COUNSELLING OF AGE-RELATED RISKS AND PRENATAL DIAGNOSIS: AN OVERVIEW OF COMMUNITY AND MEDICAL GENETICS COUNSELLING

by

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Abstract

The effectiveness of genetic counselling regarding risks associated with advanced maternal age (AMA) and prenatal diagnosis (PND) was assessed in three groups of women; those with AMA only who are counselled by their primary care physician or obstetrician within the community (AO, N=311), those with AMA plus a minor concern(s) who are counselled by genetic counsellors (AP, N=52), and those with AMA as well as complex indications for prenatal diagnosis such that they are counselled by genetic counsellors and medical geneticists (AC, N=36).

Subjects were asked to complete two questionnaires. Patients in AO completed the first questionnaire (Q1) after receiving counselling from their primary care physician in the community and before having a prenatal diagnostic procedure. Patients in AP and AC completed Q1 at the Medical Genetics clinic immediately before their genetic counselling. Patients in all three groups completed the second questionnaire (Q2) either immediately after their procedure before leaving the hospital or within four weeks post-procedure, prior to receiving their test results. The questionnaires were designed to look at subjects' knowledge of the information normally presented in AMA counselling and to assess the emotional responses of women regarding their involvement with PND.

Patients in all three groups were more informed in Q1 and Q2 regarding risks associated with having a procedure than risks associated with a chromosomal abnormality. While a majority of the women in each group said that they had been told the risk estimates requested of them, the number of women in each group who subsequently reproduced these figures was less than a majority. Finally, patients in all three groups demonstrated a decrease in anxiety once the procedure was complete.

The effectiveness of the genetic counselling process does not appear to be related to those providing the genetic counselling or the patient's ability to recall factual information.

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Part A

Introduction

Genetic counselling regarding age-related risks for having a child with a chromosomal abnormality is a service many women wish to have. The changing roles of women have caused the maternal age distribution at time of pregnancy to change. According to Vital Statistics Canada for the Province of British Columbia (B.C.), the percentage of total livebirths to women aged 35 and over has increased from 4.4% in 1978 to 9.6% in 1988. Although the number of total births per year has not changed, the number of women giving birth at a later age has. A major contribution to this statistic is the "baby boom" which occurred in the middle of this century (Adams, Oakley, & Marks, 1982; Hansen, 1986). As the women born in this time period approach middle age (35 and over) in the 80s and 90s, the percentage of births to women of advanced maternal age is increasing. Several reasons for this delayed childbearing have been discussed (Lehmann & Chism, 1987; Robinson, Garner, Gare, & Crawford, 1987; Sjogren & Uddenberg, 1990), such as changing professional or personal goals, financial stability, late acquaintance of the right partner, and the wish to be free of responsibility. Irrespective of the effects this trend is imposing from a social point-of-view, this shift in the maternal age distribution has important medical implications.

It is well documented that women of advanced maternal age have an increased risk of having a child with Down's

syndrome and other chromosomal abnormalities (Hook, 1981; Simpson et al., 1976). As a result, prenatal diagnostic services have become available to women worldwide, although the age limit and utilization varies between and within countries (Baird, Sadovnick, & McGillivray, 1985; Sjogren & Uddenberg, 1988). In 1985, Baird et al. reported on the overall utilization of amniocentesis in B.C. for prenatal diagnosis by women of advanced maternal age. The study revealed that between 1976-1983, approximately one-out-of-three women aged 38 and over at delivery chose to have prenatal diagnosis, with evidence that utilization would continue to increase. The most common prenatal diagnostic techniques provided to women include amniocentesis and chorionic villus sampling (CVS), both of which are carefully guided by ultrasound scan. Acceptability and attitudes of women towards the procedures available have been studied elsewhere (McGovern, 1986; Sjogren & Uddenberg, 1989; Spencer & Cox, 1987, 1988), and showed a growing reliance and use of CVS due to the advantage of earlier sampling.

One of the most important components of a prenatal diagnostic service is the provision of genetic counselling. Its basic functions which include informing the patient regarding prenatal diagnosis and her indication for it, delivering all relevant information in a non-directive manner, and providing support and autonomy at all times are

dependent upon the counsellor's ability to communicate with patients. As the field of genetic counselling grows with an increasing demand for prenatal diagnosis, evaluations of the effectiveness of genetic counselling are necessary to verify that the service is performing its functions adequately and according to the needs of the patients requesting prenatal diagnosis (Emery, 1984; Evers-Kiebooms & van den Berghe, 1979; Frets & Niermeijer, 1990; Kessler, 1990; Somer, Mustonen, & Norio, 1988). Chapter One reviews the impact of genetic counselling for prenatal diagnosis.

A prerequisite to an informed decision to undergo any screening or diagnostic test is knowledge about the test. This acquired knowledge and subsequent decision whether or not to have the test is dependent, in part, upon the counselling given to the patient and her ability to give a truly informed consent (Annas & Elias, 1990; Bernhardt, 1989; Cassileth, Zupkis, Sutton-Smith, & March, 1980; Marteau, Johnston, Plenicar, Shaw, & Slack, 1988). The assessment of what constitutes an informed decision has been reviewed extensively in the literature (Kessler, 1990; Lippman-Hand & Fraser, 1979; Sjogren & Marsk, 1989; Sjogren & Uddenberg, 1988; Wertz, Sorenson, & Heeren, 1986; Sorenson, Swazey, & Scotch, 1981), although different measures have been used. Chapter Two examines how women experience the genetic counselling offered to them with respect to knowledge and

perception of the information presented.

The decision to have prenatal testing is one that women should make once all relevant information has been provided. Autonomy is very important in genetic counselling for prenatal diagnosis, from the patient and the counsellor's point-of-view (Sjogren & Marsk, 1989). With rapidly-expanding capabilities in prenatal diagnosis and treatment, Annas and Elias (1990) report on the legal and ethical implications of fetal diagnosis on behalf of the individuals providing the information to patients. The purpose of genetic counselling, in addition to delivering information, is to help families make their own decisions. Furthermore, the counsellor should tailor each counselling session to the needs of the individual patient, as not all patients will share the same concerns and understanding regarding prenatal diagnosis (Bernhardt, 1989). Investigations of the factors involved in a woman's decision to have prenatal diagnosis (Bernhardt, 1989; Murray et al., 1980; Sjogren & Marsk, 1989; Sjogren & Uddenberg, 1988) have aided those providing the genetic counselling by allowing a better understanding of the decision-making process encountered by women. Chapter Three discusses the elements of decision-making as presented in the clinical genetics literature, with particular reference to the decisions regarding prenatal diagnosis for advanced maternal age.

Knowledge and understanding of prenatal diagnosis are most certainly important criteria for informed consent. Several studies have reviewed whether or not electing to have a procedure is solely due to the information discussed in genetic counselling (Sjogren & Uddenberg, 1990; Wertz & Sorenson, 1986). These studies and other research on the emotional responses of women encountering prenatal diagnosis indicate that the decision to have prenatal diagnosis is also dependent upon psychosocial factors (Silvestre & Fresco, 1980; Sjogren & Uddenberg, 1989; Thomassen-Brepols, 1987; Tunis et al., 1990). Chapter Four examines the reaction of patients to genetic counselling, demonstrating how the emotional response plays an essential role in the decision-making process regarding prenatal diagnosis.

Chapter One

Assessing the Effectiveness of the Genetic Counselling Process

Measuring the effectiveness of genetic counselling is a difficult task because of the multiple goals of the counselling process and the various criteria for defining what makes it "effective." Genetic counselling is a communication process in which the objectives and expectations of both the counsellor and the patient are not always the same. For this reason, the effectiveness of genetic counselling can be evaluated through either the counsellor or the patient's point-of-view. Furthermore, variation among counsellors regarding methods of practice and among patients with respect to their situation and reasons for seeking genetic counselling are additional factors that need to be taken into account when measuring the efficacy of such a complex medical service. Over the past decade, there have been numerous studies on the effectiveness of genetic counselling (Evers-Kiebooms & van den Berghe, 1979; Griffin, Kavanagh, & Sorenson, 1976, 1977; Kessler, 1980; Shiloh, Avdor, & Goodman, 1990; Sorenson et al., 1981; Wertz & Fletcher, 1988) which demonstrate the different approaches used by counsellors, and, subsequently, the different responses of patients to these methods.

The basic purpose of genetic counselling is to provide

accurate and appropriate information so that it will facilitate decision-making by the patient (Davies, 1983). Early investigations in the effectiveness of this process concentrated mostly on the level of patient medical-genetic knowledge post-counselling (Evers-Kiebooms & van den Berghe, 1979; Griffin et al., 1977; Sorenson et al., 1981). The studies revealed a wide variability in the knowledge of patients, even when measured immediately after the counselling process. However, because of the retrospective design used in these studies, this variation was not conclusively due to effective or ineffective counselling methods. For example, if patients were identified as knowledgeable, this did not necessarily mean that they had acquired their information in their counselling session (Sorenson et al., 1981). Hence, alternative methods of study were used in order to measure the level of knowledge more effectively and to establish the reasons for differential retention of information by patients.

Studies in establishing the success in genetic counselling placed more emphasis on the patients' perspective (Kessler, 1980; Sorenson et al., 1981). In the research previously mentioned, it was found that the information provided in genetic counselling often had profound psychological effects on those receiving the information (Abramovsky, Godmilow, Hirschhorn, & Smith, 1980; Emery,

1984; Emery, et al., 1979; Evers-Kiebooms & van den Berghe, 1979; Keltikangas-Jarvinen & Autio, 1983; Kessler, 1980; Reif & Baitsch, 1985). By assessing the patient's needs and reasons for genetic counselling, in addition to knowledge in medical-genetic information, a better understanding of criteria for effective communication can be established. With this shift in paradigm, studies tended to focus on the patient's questions and concerns before and after the counselling process in order to assess whether or not their objectives were being met in seeking genetic counselling. Sorenson et al. (1981) reported that the more time counsellors spent with patients, the more likely the patients were to discuss their socio-medical concerns. This approach allowed for more open communication between the counsellor and the patient, and, subsequently, a more successful counselling session. Additional studies on the elements of effective communication regarding genetic information have also been published, revealing the importance of both nonverbal and verbal communication for effective genetic counselling (Wertz & Fletcher, 1988; Kurtz & Riccardi, 1979). Kurtz and Riccardi (1979) recognized that while verbal communication was responsible for transferring and interpreting information to patients, nonverbal communication was simultaneously essential for conveying attitudes and emotions. In reviews published by Bernhardt (1989) and

Sjogren and Marsk (1989), it was also recommended that counsellors should determine the patient's level of knowledge before the counselling begins so that they could educate patients at a level that the patients could understand. Although it is possible that some patients may still not understand general concepts after several sessions, it is nevertheless the counsellor's obligation to try to find ways of communicating this complex information (Wertz et al., 1986). Finally, Lum (1987) and Keena, Jawanda, and Hall (1987) stressed the importance of an appreciation for cultural beliefs and traditions when counselling patients. Counsellors should be nonjudgemental with respect to religious and ethnic backgrounds, and be empathetic when providing information. In summary, the role of the genetic counsellor is no longer simply to educate patients, rather it is important to acknowledge and appreciate the psychosocial needs of the patient in order to communicate the information effectively.

In a recent survey of 1,053 medical geneticists in 18 nations, nearly all participants preferred the non-directive approach to genetic counselling (Wertz & Fletcher, 1988). This requires genetic counsellors to concentrate on the presentation of accurate facts in a manner which facilitates decision-making by the patient (Emery, 1984; Frets, 1990; Harris, 1988). In turn, patients are thereby helped and

supported in making decisions for themselves. Emery (1984) referred to this choice as an individual's prerogative, provided it is made in the full knowledge of all the facts and appreciation of the possible consequences. In some instances, patients from different cultures may expect to be told what to do and may have little experience with making their own decisions (Czeizel, Metneki, & Osztovics, 1981; Emery, 1984; Falek, 1984; Lubs, 1979; Wertz & Fletcher, 1988). In these cases, a non-directive approach may confuse the patient more and cause them to make irrational decisions. Thus, a more "client-directed" approach to counselling may meet the objectives in these situations. Pauker and Pauker (1977) and Pitz (1987) have established methods for directive genetic counselling using decision analysis. The models described by both research groups involve decision-aiding technologies. In general, patients are assisted in firstly, defining their genetic problem, and secondly, evaluating their situation by weighing the best outcome against relative costs or burdens of the worst outcome for them. Although such approaches are thought to be more scientific than personal, they are capable of being helpful for patients who have difficulty in assessing their own needs independently. Thus, these methods allow counsellors to help patients in making decisions for themselves, and still be non-directive in their approach.

Effective genetic counselling can be accomplished through a variety of methods. This assessment can be taken even further by using these same measures to compare the effectiveness of various sources of information. For instance, depending on the health care system, patients may receive genetic counselling for prenatal diagnosis from either their primary care physician (family physician or obstetrician) or by a medical geneticist and/or genetic counsellor. Studies have been published which review the genetic counselling offered by these two means in order to determine the cost-effectiveness. In 1980, Lippman-Hand and Cohen reported on the underuse of amniocentesis for prenatal diagnosis. It was suggested that obstetricians' lack of knowledge or negative attitude concerning prenatal diagnosis was preventing them from referring eligible women. In this respect, the physicians would be making decisions for the patient, demonstrating counselling methods contrary to the usual procedure used among medical geneticists. This trend was also observed by Fahy and Lippman (1988) through the Canadian collaborative randomized trial of C.V.S. In this study, the obstetricians felt that they should be directive and advise eligible women to have prenatal diagnosis. Although the attitude had changed from opposing prenatal diagnosis to promoting it, the approach taken by obstetricians in counselling patients still emphasized

influence and direction instead of autonomy and support. Finally, recent reviews by Shiloh et al. (1990) and Sjogren and Marsk (1989) reported that patients who received counselling by medical geneticists or genetic counsellors were less satisfied with the information given to them than patients who received information regarding non-genetic problems from various medical sources. Since a plausible measure of effectiveness in genetic counselling or any other medical service is through patient satisfaction with the services provided, this result was initially disturbing to genetic physicians and counsellors. However, Shiloh et al. (1990) explained that these results did not necessarily mean that genetic counselling was less satisfying than other medical services because there are many factors involved in patient satisfaction. For example, patients who are referred for genetic purposes tend to relate more to what is provided in genetic counselling rather than how it is provided (Shiloh et al., 1990). Thus, once again, an assessment of the effectiveness of genetic counselling depends on the patient's situation and how the service meets the objectives of both its recipients and providers.

Chapter Two

Knowledge and Perception of Information Regarding Prenatal Diagnosis

Knowledge is a prerequisite for making informed decisions about any of the prenatal tests currently being offered to women of advanced maternal age. There is evidence in the literature which demonstrates a large gap between what should be accomplished educationally in genetic counselling and what, in fact, is being accomplished (Evers-Kiebooms & van den Berghe, 1979; Griffin et al., 1977; Keltikangas-Jarvinen & Autio, 1983; Kessler, 1990; Lippman-Hand & Fraser, 1979; Seidenfeld & Antley, 1981; Somer et al., 1988; Sorenson et al., 1981). For example, the percentage of patients capable of demonstrating an adequate understanding of the genetic information post-counselling and subsequently able to provide informed consent varies. Many theories for this lack of knowledge have evolved from these observations, focusing on either the counsellor's or the patient's perspective.

The goal of the consent process in any medical service is providing a mechanism for patients to participate in decision-making with full understanding of the factors relevant to their proposed care (Cassileth et al., 1980). Studies on whether or not this goal is accomplished in services other than genetic counselling have revealed that patients are frequently unable to recall specific information

given to them even after extraordinary efforts are made to provide complete information and to ensure their understanding (Epstein & Lasagna, 1969; Schultz, 1975; Stewart, 1977). This appears to be true regardless of the amount of information delivered, the manner in which it is presented, or the type of medical procedure involved. One explanation for poor knowledge among patients is the technique of those providing the information to patients (Kessler, 1990; Sorenson et al., 1981). Although medical geneticists or family physicians/obstetricians are most often well-trained and knowledgeable in their field, they may not be especially skilled in transmitting the information at a level that the general public can understand. In a recent review published by Kessler (1990), it was suggested that perhaps counselling services of late have reached a plateau in their ability to educate, and that any increase in the effectiveness of genetic counselling will come only from patients in their attempt to understand the information presented to them and from counsellors wishing to assist their patients in making informed decisions regarding prenatal diagnosis.

The most common explanation for patients' lack of knowledge post-counselling has been attributed to the patients themselves. It has previously been mentioned that when measuring the effectiveness of genetic counselling

through patient retention of information, many considerations should be taken into account. First of all, there is the possibility that patients were never given the information, or alternatively, that they had been given the information but failed to understand it and subsequently retain or reproduce it (Hsia & Silverberg, 1973; Marteau et al., 1988). Furthermore, in assessments such as this, it is often very difficult to distinguish between patients' understanding and remembering of genetic information (Evers-Kiebooms & van den Berghe, 1979). For example, if the information patients reproduce is incorrect, it is not always certain if this is due to inadequate understanding during the counselling session or due to a failure of memory. Finally, patients may very well have understood the information, but failed to produce it for cognitive or emotional reasons (Bernhardt, 1989; Griffin et al., 1977; Marteau et al., 1988; Shiloh et al., 1990; Somer et al., 1988; Sorenson et al., 1981). This latter explanation has been reported as the most likely cause for patients' lack of knowledge post-counselling.

An appreciation of a patient's background is essential in effective genetic counselling (Bernhardt, 1989; Davies & Doran, 1982; Shiloh et al., 1990). Studies in measuring recall of information by different study groups observed a correlation between the patients' level of education and how informed they were after counselling (Davies, 1983; Emery et

al., 1973; Griffin, 1977; Kessler, 1990; Wertz et al., 1986). It was often observed that well-educated patients were more informed post-counselling than less-educated patients, possibly due to having seen the information during their schooling or simply because they were used to dealing with probabilities and could assess new estimates with ease. Another factor that can be attributed to variation in recall of information is related to the manner in which the information is provided. For example, most people are not accustomed to making decisions about their personal lives in terms of probabilities. It is possible that patients who had a poor recall of the information had, in fact, been given their risks and remembered them at the time of study measurement. However, the form in which the patients understood the information was not the same as that which was acquired (Griffin et al., 1977; Kessler, 1990; Lippman-Hand & Fraser, 1979). Patients will often transform their risk estimates into a more personal meaning.

In conclusion, it is clear from studies in genetic counselling that people will widely differ in their attitude to risk. Some may put great effort into understanding and weighing the probabilities to make the correct decision for their life situation. Others may not pay any attention at all to the information given, except for the fact that they are "at risk" and will, therefore, react to this fact

appropriately for them. In a study published by Elkins et al., (1986), a vast majority of women who received counselling for Down's syndrome felt that discussions including only risk information were not appropriate, and that genetic counsellors should concentrate more on women's personal reactions and feelings about their situation. In 1990, Kessler published a review of the literature with respect to patient education in genetic counselling and suggested that perhaps precise recall of factual information is not always necessary for informed consent. From the studies on patient retention of information and attitude towards the genetic counselling process, it appears that the prerequisite to informed consent may be knowledge expressed either in quantitative or qualitative terms.

Chapter Three

Factors Involved in the Decision-Making Process Regarding Prenatal Diagnosis

There are a multitude of factors affecting prospective parents' decision-making in prenatal diagnosis. The decision will vary depending on the woman's background and perception of associated risks. There has been much research on identifying the many variables involved so that genetic counsellors may assist patients more effectively in making informed decisions regarding prenatal diagnosis.

The basic process in making a decision is described as the integration of the relevant facts for a particular decision and the values of the decision-maker (Antley, 1979; Pauker & Pauker, 1987; Pitz, 1987). This process can be applied to any decision, but is of particular use to the decision of whether or not to have prenatal diagnosis.

Another approach to this process is based on a model of decision-making under risk. Tversky and Kahneman (1974) and Pearn (1973) describe this process as establishing, firstly, the desirability of the outcome to the individual(s), and secondly, the perceived likelihood of the desirable result occurring. Although it is ultimately the patient who makes the decision of whether or not to have prenatal diagnosis, genetic counsellors can assist in understanding the options available and the implications of having a prenatal test.

Therefore, knowledge of the factors involved in a woman's decision to have prenatal diagnosis is crucial in providing optimal genetic counselling.

The most important factors that women consider when contemplating having prenatal diagnosis are the benefits, risks, and limitations involved with having a procedure (Davies & Doran, 1982; Kessler, 1990). The most common advantage identified by women in various studies is the reassurance that the baby does not have a specific disorder (Davies & Doran, 1982; Fletcher, 1973; Murray, Chamberlain, Fletcher, Hopkins, Jackson, King, & Powledge, 1980). Women who believe that they are at risk may choose to have prenatal diagnosis to relieve their anxiety. Other benefits include providing valuable information about the fetus so that the mother and her primary care provider can prepare for optimal birth care or parents who choose to carry an affected fetus to term can make appropriate preparations, e.g., emotionally and financially (Murray et al., 1980). Finally, parents who choose not to carry an affected fetus to term have the option to terminate based on the information provided through prenatal diagnosis.

There also exist several limitations to having prenatal diagnosis. A common concern expressed by women regarding prenatal testing is the possibility of complications from the procedure (Davies & Doran, 1982; Sjogren & Uddenberg, 1989).

Even though women want to know if the baby is abnormal, some are not willing to risk injuring the fetus to relieve their anxiety in knowing that it is normal (Davies, 1983). Another limitation of prenatal diagnosis that needs to be well-understood by parents is that testing is only specific to the disorders in question, and cannot guarantee a healthy baby (Davies, 1983). Many other congenital anomalies can occur in newborns as well as problems which may develop later on in childhood that cannot be detected by prenatal diagnosis (Griffin et al., 1977; Simpson et al., 1976).

Perhaps the most basic element in a woman's decision whether to have prenatal diagnosis is her perceived susceptibility to having a genetically abnormal baby (Davies, 1983). Although the numerical risk of having a baby with a chromosomal abnormality increases with age, women's perception of risk is known to vary markedly among individuals (Davies, 1983; Frets, Duivenvoorden, Verhage, Peters-Romeyn, & Niermeijer, 1991; Griffin et al., 1977; Lippman-Hand & Fraser, 1979; Sissine et al., 1981; Somer et al., 1988; Sorenson et al., 1981; Wertz et al., 1986). In 1979, Lippman-Hand and Fraser published an important study which reviewed patients' reception of information presented to them in genetic counselling. It was observed through their interviews with patients that although the risk of having a child with a genetic defect is given by the

counsellor in terms of a percentage, the outcome of this value for the patient is binary: the child either will or will not be normal. Thus, the uncertainty concerning the actual outcome of the pregnancy is what parents are most concerned about, and is often weighed more heavily than their numerical risk estimate (Antley, 1979; Griffin et al., 1977; Lippman-Hand & Fraser, 1979; Pearn, 1973; Wertz et al., 1986). Interpretation of genetic risks have also been shown to be influenced by personality traits and past experiences (Pearn, 1973; Sorenson et al., 1981; Wertz et al., 1986). Several studies suggest that patients come to counselling with certain attitudes or beliefs about a problem or disorder which may or may not change post-counselling. For the most part, patients leave the counselling session with a lower perceived risk estimate than what they had expected. Nonetheless, the higher the risk a client believed was involved, the less likely she was to have learned the actual risk as given by the counsellor and the greater the tendency to experience the decision-making process as difficult (Abramovsky et al., 1980; Frets et al., 1991; Pearn, 1973; Sorenson et al., 1981; Wertz et al., 1984). Wertz et al. (1986) also found that most patients tended to overestimate numerically smaller risks, and alternatively, to underestimate numerically larger risks. In any event, the patient's final decision regarding prenatal diagnosis is

usually based on her own personal risk interpretation and expectations about the normalcy of the child rather than on the numerical risk alone.

Other factors which contribute to a woman's perception of risk and the decision to have prenatal testing involve the severity or the burden of the disorder in question. In Pearn's (1973) early studies on patients' subjective interpretations of risks offered in genetic counselling, it was found that the nature of the outcome was the most obvious factor influencing the patient's perception of odds. The decision-making process was believed to involve: (a) a personal view or understanding of the disorder, and (b) a subjective interpretation of the associated risk given for the disorder(s). In a recent study of Drugan et al. (1990), it was found that the severity of the chromosomal anomaly was one of the major determinants in parental decision-making; sex chromosome anomalies were felt to be less severe than autosomal chromosome anomalies. The influence of past experience or familiarity with a particular disorder may also play a role in the decision-making process. Frequently, patients have seen children with the disorder in question and understand the implications of having such a child themselves. Some may feel that they could cope well with an affected child, while others may be more concerned about the perceived burdens of having such a child. Examples of such

burdens include the effect on their personal and family life, the woman's professional life, the demands from society, or financial commitments (Davies & Doran, 1982; Pearn, 1973). In summary, patients' perception of risk estimates will highly depend on their perception of what they are taking a risk for.

Patient attitudes towards their pregnancy may also play a significant role in the decision regarding prenatal testing. The desire for children, and, in particular, healthy children, is a basic human characteristic which is dependent upon personal and cultural circumstances (Frets & Niermeijer, 1990; Lum, 1987; Murray et al., 1980; Pearn, 1973; Sissine et al., 1981; Thomassen-Brepols, 1987). For example, parents may so desperately want a living child of their own after having lost several children from a genetic disease that they will continue to try with the reassurance that prenatal diagnosis is available to them. In these cases, parents are more concerned for the health or quality of their children, and may sacrifice quantity to achieve quality through the use of prenatal diagnosis (Roghmann & Doherty, 1983). Finally, patients may perceive the benefits and risks differently depending upon whether a pregnancy was planned or not (Davies, 1983). Therefore, a patient's motivation with respect to her pregnancy is an additional factor which counsellors should explore in helping patients

to make decisions regarding prenatal diagnosis.

In conclusion, the factors discussed thus far have been identified as the most relevant components of a woman's decision regarding prenatal diagnosis, or more specifically, in the decision to have prenatal testing. Additional factors such as parental age, ethnic and educational background, or economic status have also been demonstrated as having potential to influence a woman's decision (Davies, 1983; Verp, Bombard, Simpson, & Elias, 1988; Wertz & Sorenson, 1986). Prenatal diagnosis is sometimes looked upon as involving two decisions; the first one is whether or not to have a prenatal diagnostic procedure, and the second one is what to do in the event of an abnormal result. According to the National Research Council (1975) and various centers offering prenatal genetic counselling (Fletcher, 1973; Sjogren & Uddenberg, 1988; Somer et al., 1988), these two decisions are separate. The only prerequisite to prenatal diagnosis may be an informed decision regarding prenatal testing, and not a commitment regarding a particular action if results show abnormality.

Chapter Four

Emotional Responses to Prenatal Diagnosis

When prenatal diagnosis was first recognized as a rapidly expanding component of routine obstetric care, efforts were made to assess the efficacy and safety of first and second trimester diagnostic methods. Studies on the effectiveness of genetic counselling for these procedures from the patient's perspective demonstrated the necessity for addressing the emotional issues involved with having a procedure as well. It has been acknowledged that in the majority of instances, emotional disturbance is a normal, rather than a pathological, response to pregnancy (Blumberg, 1984). Pregnancy confronts women with new emotions and adjustments, and more often than not induces periods of uncertainty and fear of fetal injury or defect. Women having prenatal diagnosis comprise a special group of pregnant women because they are undergoing a procedure due to an increased risk for giving birth to a child with a genetic anomaly (Beeson & Golbus, 1979; Blumberg, 1984; Evers-Kiebooms, 1988; Robinson, Tennes, & Robinson, 1975; Sjogren & Uddenberg, 1990; Tunis, 1990). Through studies on the emotional implications of prenatal diagnosis, the necessity of psychological counselling during the prenatal diagnostic process is apparent.

Genetic counselling deals with problems which are more

stressful than many medical problems (Schild, 1984). The information which is transmitted on risk factors regarding genetic disorders and prenatal diagnostic procedures is often considered very complex and difficult to understand even for individuals in less stressful situations. Therefore, unless genetic counsellors pay close attention to the emotional impact prenatal diagnosis places on patients, it is likely that learning and understanding will be compromised (Shiloh et al., 1990).

Studies comparing general patterns of mood states in pregnant women reveal a characteristic U-shaped pattern reflecting increases in anxiety in the first and third trimesters and a decrease occurring in the second trimester (Lubin, Gardener, & Roth, 1975; Tunis et al., 1990). When these assessments were made on women undergoing prenatal diagnosis, the extremes and shapes of the relative mood state patterns changed significantly, revealing different times and levels of anxiety (Tunis et al., 1990). Several researchers have reported additional sources of anxiety regarding the actual diagnostic procedure. For example, the expectation of the test itself has been shown to cause much distress due to the fear of promoting damage or miscarriage of the fetus, or due to the fear of the pain of the procedure (Beeson & Golbus, 1979; Silvestre & Fresco, 1980). Subsequently, waiting for the test result is also a major contributor of

increased anxiety for all women having prenatal diagnosis, regardless of the method used (Beeson & Golbus, 1979; Blumberg, 1984; Robinson et al., 1988; Silvestre & Fresco, 1980; Sjogren & Uddenberg, 1990; Spencer & Cox, 1987; Tunis, et al., 1990). From an interview-based study on reactions to prenatal diagnosis (Silvestre & Fresco, 1980), it was found that the majority of women undergoing amniocentesis indicated that the attachment to the child began only after the test results were known. This statement confirms the previous observations that prenatal diagnosis is associated with a wide spectrum of emotional responses by women which need to be recognized for effective genetic counselling (Sjogren & Uddenberg, 1990; Sorenson et al., 1981).

Many research groups have compared the anxiety experienced by women having prenatal diagnosis (Beeson & Golbus, 1979; Evers-Kiebooms, Swerts, & van den Berghe, 1988; Sjogren & Marsk, 1989; Sjogren & Uddenberg, 1990; Tunis et al., 1990). The general consensus among all reviews is that women having prenatal diagnosis for advanced maternal age (AMA) only are less anxious than those whose indications involve a personal or family history of a genetic disorder. In a review published by Sjogren and Marsk (1989), based on women's experiences with prenatal genetic counselling, it was found that AMA women were more content with the information they received in counselling than patients who received

counselling due to a genetic problem. This suggested that women who are counselled because of a known genetic disorder may require more specialized counselling. Furthermore, it has also been suggested that the elevated levels of personal stress and anxiety experienced by patients with genetic problems may reduce their tendency to take risks (Pearn, 1973). Therefore, fewer women may have prenatal diagnosis for genetic reasons post-counselling in comparison to women who are eligible for age reasons only. Finally, although it has been recommended that anxiety itself should not be a reason for prenatal diagnosis (Michelacci et al., 1984), Sjogren and Uddenberg (1990) suggest that a patient's psychological response and interpretation of her risk in addition to the actual statistical risk estimate should be the deciding factors for eligibility regarding prenatal diagnosis. In summary, through an awareness of the various emotional experiences of women having prenatal diagnosis, genetic counsellors should help patients cope with these feelings before any decisions regarding prenatal diagnosis can be made.

With the relatively recent availability of a first trimester method for prenatal diagnosis, studies have been carried out to determine which method, amniocentesis or chorionic villus sampling (CVS), was considered the more desirable alternative for women (Robinson et al., 1988;

Sjogren & Marsk, 1989; Sjogren & Uddenberg, 1988, 1989; Spencer & Cox, 1987, 1988; Tunis et al., 1990). Although amniocentesis has been the method of practice for many years, the advent of CVS offers several advantages that amniocentesis cannot. First of all, the earlier sampling time (9-12 weeks gestational age) in the first trimester, rather than the second trimester for amniocentesis (15-17 weeks gestational age), makes it possible to detect fetal abnormalities much earlier in the pregnancy. This factor is considered appealing since the entire testing period takes place before fetal movement and significant maternal attachment to the baby (Robinson et al., 1988; Sjogren & Uddenberg, 1989; Spencer & Cox, 1987, 1988). For those women who choose to terminate an abnormal pregnancy, the emotional distress caused by the loss of the child and the procedure for termination may not be as great as for those women who terminate after amniocentesis (Robinson et al., 1988; Spencer & Cox, 1987). Secondly, the waiting time for results after CVS is usually one week, in comparison to three weeks after amniocentesis (actual waiting periods will vary among centers). This factor suggests an earlier reduction in anxiety for CVS patients (Robinson et al., 1988; Sjogren & Uddenberg, 1989; Spencer & Cox, 1987).

Despite the obvious benefits of CVS, there also exist several disadvantages to this procedure. Due to the

relatively recent development of this technique, the risks of CVS due to technical difficulty in carrying out the procedure and possible miscarriage after the procedure were originally greater (1-2%) than what was reported for amniocentesis (<0.5%) (Robinson, 1988; Spencer & Cox, 1987). However, since then, reports from the Canadian (Canadian Collaborative CVS-Amniocentesis Clinical Trial Group, 1989) and American (Rhoads et al., 1989) randomized trials comparing amniocentesis and CVS showed that although a slightly higher risk of procedure failure and fetal loss may exist for CVS, the total loss rates were not significantly different.

In conclusion, it is difficult to compare the emotional impacts of the two most commonly used prenatal diagnostic procedures. Nevertheless, studies that do compare the two methods share at least one observation in common, which is that prenatal diagnosis induces a spectrum of emotions in its participants. The emotional trauma inherent in prenatal diagnosis has caused some concern about the use of these procedures (Blumberg, 1984; Clark & DeVore, 1989; Karp, 1981). However, the alternative to the knowledge that prenatal diagnosis provides is associated with even more severe emotional trauma (Blumberg, 1984; Sjogren & Uddenberg, 1987). In research regarding participants' reactions to prenatal diagnosis in general, the majority of women recognize it as a positive and worthwhile experience

(Blumberg, 1984; Evers-Kiebooms et al., 1988; Finley, Varner, Vinson, & Finley, 1977; Sjogren & Uddenberg, 1988, 1989; Tunis et al., 1990).

Conclusion

There are many issues that must be taken into account when counselling patients with different indications for prenatal diagnosis. It has been suggested that through prospective study methods, a better assessment of patient retention of information and the emotional impact of genetic counselling would be possible. The present research was designed to examine the genetic counselling offered by medical geneticists and genetic counsellors in the Department of Medical Genetics and by primary care providers within the community.

Part B

The Present Study

In the province of British Columbia, prenatal diagnosis is available to all pregnant women age 35 and over at the expected date of confinement (EDC) because of increased risks for fetal chromosome abnormalities. All patients eligible to have a prenatal diagnostic procedure are expected first to receive appropriate counselling to aid in their decision-making. The counselling involves the following: a discussion of their specific age-related risks, a description of the two most commonly used procedures (amniocentesis and chorionic villus sampling) and their respective risks, and an explanation of the meaning of abnormal results. The objective of the counselling is to impart enough information to patients to permit them to make an informed decision and give informed consent.

Prior to November, 1989, all women eligible for prenatal diagnosis were counselled by specially-trained genetic counsellors and medical geneticists through the Department of Medical Genetics, The University of British Columbia (UBC). Since then, those patients having no risks other than those associated with advanced maternal age (AMA) have received such counselling from their primary care physician or obstetrician in the community, rather than by genetic counsellors and medical geneticists at the Medical Genetics clinic. On the day of the procedure, a review of the information presented in counselling is done by a nurse

appointed to the AMA program to ensure that informed consent is provided.

Patients with AMA and additional genetic concerns have continued to be seen by genetic counsellors and medical geneticists. Individualized counselling is provided to such patients with respect to their situation in addition to the counselling given to all patients in the AMA program.

This system provided an unique opportunity to study the effectiveness of the genetic counselling done by primary care physicians in addition to that done by genetic counsellors and medical geneticists.

The subjects in this study comprised three groups of women. The first group consisted of women with AMA only as an indication for prenatal diagnosis (AO). These women were counselled by their primary care physician in the community.

The second group consisted of women with AMA plus other genetic concerns (AP) which did not impose additional genetic risks above their risk associated with AMA or give reason to manage their pregnancy any differently than the women in AO. Examples of concerns in the AP group include an obstetric history of spontaneous pregnancy losses with normal parental chromosomes, a family history of allergies, or concern about exposure to an environmental agent that, in fact, is not a teratogen. The women in AP were counselled by genetic counsellors at the Medical Genetics clinic to discuss their

concerns in addition to AMA counselling.

The third group consisted of women with AMA and complex indications for prenatal diagnosis (AC) which did impose additional fetal risks and may have required additional testing and follow-up. Examples of concerns in the AC group include a history of a previous child with a chromosome abnormality or a neural tube defect. The women in AC were counselled by genetic counsellors and medical geneticists at the Medical Genetics clinic to discuss their risks in addition to age.

The purpose of this thesis is to examine patients' knowledge and satisfaction gained through genetic counselling by genetic counsellors, medical geneticists, and primary care physicians. The study describes the patients' recall of information normally presented in AMA counselling and their emotional responses to their involvement with prenatal diagnosis. From the observations presented in this survey, it is this writer's intention to assess the effectiveness of genetic counselling provided by genetic counsellors, medical geneticists, and primary care physicians regarding risks associated with AMA.

Chapter Five

Method

Subjects

There were two criteria for selection of subjects. Firstly, they had to be pregnant women aged 35 and over at the expected date of confinement and, therefore, eligible for prenatal diagnosis. Secondly, they had to have sufficient fluency in verbal and written English to provide informed consent and to complete the questionnaires. In the event of an eligible subject who was not English-speaking, the language of preference was nonetheless recorded to establish whether or not there was a significant need to arrange for multilingual counselling within the prenatal service.

Over a period of five months, all patients eligible for prenatal diagnosis were asked to participate in this study with the following exceptions: patients were seen only on Wednesdays and Fridays during the latter two months of the study period due to other research involving the CVS patients and other academic commitments, and patients were not seen on eight non-consecutive days due to this writer's absence for various reasons. Therefore, patients who participated in this study were representative of the population from which they were drawn.

Forty-seven of 884 patients (6%) were excluded due to a

language barrier. The majority of these patients were of Oriental descent and had access to several Chinese or Cantonese-speaking physicians within the community to provide them with appropriate counselling. Thirteen of 884 patients (1%) refused to participate due to increased anxiety or simply because they were too busy. Seventy-two of 884 patients (8%) who had given consent to participate and 63 of 884 patients (7%) who had not given consent ultimately did not participate due to insufficient time available to complete the first questionnaire. Finally, there were 38 of 884 patients (5%) who agreed to participate and completed the first questionnaire, but were subsequently excluded from the study for several reasons: a missed abortion, fetal demise, or ultrasound abnormality detected at procedure, more than six weeks elapsed before completing the second questionnaire, or they chose not to have a prenatal diagnostic procedure post-counselling. Even though the data collected from these 38 patients was not included in the complete data analysis, their responses were reviewed to determine whether they differed from the other patients.

Of the initial 884 eligible patients for this study, 399 (45%) met inclusion criteria and successfully completed both questionnaires. There were 311 of 722 patients in AO (43%), 52 of 102 patients in AP (53%), and 36 of 60 patients in AC (60%).

Procedure

Patients were asked to complete two questionnaires. The first questionnaire (Q1) was completed before the patients had a prenatal diagnostic procedure, and the second questionnaire (Q2) was completed after the patients had a prenatal diagnostic procedure. Due to the change in the provision of genetic counselling regarding AMA and prenatal diagnosis, the timing of Q1 completion varied among groups.

From November, 1989, AMA only patients (AO) were counselled by their primary care physician in the community rather than by genetic counsellors at the Medical Genetics clinic. Only after counselling were such patients referred to and seen at the Prenatal Assessment Unit (PAU) on the day of their procedure. Therefore, it was not possible to access these patients prior to receiving genetic counselling. On the other hand, since the patients in AP and AC were counselled by genetic counsellors and medical geneticists at the Medical Genetics clinic (situated on the same site as the PAU), it was possible for the women in these groups to complete Q1 prior to receiving genetic counselling as a time could be arranged for them to do so at the clinic on the day of their appointment. A summary of the times at which Q1 and Q2 were completed in each group is included in Table 1.

The time elapsed between genetic counselling, the day of the procedure, and the completion of Q2 was recorded for all

TABLE 1: QUESTIONNAIRES #1 & #2; TIMES COMPLETED

	QUESTIONNAIRE 1 (Q1)	QUESTIONNAIRE 2 (Q2)
AMA ONLY	POST COUNSELLING at the Prenatal Assessment Unit(PAU) before having a prenatal test.	POST PROCEDURE either (i) at the PAU after their procedure and before leaving the hospital. (ii) at home to be mailed back within four weeks.
AMA PLUS AMA COMPLEX	PRIOR TO COUNSELLING at the Medical Genetics clinic.	

patients due to its potential to decrease patients' retention of information provided in genetic counselling. The number of women in all three groups who completed both questionnaires on the same day was: 264 of 311 patients in AO (85%), 23 of 52 patients in AP (44%), and 18 of 36 patients in AC (50%). The maximum time elapsed in AP and AC was two weeks between receiving genetic counselling and having a procedure, and up to four weeks until Q2 was completed. However, only 5 of 52 patients in AP (10%) and 5 of 36 patients in AC (14%) fell into this category. The time elapsed between receiving genetic counselling and completion of Q1 in AO was unavailable, although 48 of 311 patients (15%) in this group completed Q2 within four weeks after having a prenatal diagnostic procedure.

Patients who had previously had prenatal diagnosis were included in this study since their risk associated with advanced maternal age will have increased in comparison to their risk in a previous pregnancy. The breakdown of women in this category are described in the results section.

Presentation of Data

The questionnaires used in this study were designed to collect information from women having prenatal diagnosis for advanced maternal age (as a minimal indication). The types of information requested involved demographic variables (for example, age, gravidity, ethnic origin, level of education,

etc.), numerical risks associated with advanced maternal age and having a prenatal diagnostic procedure, and personal information regarding their involvement with prenatal diagnosis.

All questions were designed in collaboration with this writer's thesis committee with the exception of two which were obtained from questionnaires reported in the psychology literature. These two measures included the A-State scale of the State-Trait Anxiety Inventory (Spielberger, 1970), and the Subjective Stress Scale (Berkun, Bialek, Kern, & Yagi, 1962).

In the State-Trait Anxiety Inventory, patients were evaluated regarding feelings of tension, nervousness, worry, and apprehension (Spielberger, 1972). Half of the items from a list of 20 relate to the presence of these feelings, and the remaining items reflect the absence. Patients are instructed to rate each item from (1) not at all, to (4) very much so. For items in which a high rating indicates low anxiety, the scoring weights are reversed. For example, if a patient gives a score of 4 to the statement, "I feel calm," her score in the analysis is only 1. Thus, the analysis defines a continuum of increasing levels of state anxiety intensity, with low scores indicating states of calmness and serenity, intermediate scores indicating moderate levels of tension and apprehensiveness, and high

scores reflecting states of intense apprehension and fearfulness (Spielberger, 1972).

The Subjective Stress Scale is also designed to assess an emotional state by requiring patients to choose one adjective from a list of 14 which best describes how they are feeling at a particular point-in-time. The list involves a spectrum of adjectives, ranging from very positive to very negative examples.

Samples of the questionnaires used in this study are included in Appendix A. The A-State scale is included as #2 in Q2, and the Subjective Stress Scale is included as #21(b) in Q1 and #10 in Q2.

As there are no previous studies of this kind available to compare results with, measures of statistical significance will not be made in this analysis. Thus, the presentation of the data will involve a descriptive analysis only such that interpretations of "more" or "less" will refer to qualitative rather than quantitative descriptions. Due to the differences in sizes of the three groups of women in addition to the various backgrounds and indications for prenatal diagnosis, a comparison between the three groups of women was not made in this thesis.

Chapter Six

Results

Background Data

The women in all three groups did not differ greatly with respect to age, ethnic origin, and educational background. A description of these results in addition to other demographic variables is included in Table 2. As mentioned previously, women who had had prenatal diagnosis in a previous pregnancy were included in this study since it is the women's knowledge of risks associated with their present pregnancy that is of interest.

All of the women who participated in this study were pregnant, age 35 and over at the expected date of confinement, and, therefore, eligible for prenatal diagnosis. Depending on age and indications for prenatal diagnosis, women chose either amniocentesis or chorionic villus sampling (CVS). The majority of patients in each group had amniocentesis: 230 of 311 in AO (74%), 31 of 52 in AP (60%), and 25 of 36 in AC (69%).

The majority of women in each group said that their pregnancy was planned (70% in AO (N=306), 71% in AP (N=52), and 51% in AC (N=35)), and that it took less than one year to conceive (70% in AO (N=309), 75% in AP (N=52), and 79% in AC (N=34)).

TABLE 2: BACKGROUND DATA

	AMA ONLY N=311	AMA PLUS N=52	AMA COMPLEX N=36
AGE (mean)	37.089	37.615	37.083
ETHNIC ORIGIN - Caucasian	83% (257/311)	90% (47/52)	89% (32/36)
RELIGION - Protestant	45% (130/311)	48% (25/52)	58% (21/36)
EDUCATION - completed high school - ≥2 yrs. post-secondary	 25% (74/311) 63% (191/311)	 22% (11/50) 66% (33/50)	 27% (9/33) 64% (21/33)
OBSTETRIC HISTORY - gravida - ≥1 loss (SA)	 2 (35%, 61/309) 41% (126/305)	 4 (21%, 11/52) 75% (38/51)	 2 (31%, 11/35) 64% (23/36)
PREVIOUS PND	17% (52/311)	40% (21/52)	25% (9/36)

Variables Regarding Prenatal Diagnosis

In all three groups, the majority of women said that they had considered having prenatal diagnosis (PND) before being given information about the service from their doctor/ medical geneticist or other source: 77% in AO (N=298), 79% in AP (N=52), and 69% in AC (N=35).

The first and second most important "reasons for having PND" in each group was being in a high risk age group (66% in AO (N=311), 58% in AP (N=52), and 42% in AC (N=36)) and not wanting to have an abnormal baby regardless of the risk (36% in AO (N=311), 37% in AP (N=52), and 39% in AC (N=36)), respectively. This question was included in #9 in Q1. In the case where patients received counselling by genetic counsellors and medical geneticists, having a personal or family history of a genetic disorder was also indicated as the second most important reason for 12% of the patients in AP (N=52) and the most important reason for 28% of the patients in AC (N=36).

The sources identified as the most responsible for providing information about genetic problems were the patients' family doctor (48% in AO (N=254), 23% in AP (N=44), and 43% in AC (N=35)), or obstetrician (17% in AO (N=254), 20% in AP (N=44), and 29% in AC (N=35)). Other possibilities such as family and friends, media, or books were relevant for less than 10% of all patients. Patients who were counselled

by genetic counsellors and medical geneticists claimed a Genetic Associate as their source of information by only 12% of the patients in AP (N=44) and 14% of the patients in AC (N=35). A supplementary question identified the patients' family doctor as the first most helpful in making a decision to have PND (47% in AO (N=311), 40% in AP (N=52), and 33% in AC (N=36)). The second most helpful individual(s) were identified as the patients' family and friends in AO (21%, N=311) and obstetrician in AP (21%, N=52) and AC (22%, N=36).

Finally, the majority of women in all three groups said that their most important "reason for choosing to have PND" was to have the option of terminating an abnormal pregnancy: 50% in AO (N=311), 62% in AP (N=52), and 67% in AC (N=36). This question was included as #20 in Q1. The second and third most important reasons were to relieve anxiety by knowing that the baby's chromosomes were normal (52% in AO (N=311), 46% in AP (N=52), and 56% in AC (N=36)), and to prepare for an abnormal child (19% in AO (N=311), 16% in AP (N=52), and 25% in AC (N=36)), respectively.

Risks Associated with Advanced Maternal Age

In order to assess how informed the patients were through the relative knowledge gained in their counselling process, several questions included in the first questionnaire (Q1) were repeated in the second questionnaire (Q2). With respect to risks associated with advanced

maternal age (AMA), the risks of having a baby with Down's syndrome were discussed. Patients were asked to first of all indicate whether or not they had been told their risk of having a baby with Down's syndrome, and secondly, to give what the approximate risk of having a baby with Down's syndrome is at age 30, 40, and at their own age.

All responses given for numerical risk estimates were categorized into three levels; informed, somewhat informed, and uninformed. The limits used to establish these levels were derived from B.C. data for Down's syndrome (Hook, 1981), and through personal communication with Dr. B. McGillivray. A description of the levels used for different ages are included in Appendix B.

In Q1, approximately 75% of all women said that they had been told their risk of having a baby with Down's syndrome. However, patients who claimed that they had been told their risk did not always reproduce that number when requested. For example, 239 of 304 patients in AO (79%) indicated that they had been told their risk, yet only 161 of those 239 patients (67%) actually reproduced that risk when asked to do so. The other 78 of 239 patients (33%) left the entry blank and were considered as having no recall in the data analysis. An explanation for treatment of missing data will be discussed further in the discussion. A graphical representation of these results in addition to Q1 results

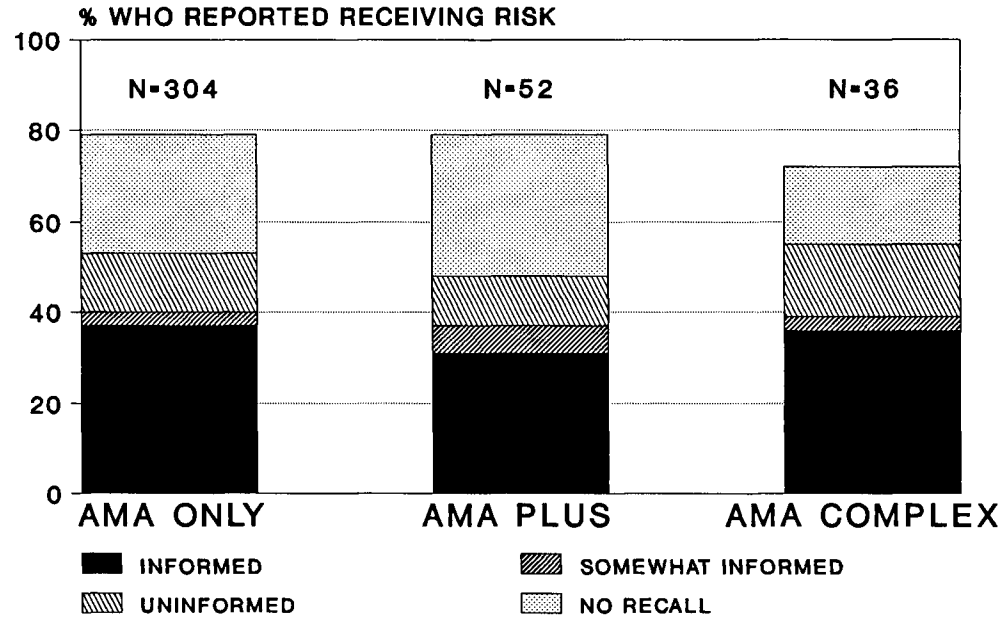
from AP and AC are included in Figure 1.

The numerical risk of having a baby with Down's syndrome at age 30 and 40 was given by less than 30% of the patients in each group in Q1. In addition to this low level of response, the majority of the risk estimates given were uninformed responses. Thus, due to such low numbers of informed patients overall, the results from these entries were not tabulated.

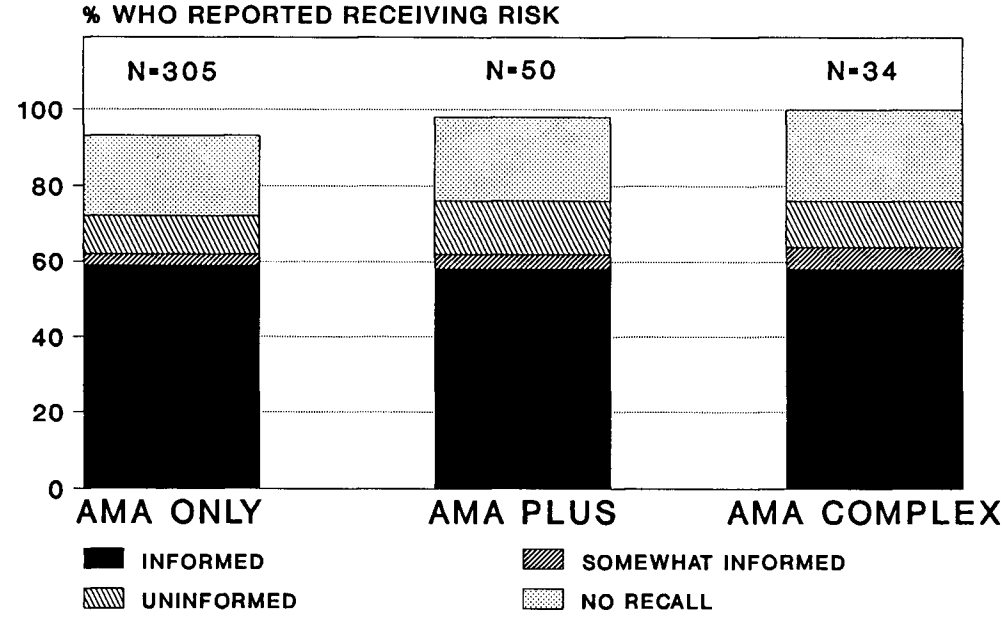
In comparing Q1 and Q2 responses, the number of patients who said that they had been told their risk of having a baby with Down's syndrome increased in each group from 79% in Q1 (N=304) to 93% in Q2 (N=305) for AO, 79% in Q1 (N=52) to 98% in Q2 (N=50) for AP, and 72% in Q1 (N=36) to 100% in Q2 (N=34) for AC. Among those patients who proceeded to reproduce a risk estimate for their own age in Q2, there was an increase in the number of informed patients: 70% in Q1 (N=161) to 82% in Q2 (N=221) for AO, 64% in Q1 (N=25) to 77% in Q2 (N=39) for AP, and 65% in Q1 (N=20) to 77% in Q2 (N=26) for AC. However, although the number of patients having no recall decreased from Q1 to Q2, approximately 25% of the women in each group were still unable to recall their risk estimate in Q2. Results for Q2 patient recall of their risk of having a baby with Down's syndrome are also included in Figure 1. Results of risk estimates for ages 30 and 40 are not included for the same reasons as in Q1.

FIGURE 1.

Q1: PATIENT RECALL OF RISK OF DOWN'S



Q2: PATIENT RECALL OF RISK OF DOWN'S



In addition to scoring the number of informed patients regarding risks associated with AMA, the data were also used to determine how many women simply recognized that the risk of having a baby with Down's syndrome increases with age. This was accomplished by scoring the number of women who gave a lower risk for age 30 than at age 40, and a risk estimate in between if their age was within these limits. The results from this analysis showed that only 13% in AO (N=311), 16% in AP (N=52), and 22% in AC (N=36) indicated this knowledge through their responses in Q1, and even fewer proportions in Q2.

Finally, patients were also asked to indicate how they felt about their own risk of having a baby with Down's syndrome. For example, they were asked to indicate whether they felt that their risk was high or low. In AO, 51% of the patients (N=288) in Q1 and 59% of the patients (N=297) in Q2 felt that their risk was low. The patients who did not respond with a low risk perception felt that their risk was either high (35% in Q1 and 31% in Q2) or average (14% in Q1 and 10% in Q2). In AP, 50% of the patients (N=46) in Q1 and 45% of the patients (N=49) in Q2 felt that their risk was high. Again, alternative responses were either low (35% in Q1 and 37% in Q2) or average (15% in Q1 and 18% in Q2). Finally, in AC, 44% of the patients (N=32) felt that their risk was low and 44% of the patients felt it was high in Q1.

Comparing this to Q2, the number of patients with a high risk perception increased to 50% (N=34), leaving 41% of the patients feeling that their risk was low, and 9% average.

Risk Associated with Neural Tube Defects

In the province of British Columbia, a woman's risk of having a baby with a neural tube defect is 1/700 (McBride, 1979). This anomaly is routinely screened for using the sample obtained from women having amniocentesis. The 1/700 risk estimate applies to the majority of women regardless of their age and may or may not be included in the prenatal counselling process. In order to assess whether or not counselling for neural tube defects was provided to women in this study, and if so, if they were informed, a question was included in exactly the same manner as the previous question. That is, the women were asked if they had been told their risk of having a baby with a neural tube defect, and if so, what did they think the approximate risk was at age 30, 40, and at their own age. The purpose of maintaining consistency in the design of these questions was not to provide the patient with any information that could reveal whether the risk was associated with age or not.

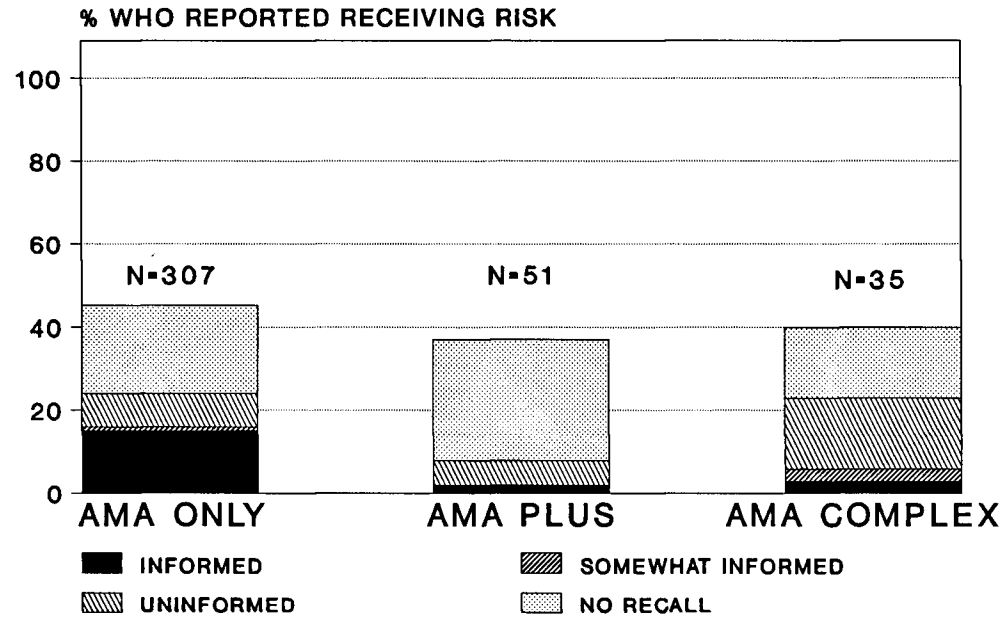
In Q1, approximately 40% of all women said that they had been told their risk of having a baby with a neural tube defect. As in the previous question, not all of the women who said that they had been told their risk were able to

reproduce that risk when requested to do so: 139 of 307 patients in AO (45%) reported receiving a risk with only 73 of those 139 patients (53%) actually reproducing a risk, 19 of 51 patients in AP (37%) reported receiving a risk with only 4 of those 19 patients (21%) reproducing a risk, and finally, 14 of 35 patients in AC (40%) reported receiving a risk with only 8 of those 14 patients (57%) reproducing a risk. The number of informed patients varied across the three groups: 45 of 73 in AO (62%), 1 of 4 in AP (25%), and 1 of 8 in AC (13%). All Q1 results are included in Figure 2, demonstrating an overall low level of response regarding risks associated with neural tube defects.

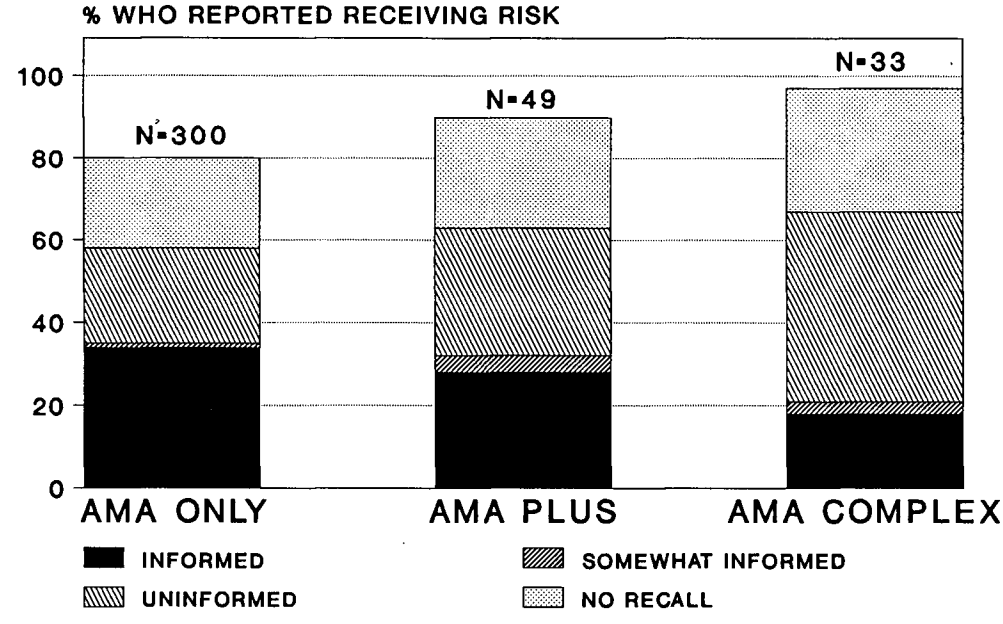
In comparing Q1 to Q2 responses, the number of women who reported having received a risk regarding neural tube defects increased to 80% in AO (N=300), 90% in AP (N=49), and 97% in AC (N=33). In addition to this, the number of women who were able to reproduce their risk also increased to 72% in AO (N=241), 70% in AP (N=44), and 69% in AC (N=32). As in Q1, the number of informed patients varied across the three groups: 104 of 174 patients in AO (60%), 14 of 31 patients in AP (45%), and 6 of 22 patients in AC (27%). In comparison to Q1 results, Figure 2 demonstrates an increase in the level of response for all groups. However, the overall numbers of informed patients continued to represent less than a majority in each group.

FIGURE 2.

Q1: PATIENT RECALL OF RISK OF NTDs



Q2: PATIENT RECALL OF RISK OF NTDs



Since the response rates to risks associated with neural tube defects at ages 30 and 40 were less than 30% by all groups in both Q1 and Q2, the results from these entries were not tabulated. However, the data was reviewed to determine how many women simply acknowledged that the risk of having a baby with a neural tube defect is not associated with age and is the same for all women in B.C. This was accomplished by scoring the number of women who gave the same risk estimate for age 30, 40, and their own age (be it informed or not), as well as those who could not recall the exact number yet indicated on the questionnaire that they were aware that it was the same for women of all ages. In Q1, only 8% of the patients in AO (N=311), 6% of the patients in AP (N=52), and 3% of the patients in AC (N=36) identified this information. Comparing these results to those found in Q2, the number of women acknowledging this information increased to 15% in AO (N=311), 24% in AP (N=52), and 14% in AC (N=36).

As in the previous question regarding Down's syndrome, patients were also asked how they felt about their risk of having a baby with a neural tube defect. In Q1, 65% of the patients in AO (N=311), 56% in AP (N=52), and 58% in AC (N=36) gave a response to this question, whereas a majority of the women in each group said that they felt that their risk was low: (159 of 203 in AO (78%), 17 of 29 in AP (59%), and 14 of 21 in AC (67%). In Q2, the response rate increased

in each group to 84% in AO (N=311), 79% in AP (N=52), and 81% in AC (N=36), and as in Q1, the majority of the women in each group continued to feel that their risk was low: 210 of 260 in AO (81%), 29 of 41 in AP (71%), and 16 of 29 in AC (55%).

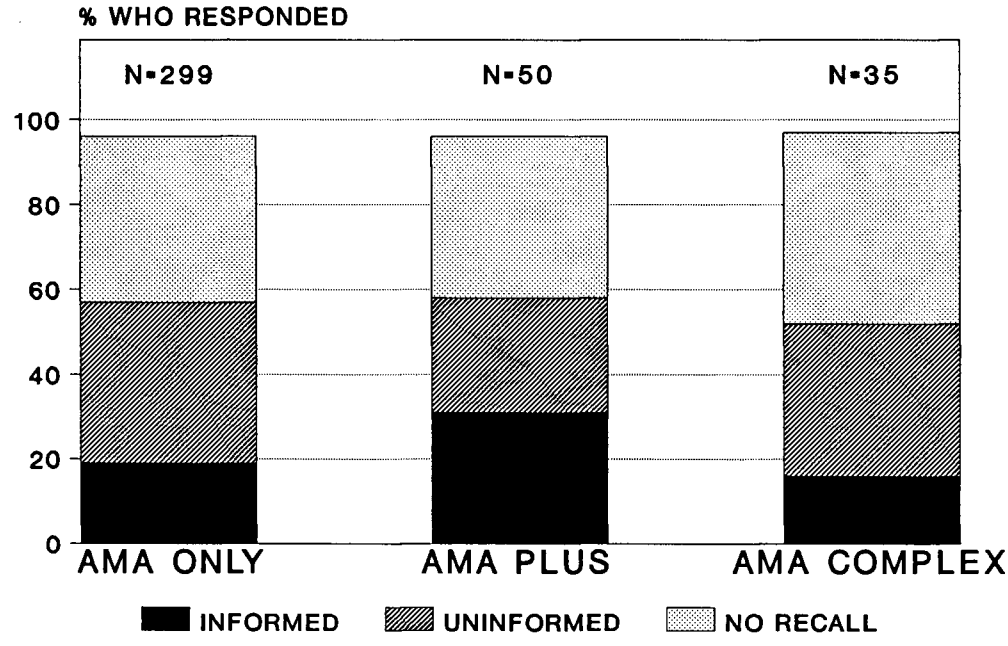
Risks Associated with Prenatal Diagnostic Procedures

A prerequisite to an informed decision to have a prenatal diagnostic procedure is knowledge about the procedure. Given that all patients in this study were eligible to have a prenatal test and, subsequently, had either an amniocentesis or chorionic villus sampling (CVS), women were asked several questions regarding risks associated with having a prenatal diagnostic procedure. For both procedures, the same spectrum of risk estimates was provided in order to reduce the number of correct responses by chance alone. Patients who left the entry blank or indicated that they did not know the risk were scored and tabulated as having no recall.

In response to the risk associated with having an amniocentesis, Figure 3 demonstrates that a large number of patients in each group advanced from having no recall in Q1 to being informed in Q2. Thus, the number of informed patients in each group increased from 20% (N=299) in Q1 to 74% (N=305) in Q2 for AO, 32% (N=50) in Q1 to 68% (N=52) in Q2 for AP, and 16% (N=35) in Q1 to 80% (N=35) in Q2 for AC. In each event, the response rate was above 90% for all groups

FIGURE 3.

Q1: PATIENT RECALL OF RISK WITH AMNIO



Q2: PATIENT RECALL OF RISK WITH AMNIO

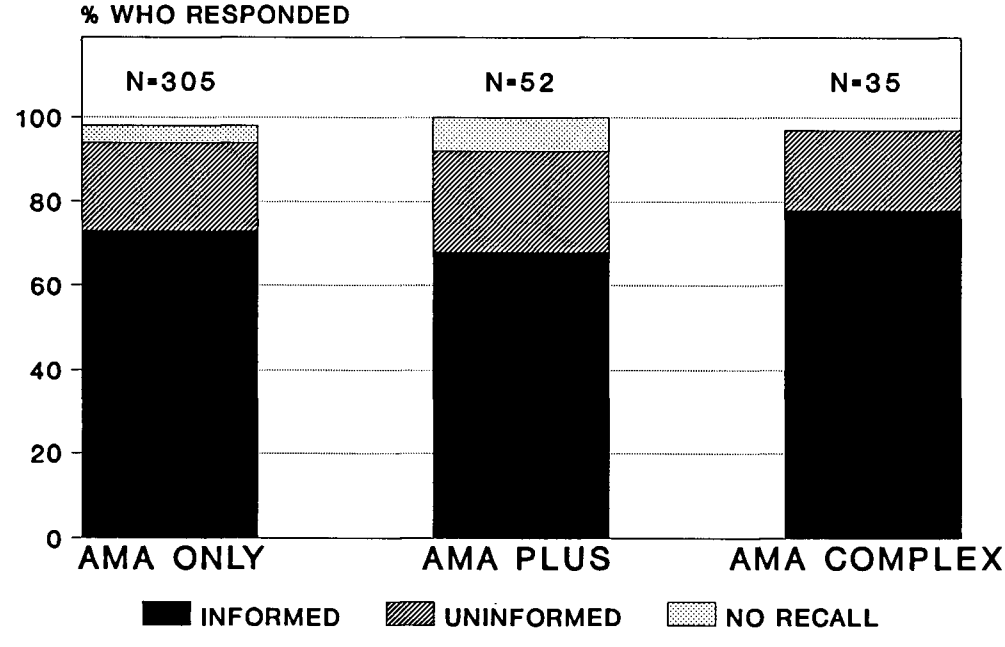
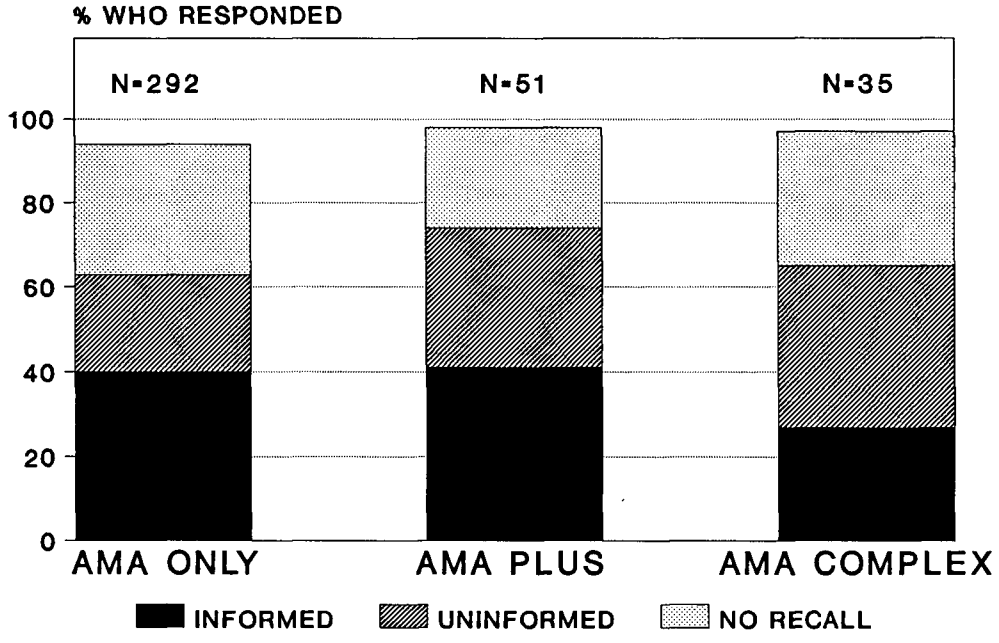
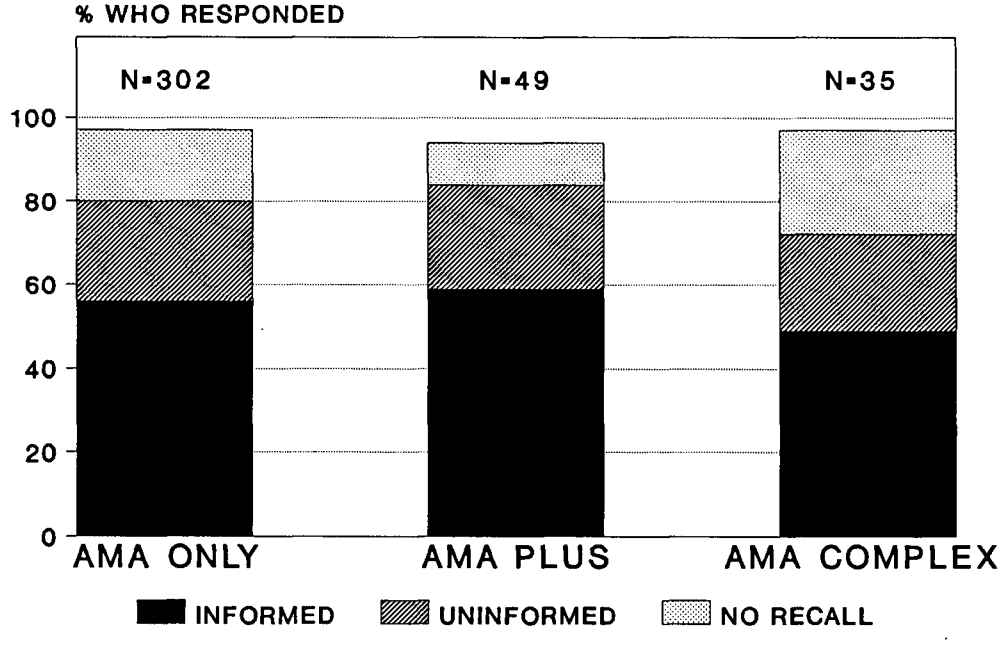


FIGURE 4.

Q1: PATIENT RECALL OF RISK WITH CVS



Q2: PATIENT RECALL OF RISK WITH CVS



concerned.

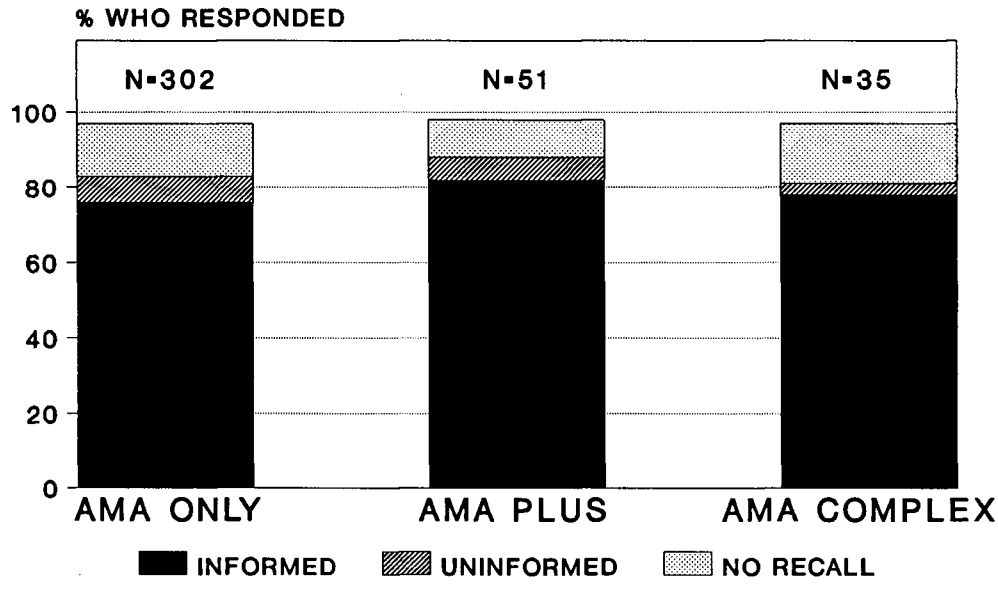
Patient recall of the risk associated with having CVS also showed an increase in the number of informed patients. A graphical representation of these results is included in Figure 4. In Q1, 43% of the patients in AO (N=292), 42% in AP (N=51), and 27% in AC (N=35) were informed. Comparing these results to Q2, the majority of patients in all three groups were informed: 58% in AO (N=302), 63% in AP (N=51), and 51% in AC (N=35). Once again, the response rate for all entries regarding the risk associated with CVS was above 90% for all three groups.

Although ultrasound is not an invasive technique, it is nonetheless a routine procedure in prenatal care, and was, therefore, included in scoring patient knowledge regarding risks associated with prenatal procedures. For the most part, the number of informed patients in Q1 remained the same in Q2 across all groups, maintaining an average of 75% of women recognizing that there is no known risk of having a miscarriage due to ultrasound. Results from this question are included in Figure 5.

In addition to information regarding risks associated with having a prenatal diagnostic procedure, patients were also asked what they thought was the approximate risk of having a miscarriage in the first twelve weeks of pregnancy. As seen in Figure 6, the results form this entry reveal that

FIGURE 5.

Q1: PATIENT RECALL OF RISK WITH ULTRASOUND



Q2: PATIENT RECALL OF RISK WITH ULTRASOUND

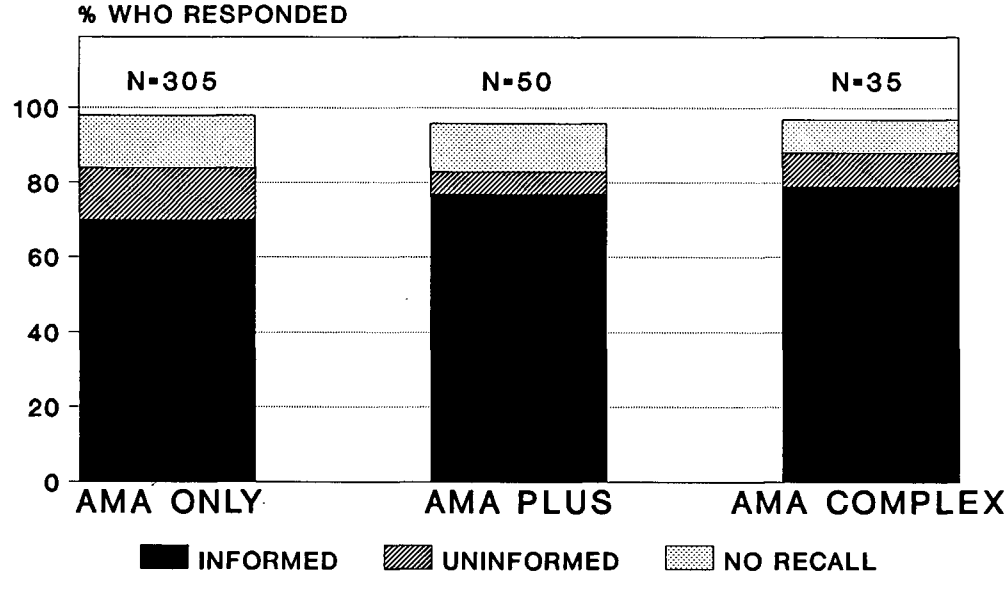
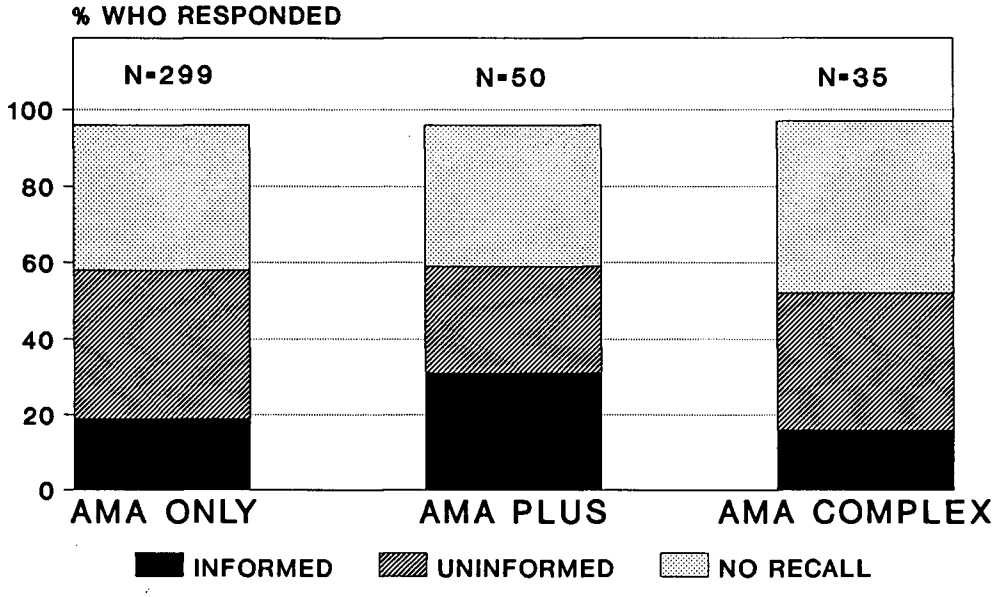
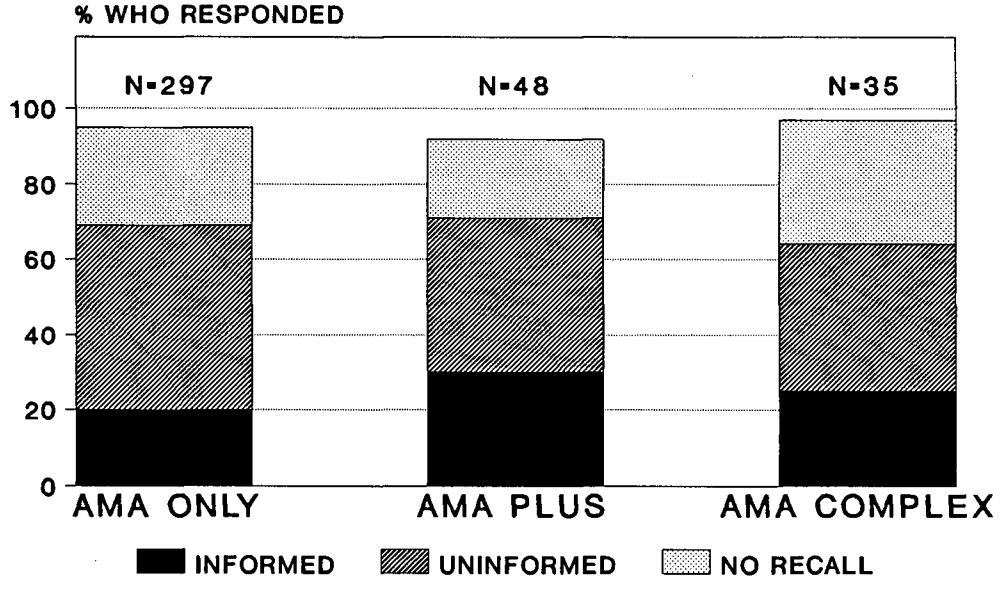


FIGURE 6.

Q1: PATIENT RECALL OF MISCARRIAGE RISK IN 1st TRIMESTER WITH NO PROCEDURE



Q2: PATIENT RECALL OF MISCARRIAGE RISK IN 1st TRIMESTER WITH NO PROCEDURE



the patients were very unaware of the magnitude of this risk in comparison to the risk estimates given regarding prenatal testing. In Q1, the proportion of informed patients were 20% in AO (N=299), 32% in AP (N=50), and 16% in AC (N=35). In Q2, the number of informed patients were 21% in AO (N=297), 33% in AP (N=48), and 26% in AC (N=35), demonstrating only a small increase in the number of informed patients in AC.

Based on the information provided with respect to options available in prenatal testing for future pregnancies, the patients were also asked which procedures they would or would not have, and why. With respect to prenatal diagnosis by amniocentesis, 79% of the patients in AO (N=295), 69% in AP (N=49), and 71% in AC (N=31) said that they would choose to have this method of testing in the event of a future pregnancy. CVS was chosen by 52% of the patients in AO (N=272), 61% in AP (N=46), and 30% in AC (N=30). Finally, ultrasound was chosen by 97% in AO (N=293), 96% in AP (N=49), and 97% in AC (N=31). One of the reasons given for not choosing a particular method of testing included the refusal to choose CVS due to its higher reported risk of miscarriage in comparison to amniocentesis: 10% of the patients in AO (N=311), 10% in AP (N=52), and 3% in AC (N=36).

Knowledge in Problems Regarding Abnormal Results

All patients who participated in this study were at increased risk of having a baby with a chromosomal problem.

For this reason, patients were asked if they were familiar with the sorts of problems that people with Down's syndrome experience.

Patients in all three groups identified several problems associated with Down's syndrome. The most common problems were mental retardation (approximately 70% in AO (N=311), AP (N=52), and AC (N=36)), physically handicapped* (20% in AO (N=311), 35% in AP (N=52), and 19% in AC (N=36)), and heart problems (15% in AO (N=311), 27% in AP (N=52), and 28% in AC (N=36)). Other problems which were identified by approximately 15% of the patients in each group included a short life-span, an unusual facial appearance, and behavioural problems. The patients' source of information regarding problems associated with Down's syndrome was not requested in this question.

Patients were also asked if they were familiar with the sorts of problems experienced by people with a neural tube defect. In most general terms, 24% of the patients in AO (N=311), 27% in AP (N=52), and 42% in AC (N=36) said that these people are physically handicapped. Other more specific problems included paralysis (29% in AO (N=311), 23% in AP (N=52), and 33% in AC (N=36)), skeletal abnormalities (15% in AO (N=311), 15% in AP (N=52), and 23% in AC (N=36)), and

---

* = exact definition of this problem not provided.

retardation (12% in AO (N=311), 25% in AP (N=52), and 6% in AC (N=36).

In order to explore whether or not the women had considered the repercussions of having an abnormal pregnancy, they were also asked what would be the most significant problem for them if they were to have a baby with Down's syndrome. The four most common problems identified in each group were having to worry about who would care for the child once the parents grew old (27% in AO (N=311), 25% in AP (N=52), and 22% in AC (N=36)), coping with the problems associated with the syndrome (30% in AO (N=311), 27% in AP (N=52), and 22% in AC (N=36)), the effect it would have on their family life (15% in AO (N=311), 29% in AP (N=52), and 17% in AC (N=36), and the emotional stress and lack of patience they would experience (15% in AO (N=311), 17% in AP (N=52), and 11% in AC (N=36).

State Anxiety and Feelings about Prenatal Diagnosis

Women who are eligible for prenatal diagnosis may experience a variety of emotions. The various responses may be induced depending upon their indication for prenatal testing, through learning about the potential problems associated with childbearing at an advanced maternal age, or from making a decision to have an invasive prenatal procedure.

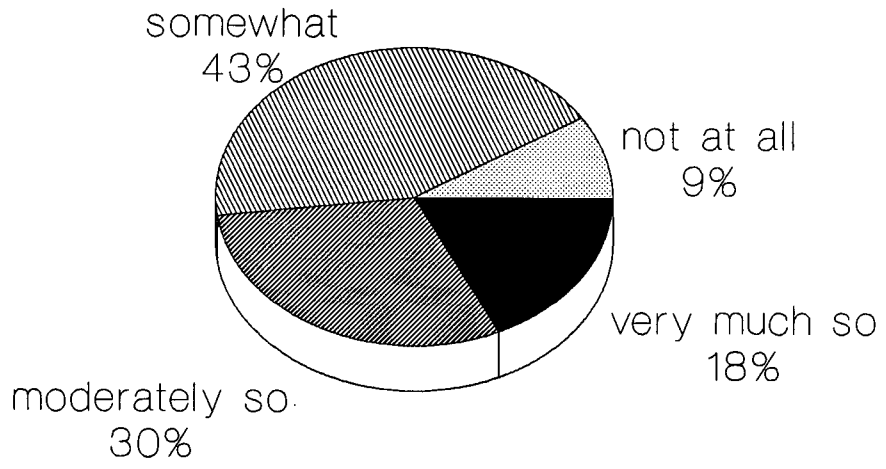
In both questionnaires, patients were asked how anxious

they were feeling at that moment on a scale from 1 to 4. Referring back to Table 1, patients in AO were, therefore, revealing their feelings in Q1 post-counselling and immediately before their procedure, patients in groups AP and AC were stating their feelings in Q1 immediately before their genetic counselling session, and patients in all three groups revealed in Q2 how they were feeling after their procedure. Even though most of the patients in each group completed both Q1 and Q2 on the same day (85% in AO (N=311), 44% in AP (N=52), and 50% in AC (N=36), there exist limitations of this analysis regarding the effect of variable time. For example, those patients who completed Q2 within four weeks after their procedure may have been less anxious than those who completed Q2 immediately after their procedure. A description of the timing of questionnaire completion and its potential effect on the results will be included in the discussion.

The state anxiety decreased from before having a procedure to after having a procedure for all groups. In Q1, the majority of patients in each group felt somewhat anxious to moderately anxious. Once the procedure was complete, the majority of patients in AO and AP felt somewhat anxious to not at all anxious. In AC, however, the majority of patients remained somewhat anxious to moderately anxious. These results are presented in Figures 7-9.

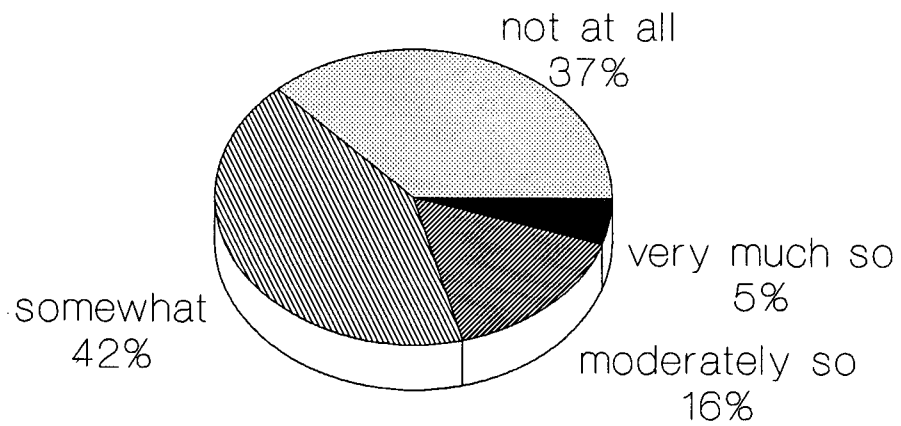
In the second questionnaire, the same type of question

FIGURE 7.
ANXIETY STATE: AMA ONLY



PRE-PROCEDURE

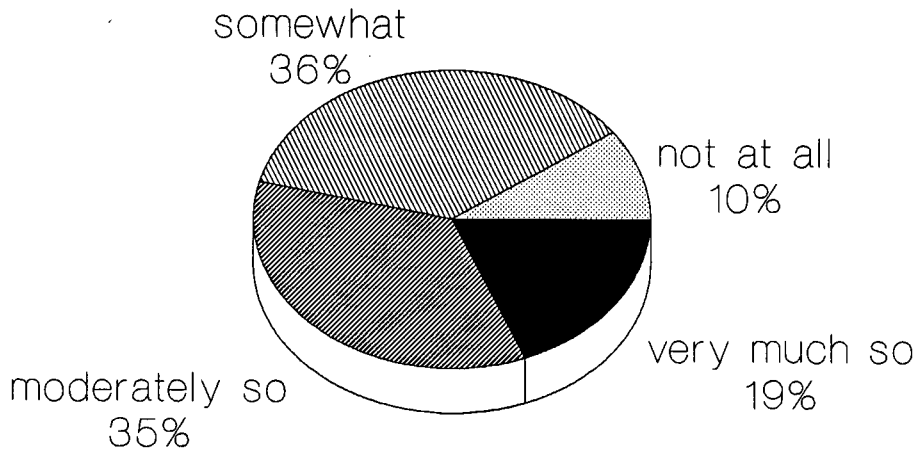
N=304



POST-PROCEDURE

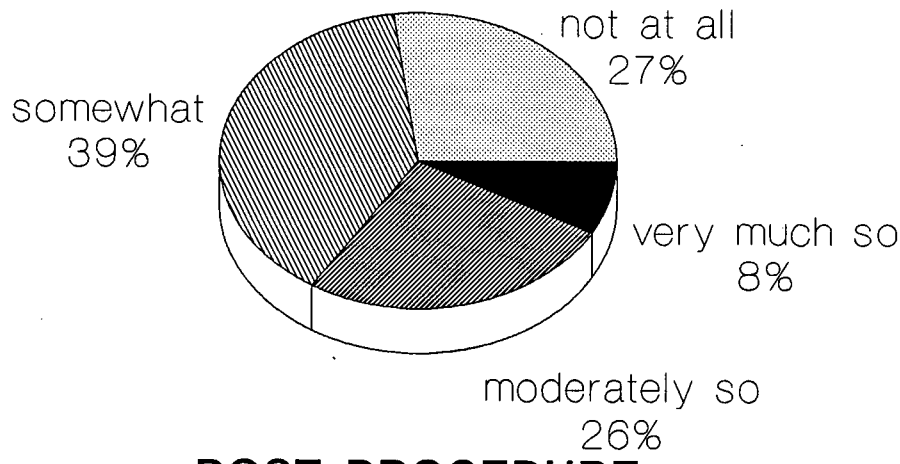
N=303

FIGURE 8.
ANXIETY STATE: AMA PLUS



PRE-PROCEDURE

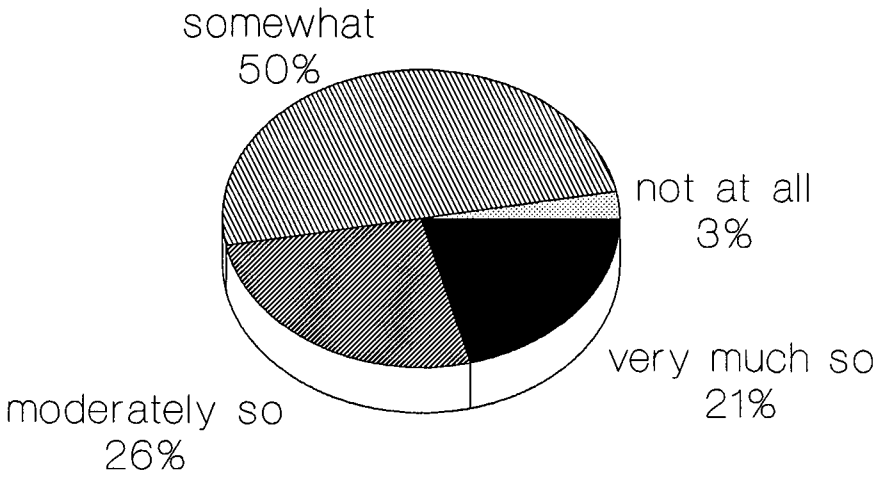
N=52



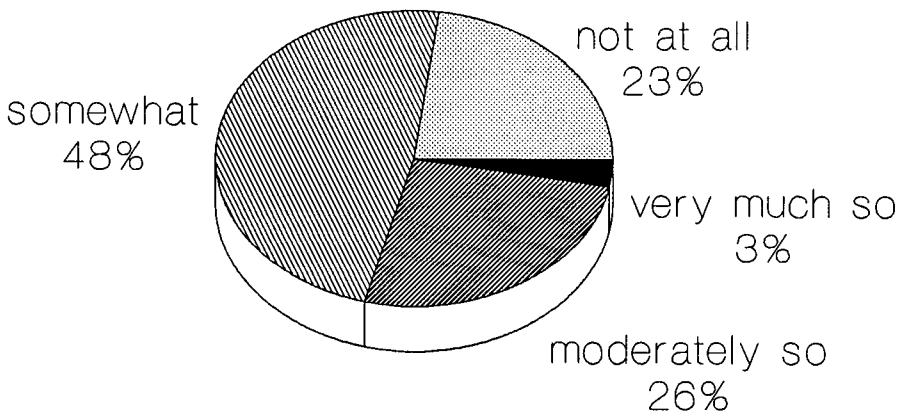
POST-PROCEDURE

N=51

FIGURE 9.
ANXIETY STATE: AMA COMPLEX



PRE-PROCEDURE
N=34



POST-PROCEDURE
N=35

was also used to ask patients how anxious they were feeling regarding the test results (see Figure 10). In this case, the majority of patients in all three groups were feeling somewhat anxious to moderately anxious. In addition to this, more patients in AP (27%, N=304), and AC (23%, N=35) felt very anxious regarding the test results than those who felt very anxious once the procedure was complete (8% in AP (N=303), 3% in AC (N=35)).

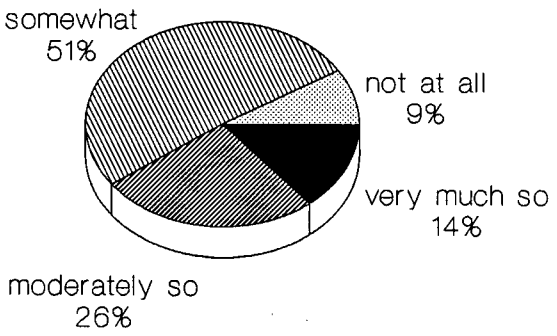
In response to the Subjective Stress Scale, most of the patients in all three groups progressed from feeling nervous in Q1 (31% in AO (N=275), 35% in AP (N=43), and 44% in AC (N=34)) to feeling fine in Q2 (31% in AO (N=238), 25% in AP (N=48), and 22% in AC (N=36)). Patients who chose more than one item to best describe how they were feeling in either Q1 or Q2 were not included in the analysis of this question.

The results from the A-State scale of the State-Trait Anxiety Inventory were tabulated for each patient using a scoring key provided by Consulting Psychologists Press (Spielberger et al., 1970). A score could range from a minimum of 20 to a maximum of 80. In each group, the scores were averaged to give a final score of 37.41 in AO (N=264), 40.77 in AP (N=44), and 42.72 in AC (N=34).

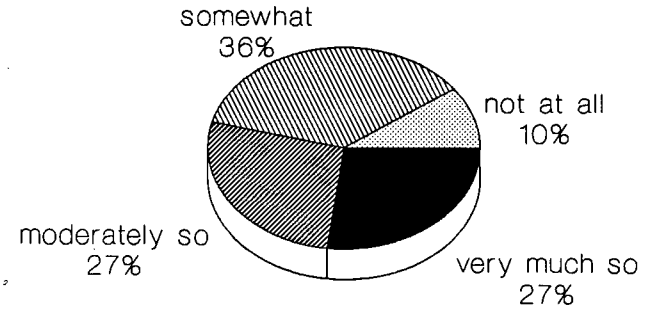
Additional Comments

Additional comments were made by the patients with respect to their overall experience with prenatal diagnosis

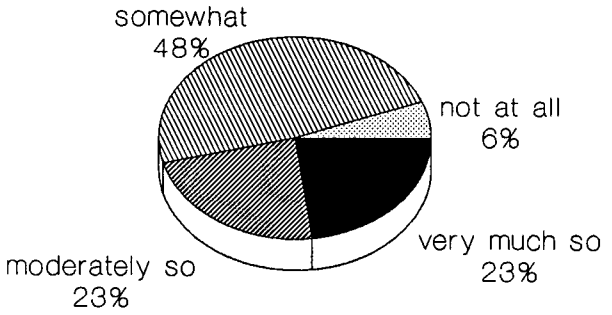
FIGURE 10.
ANXIETY STATE: TEST RESULTS



AMA ONLY
N=304



AMA PLUS
N=52



AMA COMPLEX
N=35

for advanced maternal age. First of all, 74% of the patients in AO (N=311), 69% in AP (N=52), and 69% in AC (N=36) said that nothing had happened during the procedure that they had not expected. At the end of Q2, the patients were asked how satisfied they were with the overall counselling they received. On a scale from (1) very satisfied to (4) quite disappointed, the majority of patients in all three groups said that they were very satisfied: 64% in AO (N=300), 78% in AP (N=51), and 80% in AC (N=35). Finally, a section reserved at the end of each questionnaire for any comments revealed that patients in each group (16% in AO (N=311), 33% in AP (N=52), and 19% in AC (N=36)) were very impressed by the staff involved in the actual prenatal procedure. They were described as very friendly, supportive, and informative.

A Descriptive Comparison Between the Complete Data Set and an Incomplete Data Set (Q1 Only)

Subjects were included in this study based on eligibility for prenatal diagnosis and ability to read and write English. Those who did not meet these criteria were excluded from this study, and, therefore, were never given a questionnaire. As mentioned previously, there were 38 women who did meet these criteria and completed Q1, but not Q2 for various reasons. This subset of women comprised 24 AO patients, 11 AP patients, and 3 AC patients.

A descriptive analysis of the data collected from this

subset of patients showed that there were no apparent differences between their Q1 responses and the Q1 responses of the patients who completed both questionnaires.

Chapter Seven

Discussion

The present study suggests that the effectiveness of the genetic counselling process regarding age-related risks and prenatal diagnosis will vary depending upon the patient's emotional reaction and individual circumstances regarding their involvement with prenatal diagnosis. For the most part, the genetic counselling provided by medical geneticists, genetic counsellors, and primary care physicians has been shown to be informative and supportive for patients. However, the results suggest that the factual information provided in counselling may not always be the deciding factor in the patients' decision to have prenatal diagnosis. To facilitate the interpretation of the results, findings will be discussed in the order of presentation in the previous section.

A Description of the Sample

The sample of women involved in this study represents a population of women who are pregnant at an advanced maternal age (35 years and over at EDC). For the most part, the women in each group were approximately 37 years of age, Caucasian, Protestant, and had a post-secondary education of two or more years. The obstetric history revealed that most of the women were pregnant for at least the second time, and had experienced one or more losses (spontaneous abortions).

In choosing to have a prenatal diagnostic procedure, the patient's age and indication for prenatal diagnosis played an important role. Due to a limited time schedule committed for CVS procedures at Grace Hospital, Vancouver, B.C., patients with first of all, genetic indications, and secondly, greatest age, have priority. Therefore, the fact that amniocentesis was the chosen procedure for the majority of women in each group does not suggest that there is a greater desire for amniocentesis than CVS.

The Reasons for Having Prenatal Diagnosis

The main reasons for the patients to have prenatal diagnosis were (a) because they were in a high risk age group, and (b) because they did not want to have an abnormal baby, regardless of their risk. Another reason which was identified by patients in AP and AC was a personal or family history of a genetic disorder. This was to be expected given the definitions of the three groups based on indications for prenatal diagnosis.

A total of eight patients in the entire study group responded that their most important reason to come for prenatal diagnosis was to find out the sex of the baby. Seven of these patients came from the community (AO), and one was referred to the Department of Medical Genetics due to a minor complication (AP). Through looking at these individual patients more closely, it was observed that 4 of the 8

patients were East Indian, practicing either the Sikh (2) or Hindu (2) religion, and one patient was Chinese Roman Catholic. The remaining three patients had left the entries asking their ethnic origin and religious affiliation blank. Given the fact that there were only four East Indian patients who participated in this entire study, this suggests that a correlation exists between East Indians and the use of prenatal diagnosis for sex determination. Therefore, the counselling process for such patients should concentrate on the cultural significance of sex determination in addition to the risks associated with advanced maternal age.

The Reasons for Wanting Prenatal Diagnosis

All women in this study have at least age as a common indication for prenatal diagnosis. However, the most important reason for wanting to have a prenatal test will depend on each individual and her personal experience with prenatal diagnosis.

The first and second most important reasons for wanting PND were (1) to have the option to terminate an abnormal pregnancy, and (2) to relieve anxiety by knowing that the baby's chromosomes were normal, respectively. A third reason given for wanting PND was to prepare for an abnormal pregnancy. This option was included in this question to assess how many women would not choose to terminate an abnormal pregnancy when given the opportunity.

Unfortunately, the results make this assessment difficult to make since there were women in all three groups who chose (1) as their most important reason, and the opportunity to prepare for an abnormal baby as their second or third most important reason. By responding with "to terminate" and "to not terminate" in that order to the same question, this researcher concludes that either the women did not understand the question or that they did understand it, but are undecided as to what they would do if an abnormal result was found.

The Decision-Making Process Regarding Prenatal Diagnosis

The results show that the majority of patients in each group had thought about or considered having PND prior to receiving any specific information, for example, risk of miscarriage, eligibility, etc. This suggests that there exists an awareness within the community regarding the availability of this service, and, subsequently, a corresponding interest. It has also been suggested in studies similar to this one that women may make decisions regarding prenatal testing in the absence of relevant information, and that the genetic counselling process serves to reassure them by providing information that helps them feel confident in their decision (Sorenson, 1981).

The provision of accurate information regarding specific risks and medical techniques is very important to ensure that an informed consent is given to have prenatal diagnosis. In

this study, a large number of women in all three groups had received the bulk of their information from their family doctor or obstetrician. Patients in AP had most probably never seen a genetic counsellor prior to the time when Q1 was completed. In addition to this, patients in AP or AC may have perceived the medical geneticists and genetic counsellors as "doctors," making the family doctor the main source of information for 23% of the patients in AP (N=52) and 43% in AC (N=36) an overestimation. Perhaps the patients in groups AP and AC could have identified their main source of information more specifically if the design of this question had included a medical geneticist or a genetic counsellor as an available response.

The role of the genetic counsellor is to provide the necessary information a patient needs to make an informed consent regarding prenatal diagnosis. In addition to this, counsellors should provide the patient with support and complete autonomy throughout their decision-making process. The results from this study reveal that the individuals who were identified as being the most responsible for providing them with the necessary information were also identified as the most helpful to them in making their decisions to have prenatal diagnosis. The question of whether or not they experienced autonomy in their decision-making process cannot be assessed from the data available, but will be discussed in

a subsequent research study involving the sample of women reported here.

The Effectiveness of Genetic Counselling Regarding Age-Related Risks and Prenatal Diagnosis

The objective of genetic counselling is to impart enough information to patients to enable them to make a truly informed decision. For the purpose of this study, the genetic counselling provided by primary care physicians, medical geneticists, and genetic counsellors was assessed by determining how informed the patients were through their ability to recognize and understand a series of risk estimates related to advanced maternal age and having a prenatal diagnostic procedure.

Given the data collected from questions requiring patients to recall numerical risk estimates, there were missing cases in both questionnaires and within all three groups of patients. This causes a problem in the analysis of the results since it is unknown as to whether or not a particular question was left blank because the patient was uninformed or could not remember the answer or if she did not read or understand the question.

In the event that patients left questions blank because they were uninformed or could not remember the specific risk estimates, this could suggest that their genetic counselling process was inadequate in providing them with the necessary

information that patients need to make an informed decision. On the other hand, this could also suggest that patients who had, in fact, been counselled appropriately did not remember the risk estimates because they may not have been important to them in making a decision to have prenatal diagnosis. The results presented in Tables 3-6 support the latter explanation for missing cases since approximately 25% of the patients in each group left the entries requesting risk estimates for Down's syndrome and neural tube defects blank after having reported that they had, in fact, been told these numbers. Patients who reported that they had not been told any risk estimates and subsequently left all entries requesting these numbers blank represented less an 20% of the patients in each group.

In the event that patients refrained from answering questions because they did not read or understand them, the validity of the data presented in this research would be questionable. Nonetheless, due to the observations made thus far, in addition to the fact that the total number of missing cases for any question represents a maximum of 20% of the patients in each group, this researcher concludes that the presentation of the questionnaires used in this study was not responsible for the occurrence of missing cases.

The effectiveness of the genetic counselling process will be discussed within each group, taking into account at

which time patients received their genetic counselling in relation to Q1 and Q2, and from whom the counselling was received.

(i) Risks associated with advanced maternal age. In all three groups, a majority of the women said that they had been told their risk of having a baby with Down's syndrome. Nevertheless, not all of the women who reported having received the risks were able to state what those risks were. This was demonstrated in both questionnaires, suggesting the inability of patients to recall a specific numerical risk that is provided to them in genetic counselling.

The purpose of requesting a risk estimate at age 30, 40, and the patient's own age was to see if the women could at least recognize the fact that a woman's risk of having a baby with a chromosomal problem increases with age. Unfortunately, this knowledge was not demonstrated since the majority of the women in each group responded to what they thought their own risk was, and did not respond to risks at age 30 or 40. Perhaps this knowledge could have been revealed if the question had been phrased differently. For example, instead of asking patients to state what the risk was at age 40, the question could have asked if the risk at age 40 was greater than, less than, or the same as the risk at age 30.

Looking at each group individually, the patients in AO

demonstrated an increase in the number of women who reported having received their risk (79% (N=304) in Q1 to 93% (N=305) in Q2), and an increase in the number of informed patients (47% (N=239) in Q1 to 64% (N=285) in Q2). Given the assumption that these patients had received their counselling in the community prior to completing Q1 and due to the findings in Q1 and Q2 that show an increase in the number of informed patients, this researcher concludes that at least 14% (N=305) of the patients in AO had received information regarding their risk of Down's syndrome at some point-in-time between Q1 and Q2. This percentage was derived by comparing the number of patients who reported receiving their risk in Q1 and Q2. The increase in the number of informed patients may have resulted from either patients learning their risk of Down's syndrome for the first time at Grace Hospital, or being unable to remember the exact number when completing Q1 and being reminded at Grace Hospital. Although the patients' source of this information was not indicated, they may have gained their knowledge from either the AMA nurse who reviewed their risks with them and witnessed them signing the consent form to have a prenatal diagnostic procedure, or perhaps from the individuals involved in performing the procedure.

Referring to the patients in AP, the results also show an increase in the number of patients who reported having received their risk (79% (N=52) in Q1 to 98% (N=50) in Q2) as

well as an increase in the number of informed patients (39% (N=41) in Q1 to 60% (N=50) in Q2). Since the patients in this group received their counselling regarding age-related risks and prenatal diagnosis (in addition to counselling regarding their personal or family history) between completing Q1 and Q2, this researcher concludes that any gain in knowledge shown by this group of patients may have been due to the counselling they received from a genetic counsellor. Although the AMA nurse witnessed the signing of the consent form by all patients having a prenatal diagnostic procedure, a review of their risks, etc. was only given to the patients who were counselled by their primary care physician in the community.

The results from the patients in AC also show an increase in the number of women who reported having received their risk (72% (N=36) in Q1 to 100% (N=34) in Q2) as well as in the number of informed patients (50% (N=26) in Q1 to 59% (N=34) in Q2). As in the case for patients in AP, patients in AC also received their counselling between Q1 and Q2, thus suggesting that any gain in knowledge demonstrated by these patients was due to the counselling they received from either (or both) a medical geneticist or genetic counsellor. Since this group of patients has been defined as AMA plus complex indications for prenatal diagnosis, it is likely that these patients had been seen previously by either a medical

geneticist or a genetic counsellor. Nonetheless, the women's risk associated with AMA would have been greater in this pregnancy, making her overall risk (AMA risk plus any additional risk related to their indication for prenatal diagnosis) greater as well.

(ii) Risk associated with neural tube defects. The results presented in Figure 2 suggest that counselling for neural tube defects (NTDs) was not always a routine component of the counselling process given to the patients in this study. This is not surprising since the common indication for all patients in this study to have prenatal diagnosis was because of their increased risk of having a baby with a chromosomal problem, and not due to their risk of NTDs. As mentioned previously, the samples collected from patients who had had an amniocentesis were tested for NTDs in addition to chromosomal abnormalities. Given that the majority of the patients in each group had had an amniocentesis, the results suggest that perhaps these women were not aware of being screened for this anomaly, or they were aware but were uninformed regarding its frequency.

The purpose of requesting a risk estimate at age 30, 40, and at their own age was to see if the patients could recognize that the risk of NTDs does not increase with age, and is constant for women of all ages. As in the case with the risk of Down's syndrome, this knowledge was not

demonstrated since the majority of patients in each group only responded to their own risk of NTDs, and left the risk associated at age 30 and 40 blank. By designing this question in the same manner as the question regarding Down's syndrome, this question may have only served to confuse the patients.

As in the previous question regarding Down's syndrome, there were also patients in each group who were not able to state what their risk of NTDs was after having reported that they had received this information. The management of missing cases has been discussed previously, suggesting several explanations for the patients' lack of response.

In group AO, only 45% of the patients (N=307) stated that they had received their risk in Q1. This suggests that the majority of patients who were counselled by their primary care physician had not been counselled regarding NTDs. In Q2, the number of patients who stated that they had received their risk increased to 80% (N=300), which suggests that there were patients in AO who had been told their risk of NTDs during the time period between completing Q1 and Q2. Although the source of the patients' gain in knowledge was not identified, it may have been either the AMA nurse or the individuals involved in performing the procedure who provided patients with this information. Even though the results demonstrate an increase in the number of informed patients in

AO (32% (N=139) in Q1 to 43% (N=241) in Q2), the number of informed patients continued to represent less than a majority of the patients in each group.

With reference to the results in Q1 for groups AP and AC, only 37% (N=51) and 40% (N=35) of the patients, respectively, stated that they had been told their risk of NTDs. Comparing these results to those in Q2, the numbers increased to 90% in AP (N=49) and 97% in AC (N=33). This suggests that patients in groups AP and AC had been told their risk of NTDs in their genetic counselling process. The number of informed patients in each group also increased from 5% (N=19) in Q1 to 32% (N=44) in Q2 for AP and from 7% (N=14) in Q1 to 19% (N=32) in Q2 for AC. Nevertheless, the overall numbers of informed patients in these groups, as well as in AO, represent much less than a majority of the patients in each group.

In summary, a total of 80% or more of the patients in each group stated that they had received their risk of NTDs. When asked to recall their risk, only 43% or less of the patients in each group were able to reproduce an informed risk estimate and approximately 30% of the patients in each group had no recall at all. Therefore, this researcher concludes that the specific risk of NTDs was not important to the majority of the patients in this study.

(iii) Risks associated with prenatal diagnostic procedures. All of the women included in this study had experienced having either an amniocentesis or a CVS at some point-in-time during the study period. In comparing results in Q1 and Q2, all three groups demonstrated an increase in the number of informed patients regarding risks associated with prenatal testing. Results in Q2 show that a majority of the patients in each group were informed. This suggests that, first of all, patients in AP and AC had received this information from the counselling provided by medical geneticists and genetic counsellors, and secondly, that patients in AO had received this information after their counselling and at some point-in-time between completing Q1 and Q2. The problem of missing cases was not an issue in this case such that all questions related to prenatal procedures received a greater than 90% response by the patients in each group.

In comparison to the number of informed patients in the two previous risk assessments (DS and NTDs), the patients were clearly more informed regarding their chance of miscarriage due to a procedure than their risk of having an abnormal baby. This could suggest that at the time of prenatal testing, patients were generally most concerned (due to their greater recall of the associated risks) about the repercussions from choosing to have a procedure.

Alternatively, the patients' source of information may have provided them with risks associated with procedures more often or more appropriately than risks associated with Down's syndrome or NTDs.

In addition to the risks associated with having a procedure, the patients in this study (as well as all other women who are pregnant) are also faced with a background risk of miscarriage during their first twelve weeks of pregnancy. Genetic counselling for AMA patients should inform women that prenatal tests (AMN/CVS) are used to rule out chromosomal abnormalities and are not capable of assuring them of having a normal, healthy baby. The results show that the patients in this study were very unaware of the limitations of having a prenatal diagnostic test before as well as after their counselling process. In each group, a majority of the patients thought that their background risk of miscarriage was much lower than it actually is.

In response to a general attitude towards the various methods of prenatal testing, the women were most accepting of ultrasound, which is a non-invasive technique and a routine component of prenatal care in B.C. With respect to the two most common invasive techniques, amniocentesis was more widely accepted than CVS by all three groups in this study. Unfortunately, this researcher cannot conclude from these results that more women prefer amniocentesis over CVS since

patients who knew that they were not eligible to have CVS may have chosen against it. For example, due to age priority, it would be difficult for a 35-year-old pregnant woman to have CVS if her only indication for having prenatal diagnosis was AMA. If the question had asked patients which procedures they would prefer regardless of eligibility, perhaps a general attitude towards various methods of prenatal testing would have been revealed.

The Response Regarding Abnormal Results

The minimal indication for all of the patients in this study to have prenatal diagnosis was advanced maternal age. Given the potential problems associated with child-bearing at a later age, it was important to identify whether or not the patients were aware of the sorts of abnormalities that a prenatal test can screen for so that they could make decisions regarding the management of the remainder of their pregnancy.

In each group, approximately 70% of the patients stated that children with Down's syndrome experience mental retardation, and about half of these patients also indicated other associated problems. In addition to this, less than 30% of the patients in each group were able to identify what children with NTDs experience. The patients' source of their information was not indicated in these questions.

It has been discussed previously in this section that

greater than 80% of the patients in each group had reported having received their risk of Down's syndrome and NTDs. Taking all of these results into account, this suggests that patients may have been informed regarding their risk of having a baby with a NTD without being informed regarding what this abnormality involved. On the other hand, this could also suggest that patients either could not remember what they were told or they may have been told but intentionally disregarded the question because they felt that their risk of NTDs was not as important as other risks and concerns. In any event, the effectiveness of counselling regarding neural tube defects appears to be less than the counselling regarding Down's syndrome due to its lack of association with advanced maternal age.

The Emotional Response to Prenatal Diagnosis

It has been well-documented that women experience varying levels of anxiety during their pregnancy, especially if and when a prenatal test is elected. The results from this research are no exception to these conclusions. For the purpose of this study, the analysis of patients' emotional response was related to prenatal diagnosis in general, and not related to a specific diagnostic test.

Figures 7 through 9 show that the level of state anxiety and subjective stress decreased in all three groups. For the patients in AO, this change in emotions occurred during the

time period immediately before their procedure (post-counselling) and immediately after their procedure. Those patients who preferred to complete Q2 at home and return it back to this writer within four weeks (before receiving their test results) may have felt less anxious than the patients who completed Q2 immediately after their procedure. Nevertheless, since there were only 48 of the 311 patients (15%) who did not complete Q2 immediately after their procedure, the results demonstrate that the majority of patients in AO experienced a sense of relief as soon as the prenatal test was complete.

In groups AP and AC, patients also experienced a decrease in anxiety, although 34% of the patients in AP (N=51) and 29% of the patients in AC (N=35) continued to feel moderately to very anxious once the procedure was complete. This level of anxiety can be expected of these patients due to their additional concerns related to their pregnancy. With respect to the amount of time elapsed between completing Q1 and Q2 in these groups, a large proportion of the patients in AP (44%, N=52) and in AC (50%, N=36) completed both questionnaires on the same day.

In addition to asking patients to rate their level of state anxiety at the time of completing Q2, patients were also asked to rate their anxiety regarding their test results. Comparing the results in Figures 7-9 to the results

in Figure 10, the patients' level of state anxiety appears to increase once again in each group, demonstrating approximately the same distributions as were seen in their anxiety experienced pre-procedure. This suggests that women who have PND may experience two stages during their pregnancy where anxiety may be induced: the first stage involves having the prenatal diagnostic procedure, and the second stage involves waiting for the test results.

The alternative approach used to measure state anxiety involved the State-Trait Anxiety Inventory (STAI) developed by C.D. Spielberger and associates in 1966. For the purpose of this study, the state-anxiety (A-State) analysis was applied to provide reliable, self-report measures of state anxiety at a particular moment-in-time. The patients were asked to respond to 20 different statements (A-State analysis) in Q2, thus after having had their procedure. In the interest of time and extent of other information requested of patients in Q1 and Q2, the A-State inventory was only included in Q2. Therefore, this analysis served to measure state anxiety at one specific time rather than changes in anxiety over a period of time.

The results from the A-State inventory show that all three groups revealed intermediate scores. According to Spielberger's interpretation of A-State scores (Spielberger et al., 1970), this suggests that all of the patients

experienced moderate levels of tension and apprehensiveness. Therefore, even though the three groups of women differed with respect to their indications for PND and who provided them with genetic counselling, the majority of women in each group experienced approximately the same anxiety post-procedure.

A Descriptive Comparison Between the Complete Data Set and an Incomplete Data Set

A descriptive comparison was made between the complete study group (patients who completed both Q1 and Q2) and the incomplete study group (patients who completed Q1 only) to determine whether or not there were any indications within the incomplete study group that would have predicted their exclusion from this study. Given that there were no apparent differences observed, this suggests that the patients comprising the incomplete study group were representative of the population from which they were drawn and were excluded from this study by chance alone.

Limitations of the Current Study

There are several limitations which have the potential to influence the results of this study.

The method used in this study to assess the effectiveness of the genetic counselling process in three groups of women involved the application of two questionnaires; the first was to be given prior to

counselling, and the second to be given post-counselling (after having a prenatal test). Due to the division of responsibility regarding the provision of genetic counselling for AMA patients, it was not possible to assess all of the patients in this study before and after receiving their genetic counselling. In addition to this, the amount of time elapsed between completing Q1 and Q2 varied within each group.

For example, although 264 of 311 patients in AO (85%) completed Q1 and Q2 on the same day, the remaining 48 of 311 patients in AO (15%) completed Q2 within four weeks after their procedure and before receiving their test results. In groups AP and AC, most of the patients also completed Q1 and Q2 on the same day (44% in AP (N=52), 50% in AC (N=36)), although the remaining patients in these groups completed Q1 and Q2 in a variety of ways: (a) patients completed Q1 immediately before their counselling and had their procedure on the same day but completed Q2 within four weeks after their procedure (25% of the patients in AP (N=52) and 11% in AC (N=36)); (b) patients completed Q1 and received their counselling on the same day but had their procedure and completed Q2 up to two weeks later (8% of the patients in AP (N=52) and in 11% in AC (N=36)); (c) patients completed Q1 and received their counselling on the same day, had their procedure up to two weeks later, and completed Q2 within four

weeks after their procedure (10% of the patients in AP (N=52) and 14% in AC (N=36)); and finally, (d) patients completed Q1, had their procedure, and completed Q2 within two weeks after their procedure (13% of the patients in AP (N=52) and 14% in AC (N=36)).

The existence of variable time between the completion of Q1 and Q2 should be acknowledged as a limitation of this study due to its potential to influence the patients' retention regarding factual information received through counselling, as well as their state anxiety regarding their involvement with prenatal diagnosis.

Other limitations of this study involve whether or not it was the patient's first experience with having a prenatal diagnostic procedure (AMN/CVS), and which procedure was elected in this pregnancy. Patients who had experienced having a prenatal diagnostic procedure in a previous pregnancy were included in this study because it was their knowledge of risks associated with their present pregnancy that was relevant. Nonetheless, these patients may not have been as anxious as the patients who had never experienced having a procedure because they may have known what to expect. In addition to this, the choice of procedure may have also played a role regarding the patients' state anxiety in each group. For example, the patients who had had an amniocentesis would have been further along in their

pregnancy than those who had had a CVS, and would, therefore, have had more time to experience fetal-maternal bonding through movement or vision by ultrasound (Spencer & Cox, 1988). This attachment may have caused anxiety increases above that of the CVS patients. On the other hand, the patients who had had a CVS at Grace Hospital would have been faced with a slightly higher risk of procedure than the patients who had had an amniocentesis. Therefore, previous experience with prenatal diagnosis and the nature of the method used are variables that have the potential to influence the results of this study.

With reference to the number of informed patients after having had their procedure, there was also the possibility of an intervention effect due to the application of two questionnaires. For example, patients who may not have been informed in Q1 may have been more informed in Q2 because they had asked the individual(s) providing them with genetic counselling the answers to the questions that they did not know. Therefore, patients may have done better in Q2 <u>because</u> they had taken Q1.

The issue of missing data was a common concern for several questions in this survey, especially with respect to the questions that asked patients to recall their risks associated with having an abnormal baby. A poor response rate may cause a problem in the analysis because it can place

doubt on the interpretation of results for various questions. Nevertheless, given that the patients in each group responded poorly to only certain questions in both Q1 and Q2, this suggests that it was the information specifically that caused the patients to respond poorly and not the failure of the questionnaire to ask for the information.

Through this writer's experience in meeting with all patients, either before their genetic counselling appointment or immediately before and after their procedure, this researcher collected observational data that may have contributed to the impact in which prenatal diagnosis placed on the patients in each group. First of all, the patients' composure ranged from very pleasant, calm, and cooperative to very upset, nervous, and unfriendly. Those patients whose appointments were later on in the morning or afternoon were often kept waiting prior to procedure for long periods of time, due to previous patients taking longer than expected or emergencies which took precedence over prenatal diagnostic procedures. Finally, those patients who had CVS often experienced great discomfort prior to procedure because of the necessity for a full bladder at procedure. Since all of these observations were found equally among patients in all three groups, the potential to influence the patients' state anxiety or their ability to recall information was consistent.

Conclusion

It has been reviewed in the literature that an assessment of the effectiveness of genetic counselling can be accomplished through measuring how informed patients are regarding the information presented to them in their counselling process. Using this method of assessment, the present findings suggest that informing patients regarding numerical risk estimates is not always what constitutes an effective genetic counselling process.

The consistent lack of recall of information after patients have stated that they had been told the information requested of them suggests that knowledge pertaining to actual risk estimates may not have been important or necessary for patients in making their decision whether or not to have prenatal diagnosis. Alternatively, this information may have been relevant but incomprehensible to the patient in quantitative terms. In the event where patients had understood the information presented to them in a manner other than how it was presented to them, their knowledge and understanding would not have been acknowledged by the questionnaires used in this study. Additional findings related to risk perception, reasons for "having" and "wanting" prenatal diagnosis, and anxiety felt throughout the process imply that the patients' knowing that some risk was present at all, be it big or small, was what was important

for most patients in this study. These observations coincide with the early investigations made by Lippman-Hand and Fraser (1979), demonstrating patients' binary response to risk information.

Despite the levels of no recall, missing data after confirmation of receiving information, and less than a majority of all patients being informed post-counselling, the majority of patients in each group were very satisfied with the genetic counselling that they had received. This suggests that the decision to have prenatal diagnosis is not always based on the factual information (numerical risk estimates) provided to them in counselling. Therefore, although not all of the patients in each group were informed regarding risks associated with AMA and prenatal diagnosis, their satisfaction with the counselling received suggests that their needs were met with or without such information.

In conclusion, the effectiveness of the genetic counselling process regarding age-related risks and prenatal diagnosis does not appear to be related to those who provided the patients with genetic counselling or the patients' ability to recall factual information. Counselling efforts by primary care physicians within the community and by medical geneticists and genetic counsellors in the Department of Medical Genetics may require alternative methods in presenting the factual information relevant to AMA

counselling. For example, the numerical risk estimates may be presented to women in a qualitative rather than a quantitative manner so that women might be able to appreciate their risks more appropriately.

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APPENDIX A

(i) Questionnaire 1

(ii) Questionnaire 2

PRENATAL DIAGNOSIS COUNSELLING INFORMATION SURVEY
DEPARTMENT OF MEDICAL GENETICS, UNIVERSITY HOSPITAL
DEPARTMENT OF FAMILY PRACTICE, GRACE HOSPITAL

Principal Investigator: Desmond Dwyer, M.D., & Barbara McGillivray, M.D.

QUESTIONNAIRE #1

This questionnaire is designed to obtain information which will help us ensure that the counselling presentations provide the information that parents need to make informed decision concerning prenatal testing. Please ensure that you have answered all questions as accurately as possible. If there is anything asked of you that you do not have an answer to or are uncertain of, please indicate so. We estimate that this should take you approximately 15 minutes. Completion of the questionnaire implies that you have consented to participate in the study. You have the right to refuse to participate or withdraw from the study at anytime without risk of jeopardizing further treatment or medical care.

For our reference only, we need identification on the questionnaire. We suggest that you use your mother's initials and your father's first name, since these are items you will be able to recall but that do not identify you. The telephone number will only be used to contact you should we not receive your post-counselling questionnaire.

Your mother's initials _____

Your father's first name _____

Your telephone number _____

1. What age are you? _____

2. How many times have you been pregnant? _____

   Losses _____

   Liveborn _____

   Living children: Boys _____ Girls _____

3. How long did it take you to get pregnant this time?

   _____ Less than 1 year _____ 2 to 5 years

   _____ 1 to 2 years _____ Greater than 5 years

4. Did you plan this pregnancy? Yes _____ No _____

5. What is your ethnic background?

Caucasian	East Indian	Chinese	Other

6. What is your religious affiliation?

Protestant	Roman Catholic	Buddhist	
Sikh	Hindu	Jewish	
Islam	Fundamentalist	Other	Nil

7. What level of education do you have?

_____ Did not complete high school

_____ Completed high school

_____ Post-secondary _____ Number of years

8. Had you considered having prenatal diagnosis before you were given information from your doctor/medical geneticist or other source?

_____ Yes

_____ No

9. Why did you come for prenatal diagnosis? Please number in order the reasons as: (1) most important, (2) second most important, (3) etc. If any of these reasons do not apply to you, please leave them blank.

_____ You or your family have a history of a genetic disorder.

_____ You are in a high risk age group.

_____ You wish to hear more about your risks and then decide whether or not to have prenatal diagnosis.

_____ You wish to know the sex of the baby.

_____ You do not want to have an abnormal baby regardless of the risk.

10. Who was most responsible for giving you information about genetic problems (eg. specific risks and/or discussed prenatal diagnosis)?

_____ Family doctor

_____ Obstetrician

_____ Family and friends

_____ Media (T.V., radio, newspaper, magazine)

_____ Other (please specify) _____

_____ I have not received any information about genetic problems

11. Has anyone helped you in making a decision to have prenatal diagnosis? Please number in order the following sources from: (1) the most helpful to, (5) the least helpful. If no one has helped you to make a decision, please leave the spaces blank.

_____ Family doctor

_____ Obstetrician

_____ Family and friends

_____ Media (TV, radio, newspaper, magazine)

_____ Other sources (please specify) _____

12. a) Have you been told your risks of having a baby with Down Syndrome?

_____ No

_____ Yes. If yes, what is the approximate risk (for example only, 1/2 or 1/10,000).

at age 30 _____

at age 40 _____

at your age _____

b) Do you feel that your risk is high or low?

_____

_____

c) How confident are you in your responses to this question? Please circle one.

very confident	moderately confident	pretty sure	not at all confident
1	2	3	4

13. a) Have you been told your risks of having a baby with a neural tube defect (eg. spina bifida)?

_____ No

_____ Yes. If yes, what is the approximate risk?

at age 30 _____

at age 40 _____

at your age _____

b) Do you feel that your risk is high or low?

_____

_____

c) How confident are you in your responses to this question? Please circle one.

very confident	moderately confident	pretty sure	not at all confident
1	2	3	4

14. a) What sorts of problems do people with Down syndrome have?

_____

_____

b) What sorts of problems do people with a neural tube defect have?

_____

_____

15. What would be the most significant problem for you if you had a baby with Down syndrome?

_____

_____

_____

16. Please circle what you think is the approximate risk of having a miscarriage in the first 12 weeks of pregnancy if you have no prenatal test.

1/2	1/5	1/50	1/250
1/500	1/1000	no risk	don't know

17. Please circle what you think is the risk of having a miscarriage from the following tests.

a) Amniocentesis (needle sampling of the fluid surrounding the baby in the womb)

1/5	1/50	1/100	1/200
1/500	1/1000	no risk	don't know

b) C.V.S. (chorionic villus sampling, insertion of a tube through the opening in the womb to sample the baby's placenta)

1/5	1/50	1/100	1/200
1/500	1/1000	no risk	don't know

c) Ultrasound (sonogram, sound wave test)

1/5	1/50	1/100	1/200
1/500	1/1000	no risk	don't know

18. At the present, would you choose to have the following tests? Please check which response best describes how you feel about each test.

	no	possibly	probably	yes	don't know much about it
Amniocentesis					
C.V.S.					
Ultrasound					

19. If there are certain tests that you would not choose to have, please indicate which ones and explain why you would not have them.

_____

_____

_____

20. Why would you choose to have prenatal diagnosis? Please number in order the reasons as: (1) most important, (2) second most important, (3) etc. If any of these reasons do not apply to you, please leave them blank.

_____ To have the option of terminating an abnormal pregnancy.

_____ To relieve my anxiety by knowing that the baby's chromosomes are normal.

_____ To prepare for an abnormal child.

_____ Other (please specify) _____

_____

21. a) How anxious are you feeling right now?

Not at all	Somewhat	Moderately so	Very much so
1	2	3	4

b) Which item from the list best describes your feelings right now? Please select only one.

_____ Wonderful
_____ Steady
_____ Comfortable
_____ Fine
_____ Indifferent
_____ Didn't bother me
_____ Timid
_____ Unsteady
_____ Unsafe
_____ Nervous
_____ Worried
_____ Frightened
_____ Panicky
_____ Scared stiff

COMMENTS _____

_____

_____

Thank you very much for your time and cooperation.

GENETIC COUNSELLING INFORMATION SURVEY
DEPARTMENT OF MEDICAL GENETICS, UNIVERSITY HOSPITAL
DEPARTMENT OF FAMILY PRACTICE, GRACE HOSPITAL

Principal Investigator: Desmond Dwyer, M.D., & Barbara McGillivray, M.D.

QUESTIONNAIRE #2

Now that the prenatal test is completed, a post-procedure questionnaire is designed to allow you to reflect over your experience here. Please ensure that you have answered all questions as accurately as possible. If there is anything asked of you that you do not have an answer to or are uncertain of, please indicate so. We estimate that this should take you approximately 10 minutes. Completion of the questionnaire implies that you have consented to participate in the study. You have the right to refuse to participate or withdraw from the study at anytime without risk of jeopardizing further treatment or medical care.

For our reference only, we need identification on the questionnaire. We suggest that you use your mother's initials and your father's first name, since these are items you will be able to recall but that do not identify you. The telephone number will only be used to contact you should we not receive your post-counselling questionnaire.

Your mother's initials _____

Your father's first name _____

Your telephone number _____

1. Was there anything you hadn't expected from the prenatal test procedure?

_____

_____

_____

2. Read each statement and then circle the appropriate statement indicating how you feel right now. There are no wrong answers. Please give a response to every one.

	NOT AT ALL	SOMEWHAT	MODERATELY SO	VERY MUCH SO
I feel calm	1	2	3	4
I feel secure	1	2	3	4
I am tense	1	2	3	4
I am regretful	1	2	3	4
I feel at ease	1	2	3	4
I feel upset	1	2	3	4
I am presently worrying over possible misfortunes	1	2	3	4
I feel rested	1	2	3	4
I feel anxious	1	2	3	4
I feel comfortable	1	2	3	4
I feel self-confident	1	2	3	4
I feel nervous	1	2	3	4
I am jittery	1	2	3	4
I feel "high strung"	1	2	3	4
I am relaxed	1	2	3	4
I feel content	1	2	3	4
I am worried	1	2	3	4
I feel over-excited and "rattled"	1	2	3	4
I feel joyful	1	2	3	4
I feel pleasant	1	2	3	4

3. a) Have you been told your risks of having a baby with Down syndrome?

_____ No

_____ Yes If yes, what is the approximate risk (for example only, 1/2 or 1/10,000).

at age 30 _____

at age 40 _____

at your age _____

b) Do you feel that your risk is high or low?

_____

_____

c) How confident are you in your responses to this question? Please circle one.

very confident	moderately confident	pretty sure	not at all confident
1	2	3	4

4. a) Have you been told your risks of having a baby with a neural tube defect (eg. spina bifida)?

_____ No

_____ Yes If yes, what is the approximate risk?

at age 30 _____

at age 40 _____

at your age _____

b) Do you feel that your risk is high or low?

_____

_____

c) How confident are you in your responses to this question? Please circle one.

very confident	moderately confident	pretty sure	not at all confident
1	2	3	4

5. a) What sorts of problems do people with Down syndrome have?

_____

_____

b) What sorts of problems do people with a neural tube defect have?

_____

_____

6. What would be the most significant problem for you if you had a baby with Down syndrome?

_____

_____

_____

7. Please circle what you think is the risk of having a miscarriage in the first 12 weeks of pregnancy if you have no prenatal test.

1/2	1/5	1/50	1/250
1/500	1/1000	no risk	don't know

8. Please circle what you think is the risk of having a miscarriage from the following tests.

a) Amniocentesis (needle sampling of the fluid surrounding the baby in the womb)

1/5	1/50	1/100	1/200
1/500	1/1000	no risk	don't know

b) C.V.S. (chorionic villus sampling, insertion of a tube through the opening in the womb to sample the baby's placenta)

1/5	1/50	1/100	1/200
1/500	1/1000	no risk	don't know

c) Ultrasound (sonogram, sound wave test)

1/5	1/50	1/100	1/200
1/500	1/1000	no risk	don't know

9. Please circle one only.

a) How anxious are you feeling right now?

Not at all	Somewhat	Moderately so	Very much
1	2	3	4

b) How anxious are you regarding the test results?

Not at all	Somewhat	Moderately so	Very much
1	2	3	4

10. Which item from the list best describes your feelings right now? Please select only one.

_____ Wonderful

_____ Steady

_____ Comfortable

_____ Fine

_____ Indifferent

_____ Didn't bother me

_____ Timid

_____ Unsteady

_____ Unsafe

_____ Nervous

_____ Worried

_____ Frightened

_____ Panicky

_____ Scared stiff

11. In the event of a new pregnancy, would you choose to have the following tests? Please check which response best describes how you feel about each test.

	no	possibly	probably	yes	don't know much about it
Amniocentesis					
C.V.S.					
Ultrasound					

12. If there are certain tests that you would not choose to have, please indicate which ones and explain why you would not have them.

_____

_____

_____

13. How satisfied are you with the overall counselling you have received? Please circle one.

Very satisfied			Quite disappointed
1	2	3	4

COMMENTS _____

_____

_____

Thank you very much for your time and cooperation in filling out this questionnaire. Your thoughts and suggestions are much appreciated.

APPENDIX B

(i) Risk of Down's Syndrome

(ii) Risk of Neural Tube Defects

CATEGORIES FOR EVALUATING PATIENT RECALL
FOR THE RISK OF DOWN'S SYNDROME

AGE	RISK ESTIMATE°	INFORMED	SOMEWHAT INFORMED	UNINFORMED
30	1/1140	± 1/300 (1/840-1/1440)	± 1/500 (1/640-1/1640)	> ± 1/500
35	1/360	± 1/100 (1/260-1/460)	± 1/150 (1/210-1/510)	> ± 1/150
36	1/282	± 1/100 (1/182-1/382)	± 1/150 (1/132-1/432)	> ± 1/150
37	1/220	± 1/75 (1/145-1/295)	± 1/100 (1/120-1/320)	> ± 1/100
38	1/170	± 1/75 (1/95-1/245)	± 1/100 (1/70-1/270)	> ± 1/100
39	1/130	± 1/50 (1/80-1/180)	± 1/75 (1/55-1/205)	> ± 1/75
40	1/100	± 1/50 (1/50-1/150)	± 1/75 (1/25-1/175)	> ± 1/75
41	1/80	± 1.35 (1/45-1/115)	± 1/50 (1/30-1/130)	> ± 1/50
42	1/60	± 1/60 (1/40-1/80)	± 1/35 (1/24-1/95)	> ± 1/35
43	1/48	± 1/15 (1/33-1/63)	± 1/20 (1/28-1/68)	> ± 1/20
44	1/38	± 1/10 (1/28-1/48)	± 1/15 (1/23-1/53)	> ± 1/15
45	1/30	± 1/5 (1/25-1/35)	± 1/10 (1/20-1/40)	> ± 1/10

° = data derived from B.C. data for Down's syndrome and Hook, E.B. (1981), Obstetrics & Gynecology, 58, 282-285.

CATEGORIES FOR EVALUATING PATIENT RECALL
FOR THE RISK OF NEURAL TUBE DEFECTS

AGE	RISK ESTIMATE°	INFORMED	SOMEWHAT INFORMED	UNINFORMED
All Ages	1/700	± 1/100 (1/600-1/800)	± 1/200 (1/500-1/900)	> ± 1/200

° = data derived from McBride, M.L., (1979). Sib risks of anencephaly and spina bifida in British Columbia. American Journal of Medical Genetics, 3, 377-387.