern the responsibility to deal with the psychosocial aspects of genetic conditions. The behaviour based on these boundaries involves subtle and individualized distinctions between "doing something" and remaining "non-directive". As one questionnaire respondent suggested: "You have to give advice without telling someone what to do."

SUMMARY

In this thesis I have investigated the culture of genetic counselling in Canada. Genetic counselling is a relatively recent health service aimed at providing information and assistance to families at risk from genetic disorders. The emphasis in this study has been on understanding genetic counselling, particularly the roles, responsibilities and concerns from the practitioner's point of view. My central and theoretical focus is on the "individuals' versions of the culture of [genetic counselling]" (Hahn 1985:53).

Three means of collecting information for a practitioner-centered ethnography of genetic counselling were used. Publications about genetics, the objectives of genetic counselling and several surveys of American genetic counsellors' attitudes were employed. Responses from a Canada-wide questionnaire provided information on the sociodemographics, tasks, and division of roles of a sample of 76 genetic counselling professionals--45 MD and/or PhD geneticists and 31 nonMDs/nonPhDs (primarily nurses, masters level genetic associates, social workers). The third method of data collection was ethnographic interviews with five physician geneticists and six nonMDs/nonPhDs. The interviews questions focussed on the genetic counselling professionals' concerns as practitioners attempting to assist their clients.

There were three goals in this study, each of which contributed to understanding the individual's version of genetic counselling:

1) to fill an ethnographic lacuna by studying in an historical and contemporary sociocultural context Canadian genetic services and Canadian genetic counselling professionals;

2) to investigate the variety of practitioners--physician geneticists, PhD geneticists, nurses, master's level genetic associates, and social workers, among others, who are involved in providing genetic assistance to Canadians. Here, I was concerned with the nature of their roles and training, as well as their opinions and attitudes;

3) to explore the meaning and experience of genetic counselling for its practitioners as individuals within a context shaped by historical circumstance, sociocultural values, and personal experience.

In Chapter One I began the ethnographic component of the study by attempting to establish the historical context of genetic counselling in Canada. Unfortunately, information about the history of genetic counselling in Canada is virtually nonexistent. In this thesis I outlined the general development of applied human genetics and genetic counselling in the United States and, wherever possible, in Canada. Historically, the field of medical genetics has arisen from and been influenced by a variety of disciplines. In general, however, the practitioners of genetic counselling are from three main backgrounds--science, medicine, and psychology. PhD geneticists were the original "genetic counsellors". MD or physician geneticists entered genetic counselling in the 1950s--the beginning of the "medicalization" of human genetics. More recently, nonMD/nonPhDs, such as nurses, social workers, and master's level genetic associates, entered genetic counselling during the 1970s.

Attitudinal surveys done among American genetic counsellors during the 1970s have suggested the following associations between professional training, goal, and counselling strategy. PhD geneticists are oriented toward disease prevention and practice directive counselling to ensure that goal; MD geneticists want to prevent genetic disease and educate their patients toward an informed decision; nonMDs/nonPhDs focus on their patients' goals and want to ensure those patients can deal with the psychosocial aspects of genetic disease.

My research, however, does not support this simple association of profession-goal-counselling strategy. I argue that historical process has resulted in an accumulation of diverse goals rather than a complete shift from eugenics to psychosocial issues. I also argue that individual genetic counsellors are influenced by a variety of goals and orientations. I have suggested that those orientations come from the two paradigms which characterize contemporary genetic counselling and which influence the actions of genetic counselling professionals.

The medicoscientific paradigm recognizes a biological basis to human disease and is concerned primarily with the diagnosis, treatment, cure, and prevention of those diseases. The physician is trained to arrest the disease process through intervention. The psychosocial aspects of those diseases are often seen as secondary to the biological component. Attention to the psychosocial bases and implications of disease may be left to other professionals (psychiatrists or social workers, for example) or left up to the patient to deal with. The psychosocial paradigm recognizes psychosocial as well as biological bases to human illness. Dealing with the psychosocial component is considered essential to achieving health. Psychosocial therapy is non-directive and non-interventionist and consists in large part of listening to the patient talk. In

genetic counselling, the two paradigms have been drawn together with the current concern that adequate care must deal with both the biological and the psychosocial aspects of genetic disease. The dynamic that exists through the meeting of these two paradigms underlies many of the concerns of genetic counsellors and the meaning they construct for their work.

In Chapter Two, I investigated the support for both of these paradigms that exists in the context of contemporary Canadian genetic counselling. That context is overwhelmingly a medicoscientific one. Genetic counselling is provided in medical settings--physicians' offices or clinics in hospitals. The majority of professionals working in genetics are either physician- or PhD-geneticists whose educational background is either research or clinically oriented. The knowledge available to genetic counselling professionals (and passed on to their clients) is also weighted heavily toward medical and genetic facts. In comparison to knowledge about its psychosocial implications we know a great deal about the molecular, chemical, and physical implications of genetic disease. The organizational structure of genetic services in Canada is geared to the production and dissemination of information about the diagnosis, treatment, and prevention of genetic disease.

In comparison, the ties between genetics and a psychosocial paradigm in contemporary Canadian genetic counselling are much less well-defined. There is virtually no information available about the psychosocial services available to Canadians or the impact of those services for families facing genetic disease. There are signs, however, that the importance of psychosocial aspects of genetic disease is being acknowledged. Fellowship training programmes accredited by the Canadian College of Medical Genetics for MD and PhD geneticists note that Fellows should be "able to communicate genetic

information (counselling) in such a manner that the individuals and couples are assisted in making decisions concerning themselves and reproduction" (CCMG brochure 1984). There is also a master's level Diploma in Genetic Counselling (McGill University) which aims to establish competency in both the genetics and the counselling aspects of genetic care. The recent entry of nonPhDs/nonMDs, some of whom are trained in counselling skills, into genetic counselling also suggests the influence of the psychosocial paradigm in contemporary Canadian genetic counselling. The extent to which these individuals are employed in a position in which they may utilize these skills is hard to judge. Further insight into the paradigmatic influences in genetic counselling was determined by investigating the roles that exist among genetic counselling professionals.

Chapter Three focusses the ethnographic description on the variety and nature of professional roles in contemporary genetic counselling in Canada. The bases for this description are of two sorts. Data were collected through questionnaires and interviews on counsellors' activities, including types of tasks done and amount of time involved in those tasks. Ethnographic interviews were also used to explore individuals' perceptions of their training for their work and their opinion of the kinds of educational backgrounds that exist among their colleagues and co-workers. I found that questions about training and tasks prompted answers which frequently centered on the question of who should be providing genetic counselling. In my opinion this question is related ultimately to the paradigmatic tensions underlying the goal to provide care that deals with both the medical and psychosocial aspects of genetic disease. The central issue here for genetic counsellors is who among them is best able to meet this goal: MD- or PhD-geneticists who are trained in a medicoscientific paradigm or psychosocially oriented individuals such as social workers or genetic associates?

That genetic counselling is now viewed by the public and by many of its practitioners as a medical process has provided wide support for the central role of physician geneticists. MDs see the most clients per week, they are involved in the diagnosis, treatment, and management of genetic diseases and they indicate that they do provide 'counselling'. The continued recognition of the scientific basis of human genetics has accorded a separate but complementary role for genetic counselling professionals who hold only a PhD. PhD geneticists have minimal contact with clients (their training is felt by their colleagues to impede their skills in practitioner-client communication) but have a secure and essential role in research.

In contrast, the third group, nonMDs/nonPhDs, are felt by many medical geneticists to be without a well defined role. Questionnaires provided support for this opinion in demonstrating that nonMDs/nonPhDs are involved in a variety of tasks: some have limited contact with clients in prenatal diagnosis information sessions or intake visits to obtain pedigrees and basic personal information from the client, but the majority are doing clinic-oriented clerical jobs. The counselling or psychosocial orientation they bring to genetic counselling is acknowledged as important, but it is subsumed within the medical paradigm so that psychosocial care falls within the role and responsibility of the physician geneticist. In sum, the differing roles of physician geneticists, PhD geneticists, and those with other degrees separate the professional community along lines that reflect the origin and existence of both a medicoscientific and psychosocial paradigm in contemporary genetic counselling.

In Chapter Four I turned from the context of underlying paradigms, education, organizational setting and roles to examine the meaning of genetic counselling for the individual. Virtually all genetic counselling professionals

underlined the importance of providing clients with accurate and up-to-date information on the genetic basis and medical implications of genetic conditions. This information is valuable for the client; genetic counsellors say the information aids the coping process by helping the client pass through the emotional response to genetic disease on their way to reaching an informed decision.

Although the importance and process of providing genetic and medical information was described to me in similar ways by all groups of genetic counselling professionals, dealing with the psychosocial aspects of client care showed considerable individual variation. Re-stated, there is widespread agreement that genetic counselling should include both medical/genetic and psychosocial care, but the extent to which genetic counsellors incorporate psychosocial factors and issues into their work is essentially individually based. In this, I was particularly interested in how physician geneticists, the central caregivers in genetic counselling, were dealing with the responsibility of incorporating both medical and psychosocial goals into their practice.

I argue that among the physician geneticists that I interviewed the extent to which they involve themselves in the psychosocial care of their clients is dependent, in part, upon the factors discussed in this thesis--paradigmatic pressures existing in genetic counselling, the counsellor's training, and the type of tasks they are performing. Additionally, the manner in which the genetic counsellors that I interviewed constructed their responsibility to clients was related to their view of the boundaries of medicine. Some physician geneticists draw the boundaries of medicine more narrowly than others thereby limiting their responsibilities to the provision of genetic and medical information. Others talk of the "art of medicine" as that quality which allows and encourages them to deal with their client's psychological, social, and emotional concerns.

Genetic counselling is well established as a medical service in a medical context in Canada. The "medicalization" of genetic counselling has meant physician geneticists have the central caregiving role in genetic counselling. But the criteria of adequate genetic care have changed in response to social demands, such that genetic counselling is now guided by two paradigms--one which continues to emphasize the biological basis and its medical implications of genetic disease, the other which stresses the psychosocial concomitants of that disease.

As the dominant practitioners of genetic counselling, physician geneticists face the difficult task of relating the two part of their work, the genetics and the counselling, although their training and the context in which they practise does not provide support for both parts. Genetic counselling professionals are thus guided by two conflicting principles of behaviour--"assistance" or "activism" directed towards helping their clients reach the goal of an informed decision and "non-directive counselling" which suggests clients should be free to make any decision that they wish. These two principles of action reflect the dichotomy of "genetics" and "counselling" which has arisen from the dynamic between the medicoscientific and psychosocial paradigms. The resolution for genetic counsellors then lies in the subtle act of "giving advice without telling people what to do."

REFERENCES CITED

Antley, Ray
1979 The genetic counselor as facilitator of the counselee's decision process. In Genetic Counseling: Facts, Values, and Norms. Edited by Alexander Capron *et al.* *Birth Defects (Original Article Series)* 15(2):137-168.

Applebaum, Eleanor and Stephen Firestein
1983 *A Genetic Counseling Casebook.* New York: The Free Press.

Arseneau, Richard *et al.*
1985 Examinations examined. *Med-Ed News* (University of Toronto, Faculty of Medicine) 2(2):2.

Association of Canadian Medical Colleges
1984 *Canadian Medical Education Statistics.* Ottawa: Association of Canadian Medical Colleges.

Baskin, Yvonne
1982 Spare Genes. *Omni* 53-54, 109-114.

Becker, Howard *et al.*
1961 *Boys in White: Student Culture in Medical School.* Chicago: University of Chicago Press

Black, Rita Beck
1982 Risk taking behaviour: decision making in the face of genetic uncertainty. *Social Work in Health Care* 7(1):11-25.

Brennan, James R.
1985 *Patterns of Human Heredity: An Introduction to Human Genetics.* Englewood Cliffs: Prentice-Hall.

Brewer, George and Charles Sing
1983 *Genetics.* Reading: Addison-Wesley.

Canadian Broadcasting Corporation
1986 *Drawing the line: reproductive technology.* Transcript from the Ideas Series (March 17, 24). Montreal: Canadian Broadcasting Corporation

Canadian College of Medical Geneticists
1984 Accreditation for Canadian College of Medical Geneticists (pamphlet).

Caplan, Arthur
1979 Genetic counseling, medical genetics, and theoretical genetics: an historical overview. In Genetic Counseling: Facts, Values, and Norms. Edited by A. Capron *et al. Birth Defects (Original Article Series)* 15(2):21-31.

Childs, Barton
1977 Persistent echoes of the nature-nuture argument. *American Journal of Human Genetics* 29:1-13.

1978 Genetic counseling: a critical review of the published literature. In *Genetic Issues in Public Health and Medicine.* Edited by B. Cohen *et al.*, pp. 329-357. Springfield: Charles Thomas Publishers.

Chrisman, Noel
1984 *Practical relativism in clinical settings.* Unpublished paper presented at the American Anthropological Association meetings. Denver, December 1984.

Clow, Carol
1982 Genetic services in Canada. *Progress in Clinical Biological Research* 103(B):389-398.

Cockerham, William
1982 *Medical Sociology*, second edition. Englewood Cliffs: Prentice-Hall.

Cohen, Felissa
1984 *Clinical Genetics in Nursing Practice.* Philadelphia: J.B. Lippincott.

Dagenais, Denyse L. *et al.*
1985 A cost-benefit analysis of the Quebec Network of Genetic Medicine. *Social Science and Medicine* 20(6): 601-607.

Davidson, R.G. and B. Childs
1986 Pre-publication draft of Perspectives in the teaching of human genetics. *Advances in Human Genetics* 16:79-119.

Davis, Jessica
1984 Communication skills for MD and PhD geneticists. In Genetic Disorders and Birth Defects in Families and Society: toward interdisciplinary understanding. Edited by J. Weiss *et al., Birth Defects(Original Article Series)* 20 (4):41-44.

Dodge, Wayne
1983 Communication and interpersonal skills. In *Fundamentals of Family Medicine.* Edited by R. Taylor, pp. 173-188. New York: Springer-Verlag.

Dolnick, Edward
1985 Fetus tests: collision course for America. *The Edmonton Journal* (May 19):A7

Downie, R. S.
1984 Ethics and surveys: response to 'Ethical dilemmas in clinical genetics.' *Journal of Medical Ethics* 10(2):77-78.

Doyal, Lesley
1979 *The Political Economy of Health.* London: Pluto Press.

Eisenberg, Leon and A. Kleinman
1981 Clinical social science. In *The Relevance of Social Science for Medicine.* Edited by L. Eisenberg and A. Kleinman, pp. 1-23. Dordrecht, Holland: D. Reidel.

Emery, Alan H.
1984 Introduction: the principles of genetic counselling. In *Psychological Aspects of Genetic Counselling.* Edited by A. Emery and I. Pullen, pp. 1-9. New York: Academic Press.

Engel, G.
1977 The need for a new medical model: a challenge for biomedicine. *Science* 196:129-136.

Epstein, Charles
1977 A position paper on position papers on the organization of genetic counseling. In *Genetic Counseling.* Edited by H. Lubs and F. de la Cruz, pp. 333-348. New York: Raven Press.

Evers-Kieboom, G. and H. van den Berghe
1979 Impact of genetic counseling: a review of published follow-up studies. *Clinical Genetics* 15:465-474.

Falek, Arthur
1984 Sequential aspects of coping and other issues in decision making in genetic counseling. In *Psychological Aspects of Genetic Counselling.* Edited by A. Emery and I. Pullen, pp. 23-36. New York: Academic Press.

Fletcher, John , Dorothy Wertz, James Sorenson
1985 International Survey of Medical Geneticists (Questionnaire)

Fox, Renee C.
1957 Training for uncertainty. In *The Student Physician.* Edited by R. Merton, G. Reader, and P. Kendall, pp. 207-241. Cambridge: Harvard University.

Frankenberg, Ronald
1980 Medical anthropology and development: a theoretical perspective. *Social Science and Medicine* 14B:197-207.

Fraser, F. Clarke
1974 Genetic counseling. *American Journal of Human Genetics* 26:636-659.

1979 Introduction: The development of genetic counseling. In Genetic Counseling: Facts, Values, and Norms. Edited by A. Capron *et al., Birth Defects (Original Article Series)* 15(2): 5-15.

Friedson, Elliot
1970 *Profession of Medicine.* New York: Dodd, Mead.

Gaines, Atwood and Robert Hahn
1985 Among the physicians: encounter,exchange, and transformation. In *Physicians of Western Medicine.* Edited by R. Hahn and A. Gaines, pp. 3-22. Dordrecht, Holland: D. Reidel.

Giddens, Anthony
1976 *New Rules of Sociological Method.* NewYork: Basic Books.

Gould, Stephen Jay
1981 *The Mismeasure of Man.* New York: W. Norton Co.

Hahn, Robert
1985 A world of internal medicine: portrait of an internist. In *Physicians of Western Medicine.* Edited by R.A. Hahn and A.D. Gaines, pp. 51-111. Dordrecht, Holland: D. Reidel.

Hahn, Robert and Arthur Kleinman
1982 Biomedicine as a cultural system. Prepared for Dr. Massimo Piatelli-Palmarini, General Editor, *Social History of the Biomedical Sciences.* Mss.

1983 Biomedical practice and anthropological theory. *Annual Review of Anthropology* 12:305-333.

Harper, Peter S.
1981 *Practical Genetic Counselling.* Bristol: John Wright and Sons [reprinted 1984].

Hsia, Y. Edward
1979 The genetic counselor as information giver. In Genetic Counseling: Facts, Values, and Norms. Edited by A. Capron *et al.*, *Birth Defects (Original Article Series)* 15(2):169-186.

Kelly, Patricia T.
1977 *Dealing with Dilemma: a Manual for Genetic Counselors.* New York: Springer-Verlag.

Kenen, Regina
1984 Genetic counseling: the development of a new interdisciplinary occupational field. *Social Science and Medicine* 18(7):541-549.

Kessler, Seymour
1979 The genetic counselor as psychotherapist. In Genetic Counseling: Facts, Values, and Norms. Edited by A. Capron *et al.*, *Birth Defects (Original Article Series)* 15(2):187-200.

1980 The psychological paradigm shift in genetic counseling. *Social Biology* 27:167-185.

Kevles, Daniel J.
1985 *In the Name of Eugenics: Genetics and the Uses of Human Heredity*. New York: Alfred Knopf.

Kleinman, Arthur
1980 *Patients and Healers in the Context of Culture.* Berkeley: University of California Press.

Lippman-Hand, A. and F. Clarke Fraser
1979a Genetic counseling: the postcounseling period. I. Parents' perceptions of uncertainty. *American Journal of Medical Genetics* 4:51-71.

1979b Genetic counseling: the postcounseling period. II. Making reproductive choices. *American Journal of Medical Genetics* 4:73-87.

Lock, Margaret
1985 Models and pratice in medicine: menopause as syndrome or life transition? In *Physicians of Western Medicine.* Edited by R.A. Hahn and A.D. Gaines, pp. 51-111. Dordrecht, Holland: D. Reidel.

Lynch, H.T. *et al.*
1983 *International Directory of Genetic Services.* White Plains: March of Dimes Birth Defects Foundation.

Macklin, Madge T.
1938 The inheritance of disease and its relation to the practice of medicine. *The Medical Woman's Journal* 454 (4):89-96.

Maretzki, Thomas
1985 Including the physician in healer-centered research: retrospect and prospect. In *Physicians of Western Medicine.* Edited by R. Hahn and A. Gaines, pp. 23-47. Dordrecht, Holland: D. Reidel.

Marks, Joan
1984 The genetic counselor. In Genetic Disorders and Birth Defects in Families and Society: Toward interdisciplinary understanding. Edited by J. Weiss *et al., Birth Defects (Original Article Series)* 20(4):22-24.

McGill University
1984 Centre for Human Genetics: a two year training program leading to a diploma in genetic counselling (pamphlet). Montreal: McGill University.

McKusick, Victor A.
1983 *Mendelian Inheritance in Man: Catalogs of Autosomal Dominant, Autosomal Recessive, and X-linked Phenotypes.* Baltimore: The John Hopkins University Press.

Mealey, Linda
1984 Decision making and adjustment in genetic counseling. *Health & Social Work* 9(2):124-133.

Meier, Robert
n.d. Anthropological evaluation of genetic services. Unpublished manuscript. Deptartment of Anthropology. Cleveland: Case Western Reserve University.

Mertens, Thomas, Jon Hendrix and Kathleen Kenkel
1986 The genetic associate: a career option in genetic counselling. *The Journal of Heredity* 77:175-178.

Miller, James
1975 Canadian College of Medical Genetics. *Canadian Medical Association Journal* 113:358.

Miller, Judith R.
1980 A Survey of Genetic Services in Canada. *Social Issues in Human Genetics: Genetic Screening and Counselling,* pp. 25-35 and Appendix pp. 104-107. Ottawa: Science Council of Canada.

Murray, Robert
1976 Psychosocial aspects of genetic counseling. *Social Work in Health Care* 2(1):13-23.

1984 Cultural and ethnic influences on the genetic counselling process. In Genetic Disorders and Birth Defects in Families and Society: Toward interdisciplinary understanding. Edited by J. Weiss *et al.*, *Birth Defects (Original Article Series)* 20(4): 71-74.

Norusis, Marija
1983 *SPSSx: Introductory guide to statistics.* New York: McGraw-Hill.

Oetting, L.A. and M.W. Steele
1982 A controlled retrospective follow up study of the impact of genetic counseling on parental reproduction following the birth of a Down Syndrome child. *Clinical Genetics* 21:7-13.

Olby, Robert
1985 *Origins of Mendelism.* Chicago: University of Chicago Press.

Ontario Council of Health
1976 *Genetic Services: Report of the Ontario Council of Health.* Toronto: Ontario Council of Health.

Pelto, Perti
1970 *Anthropological Research: The structure of inquiry.* New York: Harper and Row.

President's Commission for the Study of Ethical Problems in Medical and Biomedical Research and Behavioral Research
1983 *Screening and Counseling for Genetic Conditions.*

Rainer, John D.
1979 Psychiatric considerations in genetic counseling. In *Service and Education in Medical Genetics.* Edited by I. Porter and E. Hook, pp. 295-301. New York: Academic Press.

Rapp, Rayna
1985 Moral pioneers: women, men and fetuses on a frontier of reproductive technology. Paper presented at the Annual Meeting of the American Anthropological Association. Washington, DC, December 1985.

1986 Translating the genetic code: the discourse of genetic counselling. Paper presented at the American Ethnological Society Meetings, April 1986.

Reed, Sheldon C.
1974 A short history of genetic counseling. *Social Biology* 21(4):332-339.

Reif, Maria and Helmut Baitsch
1985 Psychological issues in genetic counselling. *Human Genetics* 70:193-199.

Reilly, Philip
1979 Professional identification: issues in licensing and certification. In, Genetic Counselling: Facts, Values and Norms. Edited by A. Capron *et al.*, *Birth Defects: Original Article Series* 15(2): 291-305.

Ross, James
1984 Research proposal on clients' a) reproductive and contraceptive behaviour b) perception of decision-making process. Pediatrics Department. Cleveland: Case Western Reserve University.

Rudd, Noreen
1980 Genetics: The state of the art in Canada. *Social Issues in Human Genetics: Genetic Screening and Counselling*, pp. 11-15. Ottawa: Science Council of Canada.

Sanson-Fisher, R. and P. Maguire
1980 Should skills in communicating with patients be taught in medical schools? *Lancet* (Sept 6):523-526.

Science Council of Canada
1980 *Social Issues in Human Genetics: Genetic Screening and Counselling.* Ottawa: Science Council of Canada.

Scriver, C.R.
1977 Conference on teaching clinical genetics. In Trends and Teaching in Clinical Genetics. Edited by D. Bergsma *et al.*, *Birth Defects (Original Article Series)* 13(6):187-197.

Scriver, C.R., C. Laberge, C. Clow, and F.C. Fraser
1978 Genetics and medicine: an evolving relationship. *Science* 200:946-952.

Shaw, Margery
1977 Preview of published studies of genetic counseling: a critique. In *Genetic Counseling.* Edited by H. Lubs and F. de la Cruz, pp. 35-52. New York: Raven Press.

Silverberg, Ruth and Lynn Godmilow
1979 The process of genetic counseling. In *Counseling in Genetics.* Edited by Edward Hsia, pp. 281-293. New York: Alan R. Liss.

Soltan, Hugh

1967 Madge Macklin: pioneer in medical genetics. *University of Western Ontario Medical Journal* 38:6-11.

1985 Medical genetics at Western: a personal historical view. *University of Western Ontario Medical Journal* (Jan).

Sorenson, James

1979 Family physician, public health advocate, or eugenicist: some attitudes and values of American genetic counselors. In *Service and Education in Medical Genetics*. Edited by Ian Porter and E. Hook, pp. 273-284. New York: Academic Press.

Sorenson, James and Arthur Culbert

1977 Genetic counselors and counseling orientations--unexamined topics in evaluation. In *Genetic Counseling*. Edited by H. Lubs and F. de la Cruz, pp. 131-156. New York: Raven Press.

1979 Professional orientations to contemporary genetic counseling. In Genetic Counseling: Facts, Values and Norms. Edited by A. Capron *et al.*, *Birth Defects (Original Article Series)* 15(2):85-102.

Sorenson, James *et al.*

1981 Reproductive pasts, reproductive futures: genetic counseling and its effectiveness. Edited by A. Capron *et al., Birth Defects (Original Article Series)* 17(4).

Stein, Howard

1986 'Sick People' and 'Trolls': a contribution to the understanding of the dynamics of physician explanatory models. *Culture, Medicine and Psychiatry* 10 (3):221-229.

Steltz-Lenarsky, J.S., G.N. Mahnke and Robert Meier

1985 Anthropological evaluation of genetic counseling: preliminary report of interview data. Paper presented at the 54th American Association of Physical Anthropology Meetings, Knoxville, Tenn. Abstract in *American Journal of Physical Anthropology* 66(2):234

Stephenson, S.R. and D.D. Weaver

1981 Prenatal diagnosis--a compilation of diagnosed conditions. *American Journal of Obstetrics and Gynecology* 141(3): 319-343.

Swanson, August
1984 Medical education in the United States and Canada. In, Physicians for the Twenty-first Century. Report of the Project Panel on the General Professional Education of the Physician and College Preparation for Medicine. Association of American Medical Colleges. *Journal of Medical Education* 59 (11[Supplement]):35-56.

Thompson, James and Margaret Thompson
1986 *Genetics in Medicine*, fourth edition. Revised by M. Thompson. Philadelphia: Saunders.

Twiss, Sumner
1979 The genetic counselor as moral advisor. In Genetic Counseling: Facts, Values and Norms. Edited by A. Capron *et al.*, *Birth Defects (Original Article Series)* 5(2):201-212.

Wright, Peter and Andrew Treacher
1982 Introduction. In *The problem of medical knowledge: examining the social construction of medicine.* Edited by P. Wright and A. Treacher, pp. 1-22. Edinburgh: Edinburgh University Press.

Young, Allan
1981 When rational men fall sick: an inquiry into some assumptions made by medical anthropologists. *Culture, Medicine, and Psychiatry* 5:317-35.

1982 The anthropologies of illness and sickness. *Annual Review of Anthropology* 11:257-285.

Zare, Nancy *et al.*
1984 Sex of provider as a variable in effective genetic counseling. *Social Science and Medicine* 19(7):671-675.

APPENDIX ONE

Questionnaire

QUESTIONNAIRE

No._____

I AM INTERESTED IN YOUR OPINIONS ABOUT PROVIDING GENETIC COUNSELLING. PLEASE ANSWER ALL THE QUESTIONS EVEN IF YOUR PROFESSIONAL DUTIES DO NOT INCLUDE CLIENT CONTACT.

PLEASE DO NOT WRITE YOUR NAME ON THIS QUESTIONNAIRE. ALL QUESTIONNAIRES WILL BE NUMERICALLY CODED.

I AM AWARE OF AND WILL UPHOLD MY RESPONSIBILITY TO ENSURE THAT THE PERSONAL AND PROFESSIONAL INTEGRITY AND THE ANONYMITY OF ALL PARTICIPANTS IN THIS RESEARCH IS PROTECTED.

PART ONE: SOCIO-DEMOGRAPHIC DATA

1.Age ( ) under 20 years = 1
( ) 21 to 30 years = 2
( ) 31 to 40 years = 3
( ) 41 to 50 years = 4
( ) 51 to 60 years = 5
( ) over 61 years = 6

2. Sex ( ) Male = 1 ( ) Female = 2

3. ( ) Married = 1 ( ) Unmarried = 2

4. Children ( ) Yes =1 ( ) No = 2

5. Religion ( ) Protestant = 1
( ) Roman Catholic = 2
( ) Jewish = 3
( ) Muslim = 4
( ) Hindu = 5
( ) Buddhist = 6
( ) Atheist = 7
( ) Other = 8

6. Current Occupation _____

7. How long have you been involved in genetic counselling?
( ) under 1 year = 1
( ) 1 to 5 years = 2
( ) 6 to 10 years = 3
( ) 11 to 15 years = 4
( ) 16 to 20 years = 5
( ) 21 to 25 years = 6
( ) over 25 years = 7

No._____

8. Degrees Held (please check as many as apply and indicate the year degree was awarded):
   ( ) M.D.(specify):
      ( ) Pediatrics; in 19____
      ( ) Obstetrics-Gynecology; in 19____
      ( ) Pathology; in 19____
      ( ) Other (specify)_____;in 19____

   ( ) PhD.(specify field)_____;in 19____
   ( ) Masters degree (specify field):_____;in 19____
   ( ) Genetics Associate (Canada); in 19____
   ( ) Genetics Counselor (U.S.A.); in 19____
   ( ) R.N.(specify field):_____;in 19____
   ( ) Other (specify field & degree):_____;in 19____
   ( ) Student(specify field and year of graduation above)
   ( ) Fellow (specify field and year of completion above)

9. What training have you had in human genetics?
   ( ) Course(s) in genetics
   ( ) Supervised clinical training
   ( ) Other (specify):_____
   ( ) None

10. What training have you had in counselling methods?
   ( ) Course(s) in counselling
   ( ) Training in a supervised clinic
   ( ) Other (specify):_____
   ( ) None

PART TWO: PROFESSIONAL ACTIVITIES

11. Approximately how many hours each week do you spend on the following counselling-related activities?

All counselling-related activities	_____hours/week
Seeing patients	_____hours/week
Clerical and administrative duties	_____hours/week
Laboratory work	_____hours/week
Meeting with counselling staff	_____hours/week
Other (specify)_____	_____hours/week

12. Regarding your professional activites outside of counselling, how many hours each week do you spend on the following?

Research	_____hours/week
Practice	_____hours/week
Teaching	_____hours/week
Administration	_____hours/week
Other (specify):_____	_____hours/week

13. Approximately how many clients do you counsel each week? _____

14. Please indicate as many of the following that are part of your usual activities. (Circle the number)

A. Administrative
1 Schedule appointments
2 Obtain medical records
3 Assign patients to counsellors
4 Co-ordinate clinic activities

B. Counselling
1 Take patient/family social history
2 Genetic counselling
3 Amniocentesis counselling
4 Counselling for family problems
5 Counselling for patient emotional problems
6 Suggest resources for financial problems
7 Refer to specialists
8 Follow up to counselling visit(s) (phone, mail, or in person)

C. Diagnostic
1 Take pedigree
2 Take medical history
3 Physical examination
4 Refer for diagnostic tests
5 Laboratory work
6 Make diagnosis
7 Perform amniocentesis
8 Participate in genetic screening program

D. Patient Management
1 Provide medical treatment
2 Provide information for longterm management
3 Refer to a parent's group
4 Refer to a social or educational agency
5 Refer to specialists
6 Follow up

E. Other
1 Participate in outreach programme
2 Provide education for professionals
3 Provide education for public
4 Participate in parents' and/or patient self-help groups

15. Is there anything you would like to add to your regular routine?
( ) No = 1 ( ) Yes = 2, please specify

---

16. Of your regular tasks are there any you feel should not be part of your job? ( ) None = 1 ( ) Yes = 2, please specify

---

---

17. At your present place of practice, how frequently are the following professionals directly involved (having contact with the client) in genetic counselling:

	All cases	Over 50% of cases	Less than 50% of cases	Never
a)Medical doctor	( )	( )	( )	( )
b)PhD geneticist	( )	( )	( )	( )
c)Genetics associate	( )	( )	( )	( )
d)Registered nurse	( )	( )	( )	( )
e)Social worker	( )	( )	( )	( )
f)Member of the clergy	( )	( )	( )	( )
g)Ethicist	( )	( )	( )	( )

PART THREE: GOALS AND DEFINITIONS

18. Professionals in genetic counselling may hold one or more goals for their counselling. In your opinion, how important is it that your counselling achieve each of the following.(Please circle)

a) The prevention of disease or abnormality.
1 Very important
2 Important
3 Somewhat important
4 Not at all important

b) The removal or lessening of client anxiety.
1 Very important
2 Important
3 Somewhat important
4 Not at all important

c) A reduction in the number of carriers of genetic disorders in the population.
1 Very important
2 Important
3 Somewhat important
4 Not at all important

d) Provide clients with the medical and genetic information to deal with their genetic problems.
1 Very important
2 Important
3 Somewhat important
4 Not at all important

No._____

19. In your opinion, how appropriate is it for you as a professional in genetic counselling to do each of the following.(Please circle)

a) Tell clients that decisions are theirs alone and refuse to make any decisions for them.
   1 Always appropriate
   2 Sometimes appropriate
   3 Rarely appropriate
   4 Never appropriate

b) Suggest that while you will not make decisions for clients you will support any that they make.
   1 Always appropriate
   2 Sometimes appropriate
   3 Rarely appropriate
   4 Never appropriate

c) Inform clients what most other people in their situation have done
   1 Always appropriate
   2 Sometimes appropriate
   3 Rarely appropriate
   4 Never appropriate

d) Inform clients what you would do if you were in their situation.
   1 Always appropriate
   2 Sometimes appropriate
   3 Rarely appropriate
   4 Never appropriate

e) Advise clients what they ought to do.
   1 Always appropriate
   2 Sometimes appropriate
   3 Rarely appropriate
   4 Never appropriate

f) Ensure clients have made a decision they feel they can live with.
   1 Always appropriate
   2 Sometimes appropriate
   3 Rarely appropriate
   4 Never appropriate

g) Correct 'misinformation' in your client's explanation for the genetic disorder.
   1 Always appropriate
   2 Sometimes appropriate
   3 Rarely appropriate
   4 Never appropriate

h) Determine the source of 'misinformation' and attempt to point out the error to the individual responsible.
   1 Always appropriate
   2 Sometimes appropriate
   3 Rarely appropriate
   4 Never appropriate

20. Please indicate the extent to which you, as a professional in genetic counselling, agree or disagree with the following statements:

a) You have a professional responsibility to determine your client's ideas, expectations, and attitudes about genetic counselling.
   1 Strongly agree
   2 Agree
   3 Disagree
   4 Strongly disagree

b) You have a professional reponsibility to help your clients understand the genetic and medical facts which pertain to their genetic disorder including the diagnosis, relevant principles of inheritance, recurrence risk, prognosis, and available treatment.
   1 Strongly agree
   2 Agree
   3 Disagree
   4 Strongly disagree

c) You have a professional responsibility to determine the nature of your client's explanation for the genetic disorder.
   1 Strongly agree
   2 Agree
   3 Disagree
   4 Strongly disagree

d) You have a professional responsibility to help your clients choose the course of action which seems appropriate to them in view of their family goals and the information you have provided.
   1 Strongly agree
   2 Agree
   3 Disagree
   4 Strongly disagree

e) You have a professional responsibility to help your clients choose the course of action that seems appropriate to them in view of their explanation of the genetic disorder and their family goals.
   1 Strongly agree
   2 Agree
   3 Disagree
   4 Strongly disagree

f) You have a professional responsibility to help your clients act in accordance with their decision regardless of your opinion of the basis of the decision.
   1 Strongly agree
   2 Agree
   3 Disagree
   4 Strongly disagree

g) You have a professional responsibility to ensure your client is satisfied with the counselling session.

1 Strongly agree
2 Agree
3 Disagree
4 Strongly disagree

21. Listed below are several issues that can be involved in counselling for genetic disorders. For each issue, please indicate how you prefer to include them in your counselling session. (Please circle the appropriate number)

a) Effects of genetic disorder on client's family life.

1 Allow clients to raise this issue themselves and then discuss it with them.
2 Create an opportunity for clients to raise this issue and, when they do, discuss it with them.
3 Confront clients with this issue, and then discuss it with them.
4 If clients raise this issue refer them to the following individual for discussion..... ( )Family member
( )Referring physician
( )Member of the clergy
( )Other, _____

b) Effect of genetic disorder on client's sexual life.

1 Allow clients to raise this issue themselves and then discuss it with them.
2 Create an opportunity for clients to raise this issue and, when they do, discuss it with them.
3 Confront clients with this issue, and then discuss it with them.
4 If clients raise this issue refer them to the following individual for discussion..... ( )Family member
( )Referring physician
( )Member of the clergy
( )Other, _____

c) Genetic disorder as a reason for limiting family size.

1 Allow clients to raise this issue themselves and then discuss it with them.
2 Create an opportunity for clients to raise this issue and, when they do, discuss it with them.
3 Confront clients with this issue, and then discuss it with them.
4 If clients raise this issue refer them to the following individual for discussion..... ( )Family member
( )Referring physician
( )Member of the clergy
( )Other, _____

d) Methods of contraception.

1 Allow clients to raise this issue themselves and then discuss it with them.
2 Create an opportunity for clients to raise this issue and, when they do, discuss it with them.
3 Confront clients with this issue, and then discuss it with them.
4 If clients raise this issue refer them to the following individual for discussion..... ( )Family member
( )Referring physician
( )Member of the clergy
( )Other, _____

e) Alternate forms of parenthood, including adoption and artificial insemination.

1 Allow clients to raise this issue themselves and then discuss it with them.
2 Create an opportunity for clients to raise this issue and, when they do, discuss it with them.
3 Confront clients with this issue, and then discuss it with them.
4 If clients raise this issue refer them to the following individual for discussion..... ( )Family member
( )Referring physician
( )Member of the clergy
( )Other, _____

f) Providing information for family members who may be at risk.

1 Allow clients to raise this issue themselves and then discuss it with them.
2 Create an opportunity for clients to raise this issue and, when they do, discuss it with them.
3 Confront clients with this issue, and then discuss it with them.
4 If clients raise this issue refer them to the following individual for discussion..... ( )Family member
( )Referring physician
( )Member of the clergy
( )Other, _____

g) Client's religious views.

1 Allow clients to raise this issue themselves and then discuss it with them.
2 Create an opportunity for clients to raise this issue and, when they do, discuss it with them.
3 Confront clients with this issue, and then discuss it with them.
4 If clients raise this issue refer them to the following individual for discussion..... ( )Family member
( )Referring physician
( )Member of the clergy
( )Other, _____

h) Economic cost of genetic disorder to client.

1 Allow clients to raise this issue themselves and then discuss it with them.
2 Create an opportunity for clients to raise this issue and, when they do, discuss it with them.
3 Confront clients with this issue, and then discuss it with them.
4 If clients raise this issue refer them to the following individual for discussion..... ( )Family member
( )Referring physician
( )Member of the clergy
( )Other, _____

i) Social stigma of disorder.

1 Allow clients to raise this issue themselves and then discuss it with them.
2 Create an opportunity for clients to raise this issue and, when they do, discuss it with them.
3 Confront clients with this issue, and then discuss it with them.
4 If clients raise this issue refer them to the following individual for discussion..... ( )Family member
( )Referring physician
( )Member of the clergy
( )Other, _____

22. Do you use the term "genetic counselling" to describe what you do?
( ) Yes
( ) No. I prefer the following term (please indicate why):

_____

_____

_____

_____

No._____

PART FOUR: HYPOTHETICAL CASES
This section includes descriptions of four hypothetical counselling sessions. Please read each case and answer the questions which follow each description.

CASE ONE

A 38 year old woman, now 17 weeks pregnant, was referred by her general practitioner for amniocentesis. Chromosomal analysis indicates Trisomy 21 and the woman and her husband have come for their first meeting with the cytogeneticist. The cytogeneticist explains the test results to the woman and her husband both of whom are silent throughout the explanation. The cytogeneticist tells the couple about the mental development and associated health problems of children with Down Syndrome. He asks them if they have ever thought about raising a mentally handicapped child. The wife shakes her head, but the couple remain quiet. The cytogeneticist gives the couple a pamphlet from a local Down Syndrome parental support group and suggests they contact the group. The couple thank the cytogeneticist and depart.

23. What do you feel are key issues in this session?

---

24. Would you like to make any suggestions to the counsellor?
( ) No = 1
( ) Yes = 2. I would suggest the following:

No._____

CASE TWO

A young couple has come to see a medical geneticist after learning that the wife's sister's child has been diagnosed as having cystic fibrosis.The couple is expecting a child (the woman is 14 weeks pregnant) and they are extremely anxious about the health of their own unborn child. The medical geneticist takes a family history, but information is scanty. He explains that there is no way of making a pre-natal diagnosis and that he cannot determine whether or not they are carriers. The couple is visibly upset by this information and repeatedly ask the medical geneticist,"What should we do?" The geneticist listens attentively to their concerns. He tells them he can provide them with information, but only they can decide how to use the information. The couple becomes angry with the doctor. The doctor suggests they see the hospital social worker to help them deal with their anger. The couple departs saying they expected to get help and advice from the geneticist.

25. What do you feel are key issues in this session?

---

26. Would you like to make any suggestions to the counsellor?
( ) No = 1
( ) Yes = 2. I would suggest the following:

No._____

CASE THREE

A pregnant woman is sent for genetic counselling when a neural tube defect, suspected by alpha fetoprotein levels, is confirmed by ultrasound. She has been told her unborn child has a large encephalocele. She is obviously shocked by this information and, weeping, holds her hands over her ears while the geneticist explains the infant will probably not live long after birth. She asks the geneticist not to tell her anything more . "What is the point of knowing the baby will die ?" she asks. The geneticist explains the options available to her. The woman explains that when her obstetrician told her that "there might be something wrong with the baby", she told him she didn't care about that, but she just wanted her baby. But she came because her obstetrician said he wanted her to see a "specialist for tests". She tells the geneticist she thought the tests would help the baby.

27. What do you feel are key issues in this session?

---

28. Would you like to make any suggestions to the counsellor?
( ) No = 1
( ) Yes = 2. I would suggest the following:

CASE FOUR

A forty year old man, diagnosed as having polycystic kidney disease, has been referred for genetic counselling by his family physician. The geneticist explains the relevant principles of autosomal dominant inheritance and asks about the client's family. The man says he has two young daughters. The geneticist asks the man if he will tell his wife about the diagnosis. The man is startled by this. He shakes his head and says he must bear the responsibility alone. The geneticist asks the man what he means by this. After a long pause the man tells the geneticist he knows he is being punished for being unfaithful to his wife.

29. What do you feel are key issues in this session?

---

30. Would you like to make any suggestions to the counsellor?
( ) No = 1
( ) Yes = 2. I would suggest the following:

No._____

PART FIVE: COMMENTS ON GENETIC COUNSELLING

Many people -- psychologists, sociologists, clergymen, medical doctors, and geneticists among others -- have written about counselling for genetic disorders. In this section I am interested in your opinion about some of their comments. For each comment please indicate the extent of your agreement by circling the appropriate number. Space is provided after each passage for you to add your own comments.

31. It is the genetic counsellor's responsibility to ensure clients reach a decision feel they can live with.
    1 Strongly agree
    2 Agree
    3 Disagree
    4 Strongly disagree

Comment:_____

_____

_____

_____

_____

32. The methodology of genetic counselling is still experimental in that most counselors know very little about helping clients to make sense of genetic information.
    1 Strongly agree
    2 Agree
    3 Disagree
    4 Strongly disagree

Comment:_____

_____

_____

_____

_____

No._____

33. Genetic counselling should not be within the practice of medicine.
    1 Strongly agree
    2 Agree
    3 Disagree
    4 Strongly disagree

Comment:_____

_____

_____

_____

_____

34. Professionals in genetic counselling have a responsibility to clarify their personal values for the client.
    1 Strongly agree
    2 Agree
    3 Disagree
    4 Strongly disagree

Comment:_____

_____

_____

_____

_____

THANK YOU !

If you have any additional comments you would like to make please use the following space and the back of this page.

Please note:
Questions 8-16, 18 (a,b,c) and 19(a to e) are adapted from the National Foundation-March of Dimes Genetic Counseling Study Questionnaire for Counselors reproduced in Sorenson et al (1981: 147-151).