THE REPRODUCTIVE DECISION AFTER GENETIC COUNSELING

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THE REPRODUCTIVE DECISION AFTER GENETIC COUNSELING

De beslissing over het al dan niet krijgen van kinderen
na erfelijkheidsadvies

PROEFSCHRIFT

TER VERKRIJGING VAN DE GRAAD VAN DOCTOR

AAN DE ERASMUS UNIVERSITEIT ROTTERDAM

OP GEZAG VAN DE RECTOR MAGNIFICUS

PROF. DR. C.J. RIJNVOS

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Aan Sten
Aan mijn ouders

CONTENTS

CHAPTER I.
INTRODUCTION AND AIM OF THE STUDY

Genetic and congenital disorders are now the prime cause of infant mortality and morbidity in the western industrialized countries. This is mainly due to a reduction in the incidence of infectious diseases and malnutrition brought about by economic developments, improved hygienic conditions, vaccination procedures, and the availability of antibiotics. From the end of the sixties the importance of genetics, the development of new methods for early prenatal and postnatal diagnosis, as well as carrier detection and genetic counseling[1] has increasingly been recognized in the medical world as well as in society at large (Galjaard, 1989). Moreover, people have developed a more conscious attitude towards reproduction. The introduction of the pill, the legalization of induced abortion and the change in social position of women with new possibilities for self-fulfilment, all contributed to this change in attitude towards reproduction (Galjaard, 1990). Nowadays, couples tend to restrict their family to two children at most and many want to be fully informed about their risk of having a(nother) affected child and about ways and means to prevent the birth of an affected child (ter Haar and Niermeijer, 1982; Galjaard, 1989).

In 1975, the American Society of Human Genetics defined genetic counseling as a communication process which deals with problems associated with the occurrence, or the risk of recurrence, of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained persons to help counselees:

1. comprehend the medical facts, including the diagnosis, the probable course of the disorder, and the available management;
2. appreciate the way heredity contributes to the disorder, and the risk of recurrence in specified relatives;
3. understand the options to deal with the risk of recurrence;
4. choose the course of action which seems appropriate to them in view of their risk and family goals and to act in accordance with that decision;
5. make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.

(Ad Hoc Committee on Genetic Counseling, 1975).

This definition of genetic counseling focuses on the importance of a precise diagnosis

[1] Except for the introduction and general discussion, all chapters in this thesis were initially prepared for publication in either American or English journals. Hence, inconsistencies in spelling.

and the communication of information. The principal goal of this communication process is to help counselees make "the best possible adjustment" and an "informed decision". This implies a nondirective approach, contradicting earlier beliefs that the only aim of genetic counseling was to prevent genetic disorders (Carter, 1969). Genetic counseling may have a preventive effect when it leads to a decision to: (a) refrain from having children; (b) use another person's gametes for fertilization; (c) raise adoptive or (d) foster children; (e) undergo prenatal diagnosis with the possibility of selective abortion. The last option is often erroneously viewed as inextricably linked with genetic counseling, even though many hereditary disorders cannot be detected prenatally.

Genetic counseling, as referred to in this thesis, entailed diagnostic work-up, analysis of family history, discussion of the risk of occurrence or recurrence, and various options to reduce risks[2]. The number of requests for genetic counseling handled by the Department of Clinical Genetics in Rotterdam rose from 351 in 1980 to 500 in 1984 (Annual Reports Foundation Clinical Genetics Rotterdam 1980-1983; 1984-1987), stabilizing at that level with 529 in 1989. The increase of 42% in just four years might be explained by several developments. Firstly, major advances in basic research created new possibilities for early diagnosis in the fetal or neonatal phase, treatment and counseling. Secondly, the scope of prenatal diagnosis has been widened with the introduction of chorionic villus sampling and fetal ultrasound imaging.

Approximately 40% of couples seeking genetic counseling at our Department have an affected child, 30% have an affected relative, for 10% either spouse is affected, while the remaining 20% come for a variety of reasons. A small number of couples seek genetic counseling because of consanguinity or teratogenic risk estimation, the latter mostly because of maternal use of medication or exposure to drugs, chemical agents, radiation, etc.

Genetic counseling is requested for a large variety of disorders. There are no objective standards for the severity or burden of a disorder (Emery, 1984; Ekwo et al., 1987) and therefore the personal perception of counselees was the determining factor in this dissertation.

The facilities for diagnosis of complex genetic disorders and malformation syndromes are important aspects of a clinical genetic service.
These facilities are relevant for parents, patients and referring physicians, in planning the management of the disorder, and as a basis for information about the prognosis and genetic implications of the disorder in the family. This explains why some couples with

[2] Excluding cases of uncomplicated genetic counseling, e.g. involving chromosomal disorders, metabolic diseases, or advanced maternal age.

an affected child request genetic counseling even though they do not want more children, to be informed about their child's disorder and about any risk of recurrence in future grandchildren.

Apart from the need to learn the medical and genetic facts of the disorder in the family, there are often equally important psychological aspects to be covered by the counseling-process. Couples are generally more concerned about the cause and nature of the disorder in their child or family than about the risk of recurrence or occurrence even though they claim to request genetic counseling for future offspring (Kessler and Kessler, 1988). Counselees may want to talk about their experiences with the genetic disease, because the occurrence of a genetic disorder in the family can be very disruptive psychologically. This disruption may have a negative effect on counselees' self-esteem and may attack the inner desire for personal stability (Broder and Trier, 1985; Messner and Schmidt, 1986; Porter et al., 1986; Miller et al., 1986/87; Seligman, 1987; Kessler and Kessler, 1988). Therefore, they want to be supported in their effort to make sense of it all and often feel the need to be reassured and validated by the counselor (Kessler and Kessler, 1988).

Guilt feelings may be induced by various mechanisms in parents and healthy siblings. Parents may feel guilty towards their affected child for transmitting the "defective" gene (Targum, 1981; Kessler et al., 1984; Broder and Trier, 1985; Miller, 1986/87). Alternatively, parents may fear their child's defect is due to something they did that endangered the pregnancy, e.g. taking an aspirin during pregnancy. Parents may also feel guilty because they experience the birth of their affected child as a punishment, e.g. for supposed misbehavior in the past. This implies that the recurrence of the disorder in a future child may be prevented by a change in behavior. Telling parents that they are not guilty of their child's defect takes away their power to avert the disorder. Guilt may serve as a defence against being powerless and as such a sense of guilt would be easier to bear than the feeling of being powerless (Kessler et al., 1984).

Healthy children may experience feelings of "survivor" guilt towards an affected sibling or parent; the healthy children may fantasize why they themselves were spared (Kennedy, 1985; Drotar and Crawfurd, 1985). In healthy children, guilt feelings might also be engendered by the anger towards parents and affected sibling because they feel they get less attention (Seligman, 1987). Healthy children may also feel guilty towards their parents because they blame them for having transmitted the "defective" gene.

The birth of a defective child may deprive parents from an important aspect of their life cycle (Kessler, 1979). A healthy child provides the opportunity for parents to relive their own developmental past through identification with the child. In this way parents achieve a new level of maturity (Benedek, 1970).

Genetic disorders may induce a sense of embarrassment. Consequently, a considerable amount of time and effort is required before the principle goal of genetic counseling can be realized, which is to help counselees make "the best possible adjustment" and reach an "informed decision". The procedure of genetic counseling at our department is as follows: Genetic counseling is generally provided in two consultations of 1-1½ hours each. The first consultation is used for a thorough evaluation of the diagnosis including a complete family history. Counselees' participation is essential in soliciting pertinent information from relatives. The written informed consent from the relatives will enable the clinical geneticist to obtain medical information. In some cases the relatives have to participate personally, e.g. submit to a physical examination or give blood for chromosome or DNA-studies. At the second consultation counselees are informed of the diagnosis and prognosis regarding the disorder in the family, the availability of treatment, the risk of occurrence or recurrence, and the various options available to counselees are discussed. The counselor will make every effort to facilitate the decision-making process. It is important that counselees perceive the decision appropriate to their life situation. The genetic counselor will look for any marital disfunctioning and guilt feelings (Kessler and Kessler, 1988). Marital disfunctioning in this context applies to a lack of communication between spouses and the absence of adequate means of conflict resolution. All counselees receive a comprehensive, written summary of the information presented and discussed during counseling.

In order to facilitate the reproductive decision-making process the genetic counselor should be aware of the factors that influence this process and the final decision. A review of the literature concerning several of these factors and the frequently contradictory conclusions is presented in Chapter II.

History of genetic counseling and its psychological implications

Up till the sixties, genetics held a minor position in the medical curriculum; medical genetics was studied in a few basic science institutes and only rarely in medical schools. A few clinicians specialized in genetic disorders applying their field. Risk estimation was not always based on an exact diagnosis or correct knowledge of genetics, implying that the genetic risk quoted might be incorrect.

In the Nertherlands, the University of Nijmegen was the first to start a clinical genetics unit (1971). The staff included a pediatrician/clinical geneticist, a medical cytogeneticist, a social worker, and a corps of consulting medical specialists (ter Haar and Niermeijer, 1982).

In 1977, the Dutch Health Council proposed a national, collaborative scheme for regional Clinical Genetic Centres, closely associated with Human Genetics Departments

of Medical Schools and University Hospitals. These Clinical Genetic Centers were to provide prenatal and postnatal cytogenetic analysis, diagnosis of metabolic diseases by metabolite and enzyme studies, and genetic counseling. Since 1979 pre-and postnatal cytogenetic analysis and diagnosis of metabolic diseases were paid for by the Sick Fund (collective health insurance system financed by employers, employees and government) and most private health insurance companies. Between 1979 and 1984 genetic counseling as defined by the Ad Hoc Committee on Genetic Counseling (1975) and prenatal biochemical diagnosis were funded by a national plan for extraordinary medical expenses (AWBZ). Since 1984 these services are also paid for by the Sick Fund and most private health insurance companies (Annual Report Foundation Clinical Genetics Rotterdam, 1984-1987). This funding concerns genetic counseling of complex nature which means that this cannot be provided by the family doctor or physician (Galjaard, 1986). Diagnosis of genetic diseases by DNA-analysis, which was introduced in 1985, resorted since 1-1-87 under the AWBZ with a developmental grant for four clinical genetic centers.

In Rotterdam, prenatal and postnatal cytogenetic analysis, prenatal and postnatal diagnosis of genetic metabolic diseases and genetic counseling has been provided from the early seventies involving the Departments of Cell Biology and Genetics in collaboration with the Departments of Obstetrics and Gynecology, Pediatrics, Neurology, Biochemistry etc. After the establishment of adequate financial support for diagnostic and counseling activities a separate Department of Clinical Genetics was formed, with the establishment of the Foundation of Clinical Genetics Rotterdam as the organizational and administrative body in 1979 (Annual Report Foundation Clinical Genetics Rotterdam 1980-1983). In 1987 clinical genetics became a separate medical specialism.

In the early seventies, the goal of genetic counseling was to provide medical and genetic information focusing on prevention of genetically determined disorders (Carter, 1969). It was believed that a rational attitude of the counselor facilitated the decision-making process. Emotional overtones, affects and internal conflicts of the counselees were not discussed in the literature, but probably often during consultation, as these were considered to impede the decision-making process. Psychological issues did not receive much attention (Kessler, 1979).

Towards the end of the seventies, more emphasis was given to the psychological aspects of genetic counseling for several reasons. Prevention as a major goal of genetic counseling proved biologically unattainable particularly as the risk of having an affected child could not always be foreseen. A congenital defect may arise from spontaneously occurring chromosomal aberrations, interactions of genetic and non-genetic factors, teratogenic insults, newly arisen autosomal dominant mutations, nonsymptomatic carrier

status of one or both parents for an X-linked or autosomal recessive disorders, or complications of delivery. Moreover, the means of detection and prevention were limited. Furthermore, the notion grew that prevention per se should not be the only goal of genetic counseling as it meant forcing counselees to use risk restricting means, such as refraining from having children, using other person's gametes, using prenatal diagnosis with selective abortion etc. The concept of having a risk different from other people, the provision of the genetic facts by the genetic counselor, the (presumed) reactions in society were all factors threatening counselees freedom of choice. Moreover, counselees frequently resisted using these risk restricting means (Kessler, 1979), particularly for relatively mild disorders (Epstein, 1979) or disorders variable penetrance such as hemophilia (Barrow et al., 1982; Markova et al., 1984; Beeson and Golbus, 1985). The psychological burden of terminating a pregnancy in case of fetal abnormality was also recognized (Blumberg et al., 1975; Thomassen-Brepols, 1985). It was suggested that couples deciding to terminate a pregnancy receive additional and appropriate counseling (Epstein, 1979). Moreover, genetic counselors and clinical geneticists found that counselees did not always opt for what the genetic counselor felt was the "reasonable" course of action, such as deciding not to have children when the genetic risk was considered high (arbitrary $>$ 10%). Consideration of the personal reasons motivating these couples to have a(nother) child made their decision understandable (Emery, 1984).

It was recognized that genetic information was never neutral but emotionally charged for the counselees. Furthermore, the awareness grew that genetic disorders could have long-lasting, psychological consequences for counselees and their relatives, such as chronic anxiety about the risk of occurrence or recurrence in a future child (Kessler, 1979). More insight was also obtained regarding the psychological aspects of the genetic counseling process. For example, counselees may initially deny or suppress the possibility of a future child being affected. The use of a defense mechanism was recognized as a necessary step in the process of accepting the hereditary disorder (Kessler, 1979).

The emphasis on psychological aspects and ethical principles of genetic counseling resulted in a shift in objectives with its emphasis on the need for information and on support for counselees in the adjustments they have to face. This definition has been accepted in many countries (Wertz and Fletcher, 1988). Gradually, the informed and free decision of counselees was recognized as the major goal of genetic counseling. It became the task of the genetic counselor to facilitate the decision-making process (Emery, 1984). This change to a nondirective approach, encouraging counselees to make their <u>own</u> decision, required insight into the factors that might facilitate or complicate the decision-making process. In the present study, nearly half the couples experienced problems in the

decision-making process (Chapter V). The findings in Chapter V indicated how the clinical geneticist could facilitate the decision-making process, by paying attention to specific factors.

Impact of genetic diagnosis and technology

The advance of genetic technology has been enormous in just a few decades. Following Lejeunes' discovery in 1959 that Down's syndrome was caused by an extra chromosome 21, a few hundred syndromes combining mental retardation with various physical abnormalities have proved to be caused by chromosomal aberrations (Schinzel, 1984). The number of diseases and/or syndromes that could be identified as a mendelian trait with an autosomal dominant, recessive or X-linked recessive mode of inheritance, increased from 1487 in 1966 to 4344 in 1988 (McKusick, 1988). This increase mainly resulted from better methods of diagnosis. The nature of the genetic defect is identifiable at either protein or enzyme level in approximately 700 disorders. At the DNA-level, the precise error in any of approximately 50,000-100,000 human genes involved in a single mendelian disorder has been identified for a small number of disorders only, such as hemoglobinopathies, cystic fibrosis, retinoblastoma, etc. However, refined methods of assignment of genes to individual human chromosomes (localization) have opened ways to utilize restriction fragment length polymorphisms (RFLP's) linked to the disease gene in the DNA, as markers for these genes. New methods of diagnosis and carrier testing, comparing RFLP patterns from affected and healthy family members, evolved rapidly. Gene cloning and function analysis, which would give precise insight into the nature of the genetic disorder itself, is much more time-consuming and is still at the preliminary stage.

Diagnosis of a genetic disorder using RFLP's is available for over 200 genetic diseases. With more precise localization of the disease gene, RFLP analysis will be supplanted by mutation analysis. Recent successes have been booked in this respect for cystic fibrosis and neurofibromatosis.

For some diseases with an autosomal dominant mode of inheritance such as Huntington's disease, Myotonic Dystrophy presymptomatic testing through DNA-analysis has become available. This test provides the opportunity to establish carriership long before any clinical features become apparent. It is evident that knowledge about a disabling disease 20 years ahead of clinical manifestations will cause a heavy psychological burden (Kessler et al., 1987; Meissen et al., 1988; Galjaard, 1989; Tibben et al., 1990).

DNA-analysis has also widened the scope for prenatal diagnosis. Before the early eighties, carrier detection for X-linked Duchenne Muscular Dystrophy (=DMD) had

serious limitations. This disorder affects boys, who die of respiratory and/or cardiac weakness before or around their 20th birthday. Carrier detection by means of creatinine kinase (a muscular protein) in female relatives was not always dependable. For couples at risk for DMD, sex determination was the only available option of prenatal diagnosis. They had to make the difficult decision to restrict their family to girls. Such enormously painful decisions to terminate a pregnancy in the face of great uncertainty have now come to an end. Since 1985, informative flanking markers have become available for the DMD gene. The recent discovery of deletions in the region of the DMD gene has resulted in cloning of the gene itself. This breakthrough has provided an increased precision in the methods of (prenatal) diagnosis and carrier-testing, which can often be performed at the level of the mutation specific for an individual family (Bakker, 1989). The result is impressive. Carriers of DMD may now have healthy sons, and only need to consider termination of a pregnancy when DMD is diagnosed in the 10-12th week of gestation by means of chorionic villus sampling and DNA-analysis in a male fetus.

In cystic fibrosis, similar improvements of diagnosis and carrier testing came about with the localization of the gene in chromosome 7 and - as recent as last year - the mutation occurring in the majority of patients (Kerem et al., 1989; Riordan et al., 1989; Rommens et al., 1989). These findings may eventually open the way to carrier detection of this autosomal recessive disorder in young caucasian adults. In a caucasian population, the heterozygote frequency is approximately 1:25 to 1:30.

The largest category of congenital malformations are caused by certain genetic factors in combination with environmental influences. Examples of these genetic disorders are spina bifida, congenital heart disease, club feet, cleft lip/cleft palate, etc. In addition, most major diseases occurring in adulthood have a similar multifactorial mode of inheritance, e.g. cancer, cardiovascular diseases, diabetes mellitus and various neurological and psychiatric diseases (Galjaard, 1988).

Uncovering the influence of a multitude of genetic factors and the interaction with non-genetic factors will be an enormous challenge for genetic research in the future.

Investigation of the psychological aspects of genetic counseling shows a gradual shift towards emphasis of the importance of individual freedom and recognition of problems due to new options that have become available with technological advancement. Our study was carried out in the midst of these developments.

AIMS AND METHODS OF THE STUDY

The main aims of the studies described in this thesis were to monitor the transfer of information during genetic counseling and to investigate the adequacy of the existing strategies for supporting counselees in their decision-making process, and if necessary, to

devise new strategies. This study is a continuation of a project at our Department investigating the psychosocial aspects of genetic technology. The first part of this project concerned the assessment of the psychosocial aspects of prenatal diagnosis (Thomassen-Brepols et al., 1982; Thomassen-Brepols et al., 1983; Thomassen-Brepols, 1985). In the present study the psychosocial aspects of genetic technology as provided during genetic counseling are assessed in relation to the reproductive decision and its process.

A comprehensive follow-up study was carried out in 164 couples 2-3 years after genetic counseling, focusing on the psychological aspects of the postcounseling decision-making process. The objectives were to investigate

a. which factors influenced the reproductive decision;
b. whether it was possible to identify the reproductive decision with a limited number of factors;
c. what kind of problems counselees experienced in their decision-making process,;
d. which factors were associated with these problems;
e. how counselees eventually came to a decision.

Enrollment in the study was limited to couples that requested genetic counseling for their own offspring.

The study was quantitative as well as qualitative. The quantitative part involved interviewing 164 couples 2-3 years after genetic counseling. A questionnaire with 91 items was used to reveal the factors that had influenced the reproductive decision either singly or combined. The influence of various combinations of two factors on the reproductive decision of diverse groups of couples were studied, e.g. couples with a risk over 15%, couples not eligible for prenatal diagnosis, etc. (see chapter III).

It proved impossible to identify the reproductive decision on the basis of single factors or any combination of two factors. Using combinations of eight factors enabled the construction of a model to identify the reproductive decision. The model also showed which factors were most important for the reproductive decision (see chapter IV).

Couples may experience a number of problems in their decision-making process such as (1) particular difficulty in reaching a decision, (2) inability to resolve all doubts concerning that decision, or (3) total inability to reach a decision. Insight into the factors related to these problems might indicate which couples would benefit from additional counseling (see chapter V).

The qualitative part of the study involved an in-depth interview of 30 couples, randomly selected from the total number of 164. The objective was to explore the psychological characteristics of the decision-making process. The interviews were recorded and evaluated by several independent judges, who noted the presence or absence

of any such characteristics. We looked for confirmation of the results of Lippman-Hand and Fraser (1979a; 1979b) who found that the postcounseling reproductive decision-making process was a cognitive one (see chapter VI).

Co-operation of both spouses was an essential element of the study in view of the fact that both spouses were involved in the reproductive decision. Other workers have generally interviewed women only because of more easy accessibility. Interviewing both husband and wife provided insight into the full extent of the problems related to the decision-making process including any marital disagreement in this respect.

The present study was the first one of its kind to be reported in the Netherlands and as such will enable cross-cultural comparison with similar follow-up studies carried out in other countries.

The results show that the reproductive decision-making process is too complex for the decision to be determined by one single factor.

A considerable proportion of couples had experienced problems in the decision-making process. In contrast to the results of other studies the reproductive decision-making process was generally unstructured.

The findings indicate how professionals may support individuals confronted with a genetic disorder in the family.

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CHAPTER II.
REPRODUCTIVE PLANNING AFTER GENETIC COUNSELLING: A PERSPECTIVE FROM THE LAST DECADE

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ABSTRACT

Studies from the last decade on factors influencing reproductive planning after genetic counselling were reviewed. Increased possibilities of DNA-analysis and prenatal diagnosis might have brought about a shift in paramountcy of factors influencing reproductive planning after genetic counselling. The burden of the disorder remained important. In contrast to the literature in the seventies, the magnitude of the genetic risk was no longer of major importance for reproductive planning. Instead, the interpretation of the risk as high or low and the desire to have children appeared to be paramount. The impact of new developments in DNA-analysis in prenatal diagnosis and presymptomatic testing will be an important subject for future studies on factors influencing reproductive planning.

KEY WORDS: Decision-making, follow-up, family planning, genetic counselling, prenatal diagnosis, recurrence risk, reproductive planning, reproductive uncertainty, risk interpretation.

INTRODUCTION

The aim of genetic counselling is to inform couples about the nature of a mental or physical handicap in the family and its risk of occurrence or recurrence. An important aspect of genetic counselling is to assist consultands in reaching a decision regarding

future reproductive behaviour which is appropriate to their life situation (Ad Hoc Committee, 1975). For this, it is useful to know which factors influence the decision-making process after genetic counselling. Literature reviews carried out until the end of the seventies (Shaw, 1977; Childs, 1978; Evers-Kiebooms & van den Berghe, 1979) have revealed genetic risk and the burden of a disorder to be major factors. In the last ten years, the possibility of the diagnosis of hereditary disorders has increased. The scope of prenatal diagnosis has been widened with the introduction of chorionic villus sampling and the increased possibility of fetal diagnosis using DNA technology (Galjaard, 1989).

Recently, Kessler (1989) reviewed studies on the impact of genetic counselling on reproductive planning. He posed the question whether precounselling reproductive intentions play a major role in determining postcounselling reproductive plans. The present review of follow-up studies after genetic counselling from the last decade is not specifically focused on the impact of counselling on reproductive planning, but on the associations between a variety of factors and post-counselling reproductive planning.

From the studies reviewed, the univariate association between reproductive planning and relevant single factors was investigated together with new aspects arising from several multivariate studies. Factors influencing the burden of the decision-making process and reproductive uncertainty after genetic counselling are discussed and new avenues for possible future research concerning changes in reproductive planning after genetic counselling are suggested.

FACTORS INFLUENCING REPRODUCTIVE PLANNING AFTER GENETIC COUNSELLING

The magnitude of the genetic risk

The genetic risk is defined as the risk established during genetic counselling that a disorder in the family might occur or recur in an individual or in a future pregnancy. Emphasis is placed upon the risk for a disorder in a future child since this paper will mainly address factors influencing reproductive planning.

The magnitude of the genetic risk was earlier thought to be one of the decisive factors in reproductive planning after genetic counselling (Carter et al., 1971). More recent studies suggest that this process is more complicated. Table I shows that approximately half the couples with a genetic risk of 10% or more, chose to have children (Emery et al., 1979; Bocsknov, 1979; Abramovsky et al., 1980; Sorenson et al., 1987). In a study in the Netherlands (Frets et al., 1990a), this figure rose to 71% for the whole study population; if prenatal diagnosis was not possible, only 52% chose to have

children. This figure is similar to that in the study of Sorenson et al. (1987) which only assessed consultands who were not eligible for prenatal diagnosis. Despite a directive approach to counselling in one centre in West Germany, 10% opted to have children while being aware of a genetic risk of 10% or more (Rott & Petzold, 1981).

TABLE I. Percentages of couples that decided to have children after genetic counselling associated with the genetic risk

Study	Country of couples	Total number	Genetic risk	
			< 10%	≥ 10%
Bocsknov (1979)	USSR	225	86%	43%
Emery (1979)	UK	200	82%	47%
Abramovsky (1980)	USA	212	70%	50%
Czeizel (1981)	HUNG	1841	95%	39%
Rott (1981)	FRG	424	51%	10%
Sorenson (1987)*	USA	181	59%	42%
Frets (1990a)	NETH			
-Entire study population		164	86%	71%
-Population eligible for prenatal diagnosis		71	80%	83%
-Population not eligible for prenatal diagnosis		90	89%	52%

* Only those consultands were assessed who were not eligible for prenatal diagnosis

Furthermore, other studies on various disorders, when the genetic risk was split at other levels, showed that the magnitude of the genetic risk was not directly related to reproductive planning (Bocsknov, 1979; Czeizel et al., 1981; Steele et al., 1986; Somer et al., 1988).

These results show that in non-directive counselling the genetic risk is a poor measure of reproductive planning and is of relative importance (Kessler & Levine, 1987b). Only in the study of Rott & Petzold (1981) did the influence of a directive approach become apparent (Table I). Less compliance was found in Eastern European countries where consultands having a genetic risk of 20% or more were recommended to refrain from reproduction (Bocsknov, 1979; Czeizel et al., 1981).

Overall, where prenatal diagnosis was not available, reproductive planning was strongly influenced by a genetic risk of 25% or more for cystic fibrosis, thalassemia or Huntington's disease in future offspring. A considerable proportion of parents of a child with cystic fibrosis changed their family size or abstained from further childbearing (Kaback et al.,

1984; Passarge et al., 1984). However, these were observations during the time that prenatal diagnosis was not yet available. Clearly, new studies are needed since DNA-tests by chorionic villus sampling became available to parents of a CF-child from 1985 onwards (Harper, 1986).

Of the Cypriot couples in North London who were informed of their 25% risk for thalassemia, before prenatal diagnosis became available, the pregnancy rate fell by half (Modell et al., 1982). The influence of knowing the risk for Huntington's disease on reproductive planning is unclear (McCormack et al., 1983; Carter et al., 1983; Tyler & Harper, 1983).

Results of the studies on the influence of a low genetic risk (< 5%) in parents of children with spina bifida or Down's syndrome for which prenatal diagnosis was available were inconsistent (Evers-Kiebooms, 1980; Laurence & Morris, 1981; Adams et al., 1984; Steele et al., 1986; Boon, 1986). These inconsistencies might be due to the small study populations or the varying degree of acceptance of prenatal diagnosis.

In most of the studies of various disorders, no direct relationship between reproductive planning and genetic risk was found. However, in specific recessive and autosomal dominant disorders where prenatal diagnosis was not available, the proportion of couples choosing to have children dropped significantly. This might be due to the combined effects of genetic risk and the familiarity with a "severe" disorder.

Risk recall

Correct recall of the risk indicates that consultands correctly recall the risk figure or risk range supplied in genetic counselling. Most studies revealed that half to three-quarters of the couples correctly recalled their genetic risk figure or its range (Table II).

It is unknown whether oral or written provision of information influenced the correct recall rate, as only a few studies reported on the method of information transfer. Such data would be helpful, however, to evaluate the methodology of genetic counselling.

The relationship between correct recall and reproductive planning after genetic counselling has rarely been studied. Couples who were planning a pregnancy had no better risk recall than other couples (Sorenson, 1981a; Burns et al., 1984; Frets (unpublished results)).

These limited data does not allow definite conclusions. It might be more informative to assess the influence on reproductive planning of a correct or incorrect recall of a certain risk category (low-high). Couples who know the magnitude of the risk might face an internal conflict between the desire to have children while knowing that a future pregnancy may lead to the birth of a (severely) affected child.

TABLE II. Proportion of couples correctly recalling their genetic risk quoted in genetic counselling

< 20%	approx. 30%	approx. 50%	approx. 75%
Black (1979)[a] [Down's syndrome]	Bocsknov (1979)[c,e]	Emery (1979)	Black (1979)[a] [Mental retardation]
Keltikangas (1983)[b,f]	Czeizel (1981)	Sorenson (1981a)	Springer (1980)[c]
	Keltikangas (1983)[d,f]	Oeting (1982)[a]	Abramovsky('80)[c]
		Evers-Kiebooms ('84)[a]	Seidenfeld (1981)[c]
		Passarge (1984)	Swerts (1987)[a]
			Somer (1988)
			Frets[e,g]

a = it is not clear how disagreement between the spouses were handled
b = scored range of mothers
c = criteria for correctness of risk recall were missing
d = scored range of fathers
e = risk range correctly recalled
f = genetic information given by physician
g = unpublished results

Risk interpretation

Risk interpretation indicates how the magnitude of the genetic risk as supplied during counselling is interpreted by the consultands, i.e. whether the risk is interpreted as high or low. The significance of the interpretation of the risk for reproductive planning has been stressed by many authors (Pearn, 1979; Lippman-Hand & Fraser, 1979b; Lubs, 1979; Antley, 1979; Beeson & Golbus, 1985; Wertz et al, 1986; Sorenson et al, 1987; Shiloh and Saxe, 1989; Frets et al., 1990a). Couples who interpreted their risk as high were less likely to plan a pregnancy than couples who interpreted their risk as low.

No significant relationship was found between reproductive planning and the discrepancy of genetic risk and its interpretation, i.e. when the genetic risk is over 15% and the risk is interpreted as low (Frets et al., 1990a).

The interpretation of the risk is a paramount factor in reproductive planning and is more important than the genetic risk, as will be shown later in the multivariate studies.

The burden of the disorder

The burden of a genetic disorder represents the psychological, social, and financial problems associated with such a disorder (Emery, 1984). The consultands perception of the burden of the disorder may differ from the medical or "objective" burden.

The major importance of the perceived burden of the disorder for reproductive planning has frequently been shown (Sorenson et al., 1987; Bocsknov, 1979; Lippman-Hand & Fraser, 1979b; Emery et al 1979; Somer et al., 1988; Frets et al., 1990a). The burden of the presence of a child with Down's syndrome was one of the reasons not to have more children (Black, 1979; Evers-Kiebooms et al., 1980 & 1984; Springer & Steele, 1980).

There is an ongoing discussion concerning which type of disorder is perceived to be more burdensome to consultands than others. In contrast to Ekwo et al. (1987), others observed that a very early death is seen as less of a burden than a lengthy and progressively downhill illness. Couples at risk of losing an affected child in the perinatal period were more likely to become pregnant again than those at risk of having a child with a progressively downhill illness (Bocsknov, 1979; Lippman-Hand & Fraser, 1979b; Emery et al 1979; Somer et al., 1988; Frets et al., 1990a).

In couples having a risk for a lethal disorder in the perinatal period, there seems to be an urge to plan a subsequent pregnancy. This might be an attempt to "replace" their lost child. In addition, there may be a desire to reconfirm their ability to have a healthy child or to assuage the "injured" feeling evoked by the birth of an affected child (Kessler, 1979a).

Phenylketonuria (PKU) is a special kind of genetic disorder because of the possibilities of newborn screening and dietary therapy. Schild (1979) reported that since these became available, PKU tends to be viewed as a disorder of high risk and low burden. Before the option of prenatal diagnosis, PKU was not a reason to reduce the planned number of children (Burns et al., 1984). However, in the future some parents will consider prenatal diagnosis by chorionic villus sampling. Couples have to weigh this option against lifelong dietary treatment, also when such a diet might not be easily available.

These literature data clearly show that the perceived burden of the disorder is an important factor in reproductive planning. More research is needed to establish which type of disorder is perceived as more burdensome than others. However, treatability of the disorders has not been considered as a separate factor in most studies. For the majority of the disorders for which genetic counselling is requested no effective treatment is available.

When an affected child is born, the parents will experience the full burden of the disorder in their child. The consistency about the significance of the burden of the disorder for reproductive planning does not exist for the presence or absence of an already born affected child and its impact on reproductive planning (Bocsknov, 1979; Wyss-Hutin, 1979; Lippman-Hand & Fraser, 1979b: Sorenson et al., 1987; Frets et al., 1990a).

In the course of time the perceived burden of an affected child with a certain disorder will hardly change, unless possibilities of treatment will increase. When parents already have a child with a disorder which they perceive to be highly burdensome, they might plan a subsequent pregnancy because prenatal diagnosis has become available. Therefore, the impact of the perceived burden of the disorder on subsequent reproductive planning may become less important. If so, the perceived burden can no longer be inferred from reproductive planning after the birth of an affected child and has to be assessed as a separate factor.

Availability of prenatal diagnosis

There is substantial evidence that the availability of prenatal diagnosis is especially valuable for couples with a genetic risk of 10% or more. Table III shows that after prenatal diagnosis became available, more than 90% of the couples at risk for thalassemia in a future child used prenatal diagnosis (Modell & Mouzouras, 1982; Cao et al., 1987).

TABLE III. Proportion of couples undergoing prenatal diagnosis in relation portion to the disorder in their child and the risk of recurrence

Recurrence risk for:	Genetic risk %	Utilization %	Country	Authors
Down's syndrome	1	75	BELGIUM	Evers-Kiebooms (1984)*
Down's syndrome	1	71	USA	Black (1979)*
Down's syndrome	1	22	USA	Oeting (1982)*
Spina bifida	2-5	88	WALES	Laurence (1981)
Spina bifida	2-5	74	USA	Adams (1984)
Spina bifida	3-5	70	BELGIUM	Swerts (1987)
Thalassemia	25	96	UK	Modell (1982)
Thalassemia	25	99	SARDINIA	Cao (1987)
Duchenne Muscular dystrophy	50	83	USA	Beeson (1985)
Haemophilia	50** for a boy	5	SCOTLAND/ CANADA	Markova (1984)
Haemophilia	50** for a boy	14	USA	Barrow (1982)
Haemophilia	50** for a boy	93	USA	Beeson (1985)

* all mothers were < 36 years at the birth of their child with Down's syndrome
** if mother is a proven carrier; otherwise individual risk assessment

The actual availability of prenatal diagnosis induced change in reproductive planning in one-third of the parents of a CF-child (Evers-Kiebooms et al., 1988) and in nearly one-fifth of the parents at risk for haemophilia (A or B) or Duchenne muscular dystrophy (Lubs, 1979). At that time sex-determination was the only available option for the latter two disorders.

In the studies of various disorders, prenatal diagnosis became significant for couples with a genetic risk of 10% or more (Emery et al, 1979; Frets et al., 1990a). As shown in table I, it is clearly corroborated in our own study (Frets et al., 1990a) by the significant difference in the proportion of couples having a genetic risk of 10% or more choosing to have children in those eligible and not eligible for prenatal diagnosis.

Although the effect of the use of prenatal diagnosis was more pronounced in couples having a genetic risk of 10% or more, in nearly all studies at least two-thirds of the couples with a genetic risk of 5% or less underwent prenatal diagnosis when the disorder could be detected prenatally (Table III). Laurence and Morris (1981) found that reproductive planning did not change after the introduction of amniocentesis in the whole sample of parents of spina bifida children. However, reproductive planning only increased in those who had a genetic risk of more than 10% or in those who had a surviving spina bifida child (Laurence & Morris, 1981).

Women at risk for a chromosome abnormality because of advanced maternal age are excluded here because this population is different from couples requesting genetic counselling because of a disorder in the family.

The majority of the couples in a small study in the USA at risk for Duchenne muscular dystrophy in their sons used prenatal diagnosis (Beeson & Golbus, 1985). Despite their high genetic risk, the use of prenatal diagnosis in parents at risk for haemophilia in their sons was inconsistent (Barrow et al., 1982; Markova et al., 1984; Beeson & Golbus, 1985). This might be due to the varying degree of acceptance of prenatal sex-determination as the only option at that time. When foetoscopy became available to identify haemophilia in a male foetus, the acceptance of prenatal diagnosis still varied (Hoyer et al., 1985; Miller et al., 1987). Due to recent development of DNA-analysis an affected male foetus can be identified in the majority of the families at risk for haemophilia and Duchenne muscular dystrophy. The impact of this development on reproductive planning will be an interesting subject for future studies.

The desire to have children

The strength of the desire to have children can be inferred from the number of children the couples had during counselling. Couples who had no children during counselling were more likely to plan a pregnancy than those who already had children (Bocsknov, 1979; Frets

et al., 1990a).

The desire to have children is also reflected in the birth order of the affected child. Table IV shows that couples were more likely to plan a subsequent pregnancy when the affected child was the firstborn, even when they were at risk for an autosomal recessive disorder like cystic fibrosis or phenylketonuria, for which prenatal diagnosis was available for a minority of the families at that time (Steele et al., 1986; Evers-Kiebooms et al, 1984; Burns et al., 1984).

In some studies the desire to have children was explicitly stated as a reason for the reproductive decision (Emery et al., 1979; Springer & Steele, 1980; Frets et al., 1990b). Sorenson (1981b) claimed that the major impact of genetic counselling on reproductive planning was the reinforcement of consultands' reproductive plans made prior to counselling.

These univariate studies show that the desire to have children is strongly related to reproductive planning after genetic counselling. Even in the face of a genetic risk of more than 10%, the desire to have children can prevail.

TABLE IV. Proportion of couples opting for a subsequent pregnancy in relation to the birth order of the affected child

Disorder	Birth order of affected child		Genetic risk %	Authors	
	1st %	> 2nd %			
Cystic Fibrosis	56	12	25	Steele	(1986)
PKU	55	22	25	Burns	(1984)
Down's syndrome/ Spina Bifida	39	8	1-6	Steele	(1986)
Down's syndrome	81	42	1	Evers-Kiebooms	(1984)

Influence of parental age

Except for the study of Sorenson et al., (1987), parental age was repeatedly found to be significantly related to reproductive planning (Bocsknov, 1979; Evers-Kiebooms et al., 1984; Burns et al., 1984; Mazurczak et al., 1985; Steele et al., 1986; Frets et al., 1990a). Women under 30 years of age were more likely to plan a pregnancy than older women.

Diverse factors assessed in multivariate studies

The assessment of a number of factors jointly reveals the relative importance of these factors for reproductive planning. The following factors were found to be most significant. The desire to have children during counselling (Sorenson et al., 1987; Frets et al., 1990b; Sissine et al., 1981).

Furthermore the interpretation of the risk as high or low was another significant factor (Lippman-Hand & Fraser, 1979c; Sorenson et al., 1987; Frets et al., 1990b). In contrast to Sissine et al. (1981) others found that the burden of the disorder was a paramount factor in reproductive planning (Sorenson et al., 1987). Lippman-Hand and Fraser (1979c) emphasized the influence of the consequences of the diagnosis and implications of the prognosis. The significance for the individual couple of the availability of prenatal diagnosis was a paramount factor for reproductive planning (Lippman-Hand & Fraser, 1979c; Frets et al., 1990b). The importance of past reproductive experiences for reproductive planning was also stressed (Lippman-Hand & Fraser, 1979c; Sissine et al., 1981; Frets et al., 1990b).

The factors indicated by the multivariate studies as important do not imply that these factors solely determine reproductive planning. The individual motives for reproductive planning can probably only be derived from a comprehensive personal in-depth interview with both spouses. Such studies are expensive, but the interviews in the study of Lippman-Hand and Fraser (1979a; 1979b) are highly informative on the psychodynamics and emotional contents of the counselling and the post-counselling period. Furthermore, comprehensive reviews on this subject became available and are partly reviewed in this paper (Epstein, 1979; Kessler, 1979b; Emery & Pullen, 1984; Evers-Kiebooms, 1987a).

It appears that the desire to have children, the perception of the burden of the disorder, the interpretation of the genetic risk and the significance of prenatal diagnosis for the individual couple are paramount in reproductive planning. These factors have also been identified in the univariate studies. The multivariate studies have elucidated the significant value of the joint influence of these factors on reproductive planning. However, the magnitude of the genetic risk as supplied in genetic counselling is of relative importance.

THE BURDEN OF THE DECISION-MAKING PROCESS AND REPRODUCTIVE UNCERTAINTY

The burden of the decision-making process needs to be distinguished from the burden of the disorder. The burden of the decision-making process is high when consultands experience particular difficulty in reaching a decision. However, very limited data is available on factors influencing that burden.

The absence of a healthy child, the inability to shirk the responsibility for the decision to

others, and the fear of not being able to cope with an affected child complicated the decision-making process (Lippman-Hand and Fraser, 1979b). Others found that the availability of prenatal diagnosis facilitated the decision-making process in couples at risk for having a child with cystic fibrosis or a neural tube defect (Evers-Kiebooms et al., 1988; Laurence & Morris, 1981).

For some couples the decision-making process can be so difficult that they remain undecided. Reproductive uncertainty means that a couple has not made a childbearing decision after genetic counselling at the time of the follow-up. Table V shows that the proportion of couples who remain undecided after genetic counselling is rather similar, after correction for variations in the follow-up intervals. However, Emery et al. (1979) did not find any couple to be undecided at 2 years' follow-up. This may be because at that stage consultands had been assessed 3 times which might have provided additional support in the decision-making process.

Couples who were likely to remain undecided were those who were uncertain about their reproductive plans before genetic counselling (Wertz et al., 1984), who perceived their risk and burden of the disorder as high (Abramovsky et al., 1980; Wertz et al., 1984) and those who already had an affected child living at home (Sorenson, 1981b; Wertz et al., 1984). These preliminary data suggest at least that these couples might benefit from additional counselling.

TABLE V. **The proportion of couples at risk for various disorders in their offspring who were undecided after genetic counselling**

Time between counselling and follow-up	Undecided %	Authors	
2 years	0	Emery	(1979)
5 months - 15 years	19	Lippman-Hand	(1979b)
about 3 - 8 years	9	Abramovsky	(1980)
6 months	30	Wertz	(1984)
7 - 10 days	24	Sorenson	(1986)
2 - 3 years	11	Frets	(1990a)

CONCLUDING REMARKS AND RECOMMENDATIONS

Before 1979 the genetic risk and the burden of the disorder emerged as the predominant factors for reproductive planning after genetic counselling (Shaw, 1977; Childs, 1978; Evers-Kiebooms & van den Berghe, 1979). In the last decade the follow-up studies after genetic counselling applying multivariate analyses gave probably a more realistic perspective on the complexity of the decision-making process. The burden of the disorder remained paramount. The magnitude of the genetic risk became less important as a single factor. Instead, the interpretation of the risk as high or low and the desire to have children turned out to be one of the most important factors. This change may be partly caused by an in-depth analysis of relevant factors in recent years, and partly by the effects of non-directive counselling (Emery, 1984).

A recent survey revealed that nearly all genetic counsellors preferred the non-directive approach to genetic counselling (Wertz and Fletcher, 1988). This approach is a logical consequence of the principle that consultands must be assisted in reaching an informed and autonomous decision which is appropriate to their life situation. Non-directiveness for counsellors might not always be easy to pursue. A counsellor has to deal with a personal conflict of prevention of genetic disorders on the one hand and respect for consultand's decisions on the other. When the extent of this personal conflict is not wholly in the counsellors consciousness it can be a hazard to a non-directive attitude (Kessler, 1979a).

It is of major concern to identify those couples who are at risk for a long and burdensome decision-making process after genetic counselling. Perceptiveness of counsellors for this problem is an ability to be developed. Supportive counselling by a social worker or clinical psychologist attached to clinical genetics centers can be essential in helping consultands with problems in accepting their situation or in their decision-making process or both.

In the future this psychosocial discipline will become increasingly important when presymptomatic testing will become available for an increasing number of late onset genetic disorders. The decision to take the presymptomatic test can be very difficult because the results can have great psychological and social impact on the candidates, their partners and relatives (Evers-Kiebooms, 1987b; Kessler et al., 1987a; Markel et al., 1987; Mastromauro et al., 1987; Meissen & Berchek, 1987; Meissen et al., 1988; Lamport, 1987).

Presymptomatic testing will also introduce an extra burdensome factor for the reproductive decision-making process. Experience in Huntington's chorea shows that for the test candidates the result would have the greatest impact on family planning, which might urge people to obtain information about their risk status that they otherwise would not have sought (Bloch et al., 1989). The influence of the availability of presymptomatic testing needs

specific emphasis in future follow-up studies after genetic counselling.

Development of new technology (DNA-analysis) could have tremendous social consequences. Focusing on the individual couple is therefore of utmost concern because couples in seemingly similar circumstances may make different decisions or may experience the decision-making process as more or less burdensome than others. It is therefore paramount to safeguard the freedom of couples in that they can make their own decision which they perceive as appropriate to their life situation (Galjaard, 1989).

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CHAPTER III.
FACTORS INFLUENCING THE REPRODUCTIVE DECISION AFTER GENETIC COUNSELING

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ABSTRACT

Here we report a follow-up study involving interviews with 164 couples 2-3 years after genetic counseling to assess the influence of various factors on their reproductive planning. The results show that the desire to have children and the absence of personal experience with the disorder (no close relative being affected) are important single factors for the decision to opt for having children after genetic counseling.

The magnitude of the genetic risk is of relative importance in reproductive planning. Seventy per cent of the couples with a high genetic risk (>15%) opted for having children. When the disorder was perceived as severe and the risk was interpreted as high, 72% opted for having children. The availability of prenatal diagnosis became important only in combination with a high genetic risk (>15%). Forty-seven per cent of the couples with a high genetic risk opted for having children when prenatal diagnosis was not available. In the absence of prenatal diagnosis, couples who had an affected child were more cautious about trying again than those who did not - 50% versus 14% decided to abstain.

This study has provided some insight into the complexity of reproductive decision-making after genetic counseling. The findings may help genetic counselors and clinical geneticists to understand and support counselees in their decision-making process, which is "multi-factorial".

KEY WORDS: genetic counseling, reproductive decision-making, family planning, follow-up, prenatal diagnosis

INTRODUCTION

The aim of genetic counseling is to inform couples about the causal nature of a mental or physical handicap in the family and its occurrence or recurrence risk. The genetic counselor lists various reproductive options such as refraining from having children, prenatal diagnosis, selective abortion and fertilization with another person's gametes. Genetic counseling should facilitate informed reproductive decision-making, allowing for personal and social considerations (Emery, 1984).

The literature does not present a coherent picture of the factors influencing the reproductive decision. Many authors emphasize the combination of the burden of the disorder and the factual genetic risk (Carter et al., 1971; Leonard et al., 1972; Lubs and Falke, 1977; Emery et al., 1979; Bochknov, 1979; Abramovsky et al., 1980).
Others stress the importance of the interpretation of the genetic risk for the reproductive decision (Pearn, 1973; Lippman-Hand and Fraser, 1979; Lubs, 1979; Ekwo et al., 1985; Wertz et al., 1986). The availability of prenatal diagnosis is also mentioned as an important factor (Fraser, 1974; Emery et al., 1979; Lubs, 1979; Evers-Kiebooms et al., 1984; Sorenson et al., 1987). Others found that prenatal diagnosis was only of limited benefit for couples with an affected child (Black, 1979; Oeting and Steele, 1982). In view of cultural, social and economic differences between countries, there is a need for separate national studies in this field (Somer et al., 1988).

An extensive network of facilities for early diagnosis and prevention of congenital disorders has been operational in the Netherlands for a decade. However, no psychological assessment of the results of genetic counseling has been reported in this country until now.

In a follow-up study of genetic counseling we assessed the influence of relevant factors on the reproductive decision as listed in the literature. The factors were assessed separately and in combination.

POPULATION AND PROCEDURE

Study population

In 1984 500 couples sought genetic counseling at the Department of Clinical Genetics of the University Hospital in Rotterdam, the Netherlands. Genetic counseling entailed

diagnostic work-up, analysis of family history, discussion of the risk of occurrence or recurrence, and various options to reduce risks. Each couple had two consultations with the same clinical geneticist, who had subsequently supplied the family with a comprehensive written summary of the information presented and discussed during counseling. A follow-up study was carried out in 164 couples, 2-3 years later, to establish the counselees' subsequent reproductive decision. This study focussed on couples who had come to a decision either to opt for or against having children.

Criteria for enrollment were (a) genetic counseling should apply to own offspring, (b) there should be no history of genetic counseling in another genetic center, and (c) couples should have sufficient command of Dutch, to exclude misconceptions due to language difficulties on the part of e.g. ethnic minorities. Of 190 couples eligible for the study, 16 refused to participate because they were not interested (n=8) or because the subject was too emotional (n=8). Five couples could not be traced. Three couples were excluded because the husband would not participate. Two couples failed to understand the questions and were therefore excluded.

Thus 164 couples were enrolled in the study. Table I shows the reasons for referral to genetic counseling.

TABLE I. Reasons for referrals to genetic counseling

Reasons	Number of Couples	Percentage of Total
Child affected	53	32
Relative affected	50	31
Parent affected	9	5
Combination of these reasons	52	32
Total	164	100

The age of the counselees ranged from 24-59 years for the husbands, with an average of 33.5, and from 24-44 years for the wives, with an average of 30.5. The educational level of the husbands was significantly above that of the general male population[1] (Chi-square 73.26, df 6, $p < .05$). Twenty-eight per cent of the husbands had college experience compared with 18% of the general male population. Of the wives,

[1] Dutch Office for Statistics, Voorburg, the Netherlands

14% had college experience, which did not differ from that of the general female population[1]. Of the rest, 27% per cent of the wives went to high school compared with 11% in the general female population (Chi-square 68.53, df 6, $p < .05$). Religious affiliations were representative of the general population[1].

For the purpose of the study 3 levels of genetic risk were distinguished. Category I involved a genetic risk of less than 5%, most often implying multifactorial determination (classified as "low" genetic risk). Category II involved a genetic risk of 5 to 15% presumably involving multifactorial determination, or cases in which this risk-estimate included various possible modes of inheritance (classified as "moderate" genetic risk). Category III involved a genetic risk higher than 15%, involving usually a monogenic type of inheritance. Couples from the "lowest" risk category (<5%) were randomly selected to achieve equal representation of the three risk categories because the "lowest" risk category was overrepresented in the whole population. The trichotomy of risk level has been used because in our clinical experience these categories fit in with the risk division often made by counselees themselves.

Procedure

Between 1986 and 1987, the couples under study were interviewed at home by a psychologist or one of 3 senior medical students, trained for this purpose. An interval of two years between counseling and follow-up study was considered necessary to allow time for counselees to digest the information and make up their minds.

Participation of both husband and wife was required, to estimate possible interpersonal differences concerning interpretation of the results of genetic counseling and subsequent reproduction. The spouses were interviewed together. In case of disagreement between the spouses the answer which could evoke worries concerning a future child was selected for instance perceiving the disorder as severe. When one of the spouses had expressed this feeling the couple had to deal with this in their decision-making process. Thus the couple was considered perceiving the disorder as severe when one of the spouses perceived the disorder as severe. In this paper interpersonal differences were also at stake concerning risk interpretation and personal experience with the disorder. The parental age was equal or under 30 years when the wife was equal or under 30 years.

A specifically-designed questionnaire was used which had been tested in a pilot study involving 8 couples. The questionnaire listed 91 items inquiring into counselees' socioeconomic position, their reproductive history, and their understanding of the medical and genetic issues. Counselees were asked about their level of genetic risk as presented to them during counseling, and about their own interpretation of this risk. They were also asked about their perception of the severity of the disorder(s) at the time of the

interview and whether they had subsequently come to a reproductive decision. Couples who were expecting a child at the time of their first consultation were asked about their reproductive plans after completion of that particular pregnancy. Counselees were also asked whether the clinical geneticist had answered their questions and concerns.

In this paper reproductive planning was examined in terms of single factors and in terms of combinations of these factors.

Table II lists the type and numbers of disorders which were the indication for genetic counseling in this study. The disorders were classified after Ekwo et al. (1987) into categories variable for early or late death, physical, facial or skeletal disorder, presence or absence of mental retardation. The total number of disorders exceeded 164, because some couples were at risk for more than one disorder. The classification "other" entailed for instance risks from consanguinity or behavioral disturbances. For reasons of clarity the influence of the disorder on the reproductive decision was assessed only in couples at risk for *one* disorder.

TABLE II. Type and number of disorders involved in genetic counseling and the follow-up study

Type of Disorder	Total Number of Couples	Number of Couples That had Decided
- Mentally normal, facial abnormalities	22	19
- Mentally normal, problems with legs and other limb defects	15	14
- Mental retardation, will live into adulthood	40	48
- Early death at 2 - 6 months of age	44	40
- Chronic physically incapacitating illness	74	65
- Other	18	15

Classification after Ekwo et al. (1987).

Statistical Analysis

To identify differences the relative risk (RR) was estimated by the odds ratio with the levels of significance (p values) being two-tailed. In case of trichotomies such as the genetic risk level the Chi-square test for trend was applied (Breslow and Day, 1980). If cell entries in the table equalled zero, 0.5 was added to each cell to estimate the odds ratio (Woolf, 1955).

RESULTS

Of 164 couples 115 (70%) opted for having children. Eighty-seven of those couples (53%) actually became pregnant. Twenty-eight couples (17%) decided not to have any (more) children. Two couples (1%) opted for artificial insemination by donor and one (1%) couple waited for prenatal diagnosis becoming available in the near future. These three couples were excluded from the statistical analysis because their decision differed from the other options and the numbers were too small to treat them as separate groups.

Eleven per cent (18 couples) of the total study population were still undecided at the time of the follow-up. This group will be discussed elsewhere in the context of problems in reproductive decision-making after genetic counseling (Frets et al., 1990a).

Single Factors Associated With the Reproductive Decision

The significant associations between reproductive planning and the single factors are given in Table III. Exclusion concerned couples that had *both* a child and a relative affected by a disorder, because we were interested in the separate influence of an affected child and of an affected relative. One couple was excluded that failed to complete the question about the interpretation of the risk.

The association was most marked between reproductive planning and familiarity with the disorder, followed by the number of any living children during genetic counseling and when a child or a relative was affected (relative risk 11.52, 8.30 and 8.07, respectively). All couples who were not personally acquainted with the disorder, but asked for genetic counseling because of a distant relative, opted for having children. Most of these couples had a genetic risk under 5%. Ninety-four per cent of the couples who had no living children at the time of genetic counseling opted for having children. Most of the couples with an affected relative opted for having children (96%) compared with nearly three-quarter of the couples with an affected child (73%). Two-third of the couples (65%) with an affected relative (close or distant) belonged to the lowest risk category (<5%). Most of the couples with a low genetic risk level as well as those interpreting their risk as low, opted for having children (91% and 89%, respectively). In

comparison, two-third of the couples with a high genetic risk level as well as those interpreting their risk as high, decided in favour of having children (70% and 67%, respectively).

TABLE III. Single factors significantly associated with the reproductive decision after genetic counseling

	Opted for having Children?			RR	Chi-square	
	Yes %	No %	Total N		homo-geneity	trend
Personal experience with disorder?						
Yes	77	23	124	1.0		
No	100	0	19	11.52	4.41*	
Any living children during counseling						
Yes	64	36	64	1.0		
No	94	6	79	8.30	19.55***	
Child affected	73	27	41	1.0		
Relative affected	96	4	46	8.07	8.52**	
Genetic risk < 5%	91	9	57	1.0		
5-15%	76	24	46	.31		
> 15%	70	30	40	.23	7.48**	6.41*
Risk interpreted as low?						
Yes	89	11	85	1.0		
No	67	33	57	.23	11.07***	
Parental age > 30?						
Yes	73	27	74	1.0		
No	88	12	69	2.82	5.37*	

*** $p < .001$ ** $p < .01$ * $p \leq .05$ RR = relative risk

In the assessment of single factors, reproductive planning appeared not to be influenced by educational level, religious affiliation, the availability of prenatal diagnosis, the perceived severity of the disorder or whether the disorder was accompanied by mental retardation ($p > .05$). Nearly two-third (62%) of the couples that

opted for having children perceived the disorder that concerned them as severe. The association between the type of disorder of couples at risk for *one* disorder and the reproductive decision is shown in Table IV. Nineteen couples who were at risk for a child that would die young opted for having children. Eleven of these 19 couples had a genetic risk under 5%. Most of the couples at risk for a chronic physically incapacitating illness in a future child belonged to the highest risk category (>15%) and opted for having children.

TABLE IV. Relationship between the type of disorder of couples at risk for one disorder and the reproductive decision

Type of Disorder	Reproductive Decision		
	Opted for Having Children	Refrained from Having Children	Total
- Mentally normal, facial abnormalities	7	3	10
- Mentally normal, problems with legs and other limb defects	5	1	6
- Mental retardation, will live into adulthood	13	4	17
- Early death at 2 - 6 months of age	19	0	19*
- Chronic physically incapacitating illness	22	8	30*
- Other	5	1	6
Total	71	17	88

* Significant difference between early death and chronic physically incapacitating illness ($p < .05$), the other differences were not significant (Chi-square).

Multiple Factors Associated With the Reproductive Decision

Combinations of two single factors significantly related to reproductive planning are shown in Table V. The strongest association was found between the reproductive decision and the combination of a low genetic risk (<5%) and the number of children during counseling (relative risk 20.51). All couples with a low genetic risk and no children during genetic counseling, opted for having children. Of the couples who already had one or more children during genetic counseling and who had a genetic risk

less than 5%, 22% refrained from having any more. The proportion of these couples with an affected or healthy child avoiding further reproduction was nearly the same (25% and 18%, respectively). Of the couples who interpreted their risk as high and the disease as severe, 72% opted for having children. This percentage decreased to 47% when a high genetic risk (>15%) was combined with the unavailability of prenatal diagnosis (see Table V). Thus, the availability of prenatal diagnosis became important primarily in combination with a high genetic risk.

TABLE V. Combinations of factors significantly associated with the reproductive decision after genetic counseling

	Opted for Having Children?			RR	Chi-square	
	Yes %	No %	Total N		homo-geneity	trend
Risk < 5% and any living children during counseling?						
Yes	78	22	23	1.0		
No	100	0	34	20.51	7.07*	
Disorder perceived as severe and risk interpreted as low?						
Yes	94	6	48	1.0		
No	72	28	36	.17	7.20*	
No prenatal diagnosis available and genetic risk < 5%	94	6	36	1.0		
5-15%	80	20	25	.24		
> 15%	47	53	17	.05	15.70*	13.53*
No prenatal diagnosis available and child affected ?						
Yes	50	50	14	1.0		
No	86	14	64	6.11	8.98*	

* $p < .01$ RR = relative risk

There was no significant difference in the reproductive decision when a low genetic risk was interpreted as high compared with a high genetic risk (>15%) as low. Altogether 10 couples (18%) with a low genetic risk (<5%) interpreted this risk as high and 8 couples of those opted for having children. Sixteen couples (35%) with a moderate genetic risk (5 - 15%) interpreted this risk as high and 9 couples of those opted for having children. Of the couples with a high genetic risk (>15%) 9 couples (23%) interpreted this risk as low and 7 of those opted for having children.

Outcome of pregnancies after genetic counseling

The outcome of the ongoing pregnancy at the first consultation was not significantly related to the subsequent reproductive decision (p>.05) (Table VI).

TABLE VI. Relationship between outcome of pregnancy present at first consultation and subsequent reproductive decision (for future children).

	The Subsequent Reproductive Decision				
Outcome Pregnancy	Opted for having children	Opted against having children	Total	RR	Chi-square homogeneity
Healthy child	14	7	21	1.00	
Affected child	4	2	6	1.00	0.00
	18	9	27		

p > .05

In 6 of the 27 couples the expected child was affected. Four of these 6 couples decided to have at least one more child of whom one couple was eligible for prenatal diagnosis. The two other couples refraining from having children had no possibility of prenatal diagnosis.

Table VII shows the outcome of the pregnancies undertaken after the reproductive decision had been made. The majority of the children were healthy. In case of an affected child, a distinction was made to indicate whether this concerned the disorder for

which the couple was at risk.

TABLE VII. Outcome of pregnancies undertaken after the reproductive decision had been made

Genetic Risk Category	Healthy	Disorder Relevant to Particular Family	Other Disorders
I. < 5%	36	-	6
II. 5 - 15%	24	1	1
III. > 15%	16	2	1
Total	76	3	8

DISCUSSION

The diversity of the disorders under study reflects the case-load of a genetic counseling center. Limiting this investigation to one type of disorder might have simplified this study. This might have resulted in an unwanted limitation of the factors that might influence the reproductive decision. Regarding single factors associated with the reproductive decision, the familiarity with the disorder appeared to be most important. In the absence of personal acquaintance with the disorder of a distant relative, all couples opted for having children. However, this absence of personal experience mainly concerned couples with a low genetic risk. Even so, these results still reflect a tendency to deny the possibility of severe symptoms of the familial disorder in a future child (Falek, 1977; 1984; Rosenstock, 1979).

The second strongest relationship was found between the reproductive decision and the absence or presence of children during genetic counseling, reflecting the desire to have children. The importance of this factor in making reproductive decisions has been stressed in other studies (Pearn, 1973; Reynolds et al., 1974; Emery et al., 1979; Black, 1979; Wertz et al., 1986; Sorenson et al., 1987).

Results of other studies regarding the relationship between genetic risk and reproductive decisions are summarized in Table VIII.

TABLE VIII. Percentages of counselees that decided to have children after genetic counseling (Literature data and the present study)

Study	Country	Total Number of Counselees (N undecided)[1]	Genetic Risk % < 10%	≥10%
Carter (1970)	UK	421 (0)	76	36
Emery (1979)	UK	200 (0)	82	47
Bochknov (1979)	USSR	225 (0)	86	43
Abramovsky (1980)	USA	212[2](15)	70	50
Sorenson (1987)	USA	181 (0)	59	42
Present study*	NETH			
- Entire population		164[3](18)	86	71
- Population not eligible for prenatal diagnosis		90 (12)	89	52

* to make the results of the studies comparable, the genetic risk was split at 10%.

1 The numbers of couples who were still undecided 6 months or more after genetic counseling are given in parentheses. These couples were also included in the total number of couples.

2 No numerical risk could be offered (n = 31)

3 Three couples were excluded (see results on page 8)

Most other workers (Carter et al., 1971; Emery et al., 1979; Abramovsky et al., 1980; Sorenson et al., 1987) reported that 36-50% of the couples with a high genetic risk (≥10%) opted for having children. In contrast, in our study this amounted to 71%. However, except for the Sorenson study the results of our study were not significantly different from the results of the other studies. The significant difference between the results of the study of Sorenson et al. (1987) and our study ($p = .05$) might be due to the different time of follow-up (6 months and 2 to 3 years after genetic counseling). Compared with the other studies part of the different trend in our study might be explained by the availability of prenatal diagnosis for 44% (71/161) of the couples. Only 52% of the couples with a high genetic risk who were not eligible for prenatal monitoring opted for having children, showing the significance of prenatal diagnosis in combination with a high genetic risk. The data do not support the notion that the magnitude of the genetic risk, as a single factor, is paramount in reproductive planning. In agreement with Kessler and Levine (1987), we found that genetic risk was of relative importance.

In regards to the combination of factors associated with the reproductive decision 72% of the couples who interpreted their risk as high and perceived the disorder as severe decided in favour of having children (see Table V). An explanation for this apparent disregard for the consequences might reflect an unconscious reaction to the "unbearable" feeling of lowered self-esteem, due to the hereditary nature of the disorder (Leonard et al., 1972; Antley et al., 1973; Antley and Hartlage, 1976; McCollum and Silverberg, 1979; Corgan, 1979; Broder and Trier, 1985).

Prenatal diagnosis only became important in combination with a genetic risk higher than 15% (Modell et al., 1980; Cao et al., 1980; Kaback, 1982; Somer et al., 1988). Moreover, in the absence of prenatal diagnosis, couples who had an affected child were more cautious about trying again than those who did not - 50% vs 14% decided to abstain. Nevertheless, there was still an appreciable number (50%) who were willing to take the chance of having an affected child, even in the absence of prenatal diagnosis (Table V).

Interpreting a low genetic risk ($<5\%$) as high or a high genetic risk ($>15\%$) as low did not influence the reproductive decision ($p>.05$). Due to small numbers no definite conclusions can be drawn. In agreement with others we found that the perception of the severity of the disorder tends to be included in the interpretation of the risk (Pearn, 1973; Cote, 1982; Somer et al., 1988). The couples with a low genetic risk who interpreted their risk as high mainly perceived the disorder as severe. The majority of the couples with a high genetic risk interpreting their risk as low did not perceive the disorder as severe.

Eighteen couples whose expected child at the first consultation was born opted for having children. Four couples whose expected child at the first consultation was affected were not deterred from trying again. Only one of these 4 couples was eligible for prenatal diagnosis. The absence of prenatal diagnosis did not deter these couples from trying again (Table VI). Because of the small numbers we have to be cautious in drawing definite conclusions. The same is true for the outcome of pregnancy after the reproductive decision had been made (Table VII).

The subjective severity of the disorder for the counselees was assessed, rather than the objective burden of a disorder. This was in accordance with the existing literature at the time of the study. A recent investigation deals to a certain extent with the objective burden of a disorder (Ekwo et al., 1987). Our results on the association between the reproductive decision and the type of disorder are limited to couples being at risk for one disorder and call for caution in drawing definite conclusions. In contrast to Ekwo et al. (1987) there is evidence in the literature that a very early death is seen as less of a burden than a lengthy and progressively downhill illness (Carter et al., 1971; Bochknov,

1979; Lippman-Hand and Fraser, 1979; Emery et al., 1979; Somer et al., 1988). Considering this, our data support the notion that the burden of the disorder is an important factor influencing the reproductive decision. Couples being at risk for a defect resulting in early death mainly opted for having children whereas couples at risk for a prolonged illness were more afraid to take the risk. The use of the categories as proposed by Ekwo et al. (1987), although they might be arbitrary due to variability of genetic disorders, could serve the purpose of classification of some of the problems in reproductive planning after genetic counseling.

The primary goal of genetic counseling is to inform and support counselees. Most (78%) of the couples said they had benefited from genetic counseling in that this had enabled them to make an informed decision. From a social point of view the decision not to have children in case of a high genetic risk (>15%) would lead to prevention. Altogether, 70% of the couples with a high genetic risk opted for having children. This shows that taking an informed decision in such a high risk situation is a very complicated process, involving much more than the "rational" assumption that one should avoid further reproduction in face of a high risk.

In a forthcoming study the joint influence of eight relevant factors on the reproductive decision will be assessed, including the possibility of predicting the reproductive decision (Frets et al., 1990b).

CONCLUSION

The genetic facts provided by clinical geneticists seem to be of relative importance for the reproductive decision after genetic counseling. Issues at stake *before* genetic counseling such as the desire to have children and the familiarity with the disorder seem to be more important in reproductive planning. The data do not support the notion that the magnitude of the genetic risk, as a single factor, is paramount in reproductive planning. Couples tend to take high risks, even when the disease is perceived as severe and prenatal diagnosis is not available. This study has provided some insight into the complexity of reproductive decision-making after genetic counseling. The findings may help genetic counselors and clinical geneticists to understand and support counselees in their decision-making process, which is "multi-factorial".

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CHAPTER IV.
MODEL IDENTIFYING THE REPRODUCTIVE DECISION AFTER GENETIC COUNSELING

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ABSTRACT

To assess the identifiability of reproductive planning after genetic counseling, a model was designed to study 8 relevant factors influencing reproductive decisions after genetic counseling. Altogether 164 couples were interviewed at home 2 to 3 years after genetic counseling. The factors were arranged in a flow chart distinguishing 3 groups: reproductive outcome prior to genetic counseling, desire to have children, and interpretation of information gained from genetic counseling. The model based upon these retrospective data showed that reproductive decisions were identified correctly in 91% of the cases. The model consisted of 8 factors and documented the urgency of the desire to have children and the interpretation of the genetic risk. In addition, linear discriminant analysis of the 8 relevant factors enabled identification of the reproductive decision in 96% of the cases. This model may prove helpful to counselors and counselees by showing what other couples have decided in comparable circumstances and for which reasons.

KEY WORDS: genetic counseling, reproductive decision-making, model, family-planning, follow-up, risk interpretation.

INTRODUCTION

In 1975 the Ad Hoc Committee on Genetic Counseling of the American Society of Human Genetics claimed that the aim of genetic counseling was to inform couples about the nature of a mental or physical handicap in the family and its risk of occurrence or recurrence[1]. Furthermore, various options to prevent the birth of an affected child were discussed such as refraining from having children, prenatal diagnosis and selective abortion, or fertilization with donor gametes. Genetic counseling should facilitate informed reproductive decision-making, allowing for personal and social considerations (Emery, 1984).

The literature does not present a coherent picture of the factors influencing reproductive decisions. Many workers stressed the importance of the genetic risk (Carter et al., 1971; Leonard et al., 1972; Emery et al., 1979; Abromovsky et al., 1980). Others emphasized the importance of the interpretation of the genetic risk (Lippman-Hand et al., 1979; Pearn, 1979; Côté, 1982; Ekwo et al., 1985; Wertz et al., 1986; Sorenson et al., 1987; Kessler and Levine, 1987). In a previous study we assessed the influence of various factors on the reproductive decision after genetic counseling. From this study it appeared that the decision was mainly influenced by personal experience with the disorder and the desire to have (more) children (Frets et al., 1990a).

The present study includes assessment of relevant factors that might enable identification of couples with respect to their reproductive decision after genetic counseling.

A model was constructed based on relevant factors influencing the reproductive decision such as the desire to have children (Pearn, 1973; Black, 1979; Côté, 1982; Wertz et al., 1986; Sorenson et al., 1987; Frets et al., 1990a) and the interpretation of the risk (Ekwo et al., 1985; Wertz et al., 1986; Sorenson et al., 1987). This model correctly identified the decision made by the most of the couples under study. The constructed model may prove helpful to counselors and counselees by showing which decision is made by other couples in comparable circumstances and for which reasons.

POPULATION AND PROCEDURE

Study Population

In 1984 500 couples, mostly having complex family histories of mental or physical handicaps, sought genetic counseling at our Department. This entailed full counseling including diagnostic work-up, family history, estimating the risk of occurrence or recurrence, and various

[1] Indentation of certain sentences or entire paragraphs, leaving a large left margin, indicates that these sections cover grounds already dealt with in previous chapters.

options. A follow-up study was carried out in 164 couples, 2 to 3 years later, to establish the counselees' subsequent reproductive decision. A comprehensive description of methods and study population has been reported previously (Frets et al., 1990a).
This study focussed on couples who had arrived at a decision either to opt for or against having children. The couples who were still undecided will be discussed in a subsequent paper (Frets et al., 1990b).

Criteria for enrollment were (a) genetic counseling should apply to risk of occurrence or recurrence in own future offspring, (b) there should be no history of genetic counseling in another genetic center, and (c) couples should have sufficient command of Dutch to exclude misconceptions due to language difficulties.

Three levels of genetic risk were distinguished. Category I involved a genetic risk of less than 5%, category II entailed a genetic risk of 5 to 15% and category III consisted of a genetic risk higher than 15%. The lowest risk category was overrepresented in the whole population. Therefore, couples from this risk category were randomly selected to achieve equal representation of the 3 risk categories.

Procedure

Between 1986 and 1987, the couples under study were interviewed at home by a psychologist or one of 3 senior medical students trained for this purpose. A specifically designed questionnaire was used listing 91 items inquiring into counselees' socioeconomic position and their reproductive history. Counselees were asked about the interpretation of their risk and of the severity of the disorder. They were also asked whether they had subsequently arrived at a reproductive decision. If so, they were asked to give the reasons for their decision. The couples' answers were written down in detail, instead of only focussing on the answers to the multiple choice questions, to get more reliable insight into the issues that were important for the couples.

Table I lists the type and numbers of disorders involved in the genetic counseling and, thus in our study, and reflect the normal case-load of a genetic counseling unit. The disorders were classified after Ekwo et al. (1987) into categories variable for early/late death, physical (facial or skeletal) disorder, presence or absence of mental retardation. The total number of disorders exceeded 164, because some couples were at risk for more than one disorder. The classification "other" entailed i.e. risks from consanguinity, behavioral disturbances in the index patient and congenital malformations as a consequence of diabetes mellitus of the mother.

TABLE I. Type and numbers of disorders involved in genetic counseling and the follow-up study

Type of disorder	Total number of couples	Number of couples that had opted for or against having children
- Mentally normal, facial abnormalities	22	19
- Mentally normal, problems with legs and other limb defects	15	14
- Mental retardation, will live into adulthood	48	40
- Early death at age 2 - 6 months	44	40
- Chronic, physically incapacitating illness	74	65
- Other	18	15

Classification after Ekwo et al. (1987).

Relevant factors influencing the reproductive decision after genetic counseling were suggested by a review of the literature and from responses to the questionnaire in this study. A semi-hierarchical scheme (see Table II) was developed of single or combined factors which were almost exclusively found in couples opting for having children and similarly other factors almost exclusively found in couples refraining from having children. For example, *all* the eight couples who had lost their only child before genetic counseling opted for having children. Only those factors were included which were almost exclusive for a decision and subsequently had predictive value.

The semi-hierarchical sequence indicates that when more than one factor was relevant to or important for a couple, this couple was identified by the first factor in the hierarchy.

TABLE II. Influence of single or combined decisive factors on the reproductive decision

Factors		Reproductive decision		
	Factor relevant/ decisive	Opted for having children	Opted against having children	Total
A1. Only child died before genetic counseling	+	8	0	8
	-	107	28	135
				143
B1. Very strong desire to have children*	+	24	1	25
	-	91	27	118
				143
B2. Family perceived as complete***	+	0	14	14
	-	115	14	129
				143
B3. Genetic risk < 5% and no children before genetic counseling**	+	29	0	29
	-	86	28	114
				143
C1. Risk interpreted as low or moderate***	+	33	0	33
	-	82	28	110
				143
C2. Risk interpreted as high***	+	1	9	10
	-	114	19	133
				143

* $p < .05$ ** $p < .01$ *** $p < .001$ (continued)

TABLE II. (cont.)

Factors		Reproductive decision		
	Factor relevant/ decisive	Opted for having children	Opted against having children	Total
C3. Prenatal diagnosis decisive	+	9	0	9
	-	106	28	134
				143
C4. Disorder perceived as severe + pre-natal diagnosis available + no children before genetic counseling + parental age ≤ 30 years	+	4	0	4
	-	111	28	139
				143

A flow chart, representing these almost exclusive factors, was drawn up to serve as a model to identify the reproductive decision correctly. A correctly identified decision is that decision which is made by *all* couples when a certain factor is relevant. Given a certain factor and all couples opt for having children the decision to opt for having children is the correctly identified decision. When this particular factor is relevant and *one* couple refrains from having children implies that this couple is *wrongly* identified. A couple was considered to have given a certain reason for their decision when one of the spouses had given this reason.

In some cases the association between the almost exclusive single or combined factors and the reproductive decision was not significant. The non-significant factors were nevertheless included because we focussed on the correct identification of the decision enabled by the model as a *whole* rather than on the separate single or combined factors. Only those almost exclusive factors were entered in the *model* which contributed to the

highest number of correctly identified couples. The optimal fit of the model is achieved when the number of correctly identified couples decreases after leaving out one factor of the model.

In addition, Fisher's linear discriminant analysis (Nie et al., 1975) was used, logistic regression analysis not being applicable because the zero-values in the 2x2 table resulted in excessive standard errors. The jackknife method, whereby one factor is left out at a time, served to test the stability of the results (Lachenbruch and Mickey, 1968).

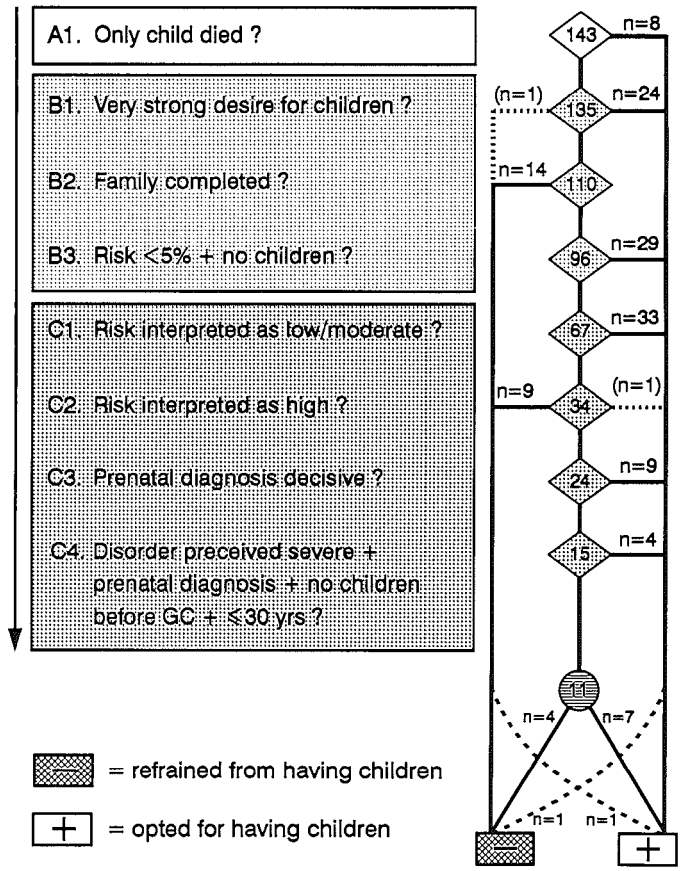
RESULTS

Altogether 115 couples opted for and 28 couples opted against having children, 18 couples were still undecided. Two couples opted for artificial insemination by donor and one couple waited for prenatal diagnosis to become available in the near future. These 3 couples were excluded from the statistical analysis because their decision differed from the other options and the numbers were too small to treat them as separate groups.

Table II shows the influence of single or combined factors on the reproductive decision. The factors were principally genetic in origin (factor A1, B3, C4), or based on the main reasons the couples gave for their decision (factor B1, B2, C1, C2, C3). For example, all 33 couples who interpreted their risk as low or moderate and gave this as a main reason for their decision opted for having children (factor C1). Those who did not give this but another (main) reason for their decision either opted for or against having children.

The joint influence of the various single or combined factors from Table II on the reproductive decision is shown in the flow chart (Fig. 1). Three groups were distinguished with respect to (A) reproductive outcome prior to genetic counseling, (B) the desire to have children, including perception of a "complete" family after genetic counseling, and (C) interpretation or evaluation of information gained from genetic counseling. The sequence of these groups of factors is not essential, but it is a way of presenting the factors in a orderly fashion. Of 143 couples who had made a decision to opt for or against having children, all 8 couples that had lost their only child before genetic counseling opted for having children. Of the other 135 couples, 24 of 25 with a very strong desire to have children decided to do so, giving that desire as their reason for having children ("We could not conceive of a life without children").

FIGURE 1. Flow chart model identifying the reproductive decision



Short horizontal lines imply affirmative answers, with the numbers in the diamonds representing total number of couples after each substraction. Numbers in brackets represent couples wrongly identified with the dotted lines indicating the actual decision.

Of the remaining 110 couples, all 14 couples, who perceived their family as complete after genetic counseling gave this as a main reason for their decision to refrain from having (more) children. Of the other 96 couples, all 29 with a low genetic risk and no children before genetic counseling opted for having children. Of the remaining 67 couples, all 33 couples that interpreted their risk as low or moderate gave this as a reason to opt for having children. Of the other 34 couples, 9 of 10 that interpreted the

risk as high, gave this as a reason to refrain from having children. Of the remaining 24 couples, 9 opted for having children, giving prenatal diagnosis as the decisive (and available) factor for their decision to opt for having children. Of the other 15 couples, all 4 couples who perceived the disorder as severe and eligible for prenatal diagnosis, who had no children prior to genetic counseling and who were not older than 30, opted for having children.

Eleven couples did not fit any correct identification. Seven of them opted for having children, with the other 4 against. Reasons for the decision of the 7 unidentified couples to opt for having children mainly entailed taking the risk despite its magnitude or perceiving the disorder as not severe. The various reasons for the decision of the 4 unidentified couples to refrain from having (more) children consisted of perceiving the disorder as very severe or the care of their affected child as too demanding.

The reason for *wrongly* identifying 2 couples (see factor B1 and C2) mainly concerned disagreement between the spouses.

Regarding the 25 couples that were expecting a child at first consultation, 10 interpreted their risk for a subsequent pregnancy as low (factor C1) and had opted for another pregnancy. Of these couples 6 had a genetic risk less than 5% and 4 a risk between 5 and 15%. Seven couples perceived their family as complete (factor B2) and decided against a subsequent pregnancy. The other 8 couples were spread out over the 3 groups of factors.

The number of correctly identified couples remained approximately the same when the hierarchical sequence of group B and C or the sequence of the decisive factors *within* group B (B1-B3) and C (C1-C4) in Fig. 1 were changed.

The types of disorders for most of the factors in the model represented the whole spectrum of early/late death, presence or absence of mental retardation or chronic illness. Couples whose only child had died before genetic counseling (factor A1) were at risk to lose a next affected child within 2 to 6 months after birth.

Couples that perceived their family as complete mainly had a genetic risk higher than 5%. Those that interpreted their risk as low or moderate mainly had a genetic risk of less than 15%. Most couples interpreting their risk as high or those claiming that prenatal diagnosis was a prerequisite for their decision had a high genetic risk ($>15\%$). In the other factors the genetic risk categories were equally represented.

Table III gives the total number of couples whose reproductive decision was correctly identified, applying the flow chart model.

TABLE III. Correctly identified reproductive behavior applying the model

	Identified decision			
Actual decision	Refrained from having children	Opted for having children	No identification	Total
Refrained from having children	23	1	4	28
Opted for having children	1	107	7	115
	24	108	11	143

	%
Correct identification	91
Decision to have children correctly identified	99
Decision not to have children correctly identified	96
Specificity (107/115)	94
Sensitivity (23/28)	82

The decision to have children made by 115 couples was correctly identified in 107 cases. Of the 28 couples that refrained from having any (more) children 23 were identified correctly. The correct identification of the decision to have children amounted to 93% (107/115) and was based on the factors A1, B1, B3, C1, C3, C4. The correct identification of the decision *not* to have children amounted to 82% (23/28) and was based on the factors B2, C2. The total rate of correct identification was 91% (130/143).

The results were also analyzed from a different perspective using a Fisher's linear discriminant analysis. This analysis yielded a significant discriminant (Chi-square= 223.51, df= 8, $p < .00001$): 96% of the patients could be identified correctly with the 8 factors from the model (114+23/143). Table IV gives the standardized discriminant function coefficients indicating the relative importance of the 8 factors, the higher the coefficient the more important the factor is in differentiating between the decision to opt for against having children. The jackknife-method provided identical results. When the 8 factors were arranged in a flow chart identical to the selection sequence of the linear

discriminant analysis, one additional couple was identified correctly (Table V). The sensitivity in identifying the couples opting against having children of the model and the linear discriminant analysis were the same.

TABLE IV. Results of the linear discriminant analysis of 8 factors correctly identifying the reproductive decision

Factors	Standardized discriminant function coefficients
B2. Family perceived as complete	.554
C2. Risk interpreted as high	.252
C1. Risk interpreted as low or moderate	.249
B3. Genetic risk < 5% and no children during Genetic Counseling	.242
B1. Very strong desire to have children	.238
C3. Prenatal diagnosis decisive	.229
A1. Only child died before genetic counseling	.218
C4. Disorder perceived as severe + prenatal diagnosis available + no children before genetic counseling + parental age ± 30 years	.195
Wilk's Lambda	.20
Chi-squared	223.51
	df = 6, p <.00001

TABLE V. Correctly identifying reproductive behavior applying linear discriminant analysis

	Identified decision		
Actual decision	Refrained from having children	Opted for having children	Total
Refrained from having children	23	5	28
Opted for having children	1	114	115
	24	119	143

	%
Correct identification	91
Decision to have children correctly identified	96
Decision not to have children correctly identified	96
Specificity (114/115)	99
Sensitivity (23/28)	82

DISCUSSION

The Model

The constructed model was successful in identifying the reproductive decision correctly in 91% of the cases. The model may prove useful in clinical practice. When a couple has problems with decision-making after genetic counseling the model can be easily applied by following the flow chart and determining which issues are relevant or important to a particular couple. When the attitude of a couple is not clear the genetic counselor can help to understand why this is the case for that individual couple. Genetic counselors and clinical geneticists may be able to show, when needed, what other couples have decided in comparable circumstances and for which reasons. In this way it provides an answer to a question often asked by counselees (Lippman-Hand and Fraser, 1979).

However, to identify factors facilitating the decision-making process after genetic counseling one may compare couples who were able to make a decision and those who were not. Assessment of these factors will be subject of a subsequent paper.

The number of factors considered proved to be essential, because the number of couples correctly identified decreased as soon as one factor was eliminated from the model.

The arrangement of the factors *within* group B (B1 - B3) or C (C1 - C4) in Fig. 1 and the sequence of group B and C in the model did not influence the results. That is, changing the hierarchical sequence of the factors within group B and C or the sequence of the groups did not alter the correct identification of the model. The groups of factors based on a common denominator were considered clinically more relevant than the statistical sequence of separate single or combined factors based on the linear discriminant analysis. The common denominators in the model were the desire to have children (group B) and the interpretation of information gained from genetic counseling (group C).

The linear discriminant analysis showed that as in the model, all 8 factors contributed significantly. Considering the percentages, the correct identification of the model and the linear discriminant analysis were slightly different (91% and 96%, respectively). These percentages are different because the linear discriminant analysis is more powerful when the factors are independent, which indicates that two or more factors do not identify the *same* couple. Both methods turned out to have similar sensitivities in identifying the couples opting against having children.

Our results from the model and the linear discriminant analysis suggest that counselees' interpretation of the risk, assessed as a main reason for the decision, and the strength of the desire to have children are paramount factors influencing the reproductive decision after genetic counseling. The perception of the couple that their family is complete reflects the absence of the desire to have children. These findings are in agreement with those of our earlier paper and those of other studies (Pearn, 1973; Lippman-Hand and Fraser, 1979; Ekwo et al., 1985; Wertz et al., 1986; Sorenson et al., 1987; Kessler and Levine, 1987).

The model is not causal in that the factors do not explain *why* a couple made a certain decision. The individual motives for their decision cannot be derived from this model. However, the *real* motives are probably related to the factors in the model. Further investigation is required to explore this field. The present study does not pretend to rationalize the decision-making process after genetic counseling. While the construction of a model involves a certain arrangement of the data, the individual motives behind the decision could be irrational, by the standards of the counselor.

Single or combined factors of the model. It may seem strange that 14 couples who perceived their family as complete after genetic counseling had requested genetic counseling in the first place. One of the criteria for enrollment in the study was that counselees were considering future children. Seven of these 14 couples, all with a low genetic risk, were expecting a child at the first consultation. Once the child was born after genetic counseling, these couples decided their family was complete. For the other 7 couples the perception of family completeness may have been caused by rationalization of the unbearable feelings of having to refrain from having (more) children. The fact that these 7 couples had a moderate or high genetic risk (> 5%) might not have been purely accidental. Additional reasons for the decision given by these couples were that they felt their risk was too high or the care of their affected child born prior to genetic counseling was too demanding.

Perception of the severity of the disorder is an implicit ingredient of the model. It concerns the death of an only child (factor A1) and prenatal diagnosis being decisive and available (factor C3). The latter factor was mainly an issue for couples at risk for disorders that will result in chronic morbidity and early death. The perception of the severity of the disorder is explicitly present in the model in the last factor (factor C4). Perception of the severity of the disorder could only correctly identify the decision when it was combined with the availability of prenatal diagnosis, no children before genetic counseling and the parental age below or equal to 30.

The influence of the factors of the model on the reproductive decision cannot be explained by the presence of a certain type of disorder. The types of disorder in 7 of the 8 factors represented the whole spectrum of early/late death, and presence or absence of mental retardation or chronic illness.

The level of genetic risk might have influenced the content of the main reason for the decision. That is, the interpretation of the risk given as a main reason for the decision was presumably affected by the level of genetic risk. Couples interpreting their risk as low or moderate and giving this as a main reason for their decision had a genetic risk less than or equal to 15%. Couples interpreting their risk as high and giving this as a main reason for their decision had a genetic risk higher than 15%. The decision of the couples for whom prenatal diagnosis was a prerequisite for their decision might have been influenced by the (high) genetic risk (>15%). The influence of the availability of prenatal diagnosis on the reproductive decision in case of a high genetic risk was also emphasized by others (Modell and Mouzouras, 1982; Cao et al., 1987). The perception that the family was complete might also have been affected by the level of the genetic risk. Couples who had lost their only child or those having a very strong desire to have children were not influenced by the genetic risk. The genetic risks of these couples were

spread out over the 3 risk categories. The genetic risk is one of the many factors to be weighed by couples, but is not experienced as a decisive factor in this study population.

The marked influence of personal acquaintance with the disorder (i.e. a close relative being affected) on the reproductive decision as found in a previous study was overridden by the joint influence of the 8 factors of the model. It appeared that all couples not being personally acquainted with the disorder opted for having children (Frets et al., 1990a). This particular factor was redundant in the model because after leaving this factor out of the model the number of correctly identified couples remained the same.

Design. The results of this study may have been distorted in several ways by its retrospective design. The reason given by the couple for their decision may have been distorted in an attempt to make their decision fit the bill. Dissonance is an unpleasant state, and the person is motivated to reduce this by either adding new cognitions or changing existing ones (Wrightsman, 1972). Recall bias may be another interfering factor. However, the reproductive decision is to a large extent influenced by feelings and not by a recall of facts.

The substantiated decision analytic model of Pauker and Pauker (1987) could not be applied to our study population because most of their study individuals were at risk for a chromosome abnormality due to advanced maternal age.

Analyzing the data of this study, the model is successful in correctly identifying the reproductive decision. To test the stability of our model the present study will be replicated in a prospective design. If this prospective study leads to similar results the model has proved to be stable.

CONCLUSION

With this study we gained insight into the joint influence of 8 important factors on the reproductive decision after genetic counseling. The model was successful in correctly identifying the reproductive decision in 91% of the cases. Counselees' interpretation of the risk, assessed as a main reason for the decision, and the strength of their desire to have children are paramount factors influencing the reproductive decision after genetic counseling. The marked influence of personal experience with the disorder on the reproductive decision as found in a previous study was overridden by the joint influence of the 8 factors in the model. The model may prove helpful to counselors and counselees by showing what other couples have decided in comparable circumstances and for which reasons.

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CHAPTER V.
ANALYSIS OF PROBLEMS IN MAKING THE REPRODUCTIVE DECISION AFTER GENETIC COUNSELLING

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ABSTRACT

A follow-up study of 164 couples to evaluate reproductive decision-making 2-3 years after genetic counselling, revealed that 43% had problems making the reproductive decision. These couples (a) had experienced the decision-making process as difficult, (b) had doubts about the decision they had made or (c) had been unable to make a decision. Logistic regression analysis revealed that the following factors as independently and significantly associated with problems in the decision-making process: (1) no postcounselling relief; (2) anticipation of a high risk level; (3) relatives' disapproval of decision; (4) the decision not to have a(nother) child; and (5) the presence of an affected child. Interestingly, 45% of the couples eligible for prenatal diagnosis who decided to have children experienced the decision-making process as difficult against 23% of couples deciding to have children while prenatal diagnosis was not available ($p<.05$). Problems in the decision-making process might become apparent after genetic counselling rather than in the course of it. Therefore, we suggest a structured follow-up 3-6 months after genetic counselling to identify couples that would benefit from additional supportive counselling.

KEY WORDS: reproductive decision-making, reproductive uncertainty, family planning, follow-up, genetic counselling, prenatal diagnosis, risk interpretation, risk recall

INTRODUCTION

> In 1975, the Ad Hoc Committee on Genetic Counselling of the American Society of Human Genetics determined that the aim of genetic counselling is to inform consultands about the nature of a mental or physical handicap in the family and its risk of occurrence or recurrence. Furthermore, the Committee advocated discussion of various options to prevent the birth of an affected child, such as refraining from having children, prenatal diagnosis and selective abortion, or fertilization with donor gametes. Genetic counselling should facilitate informed reproductive decision-making, allowing for personal and social considerations (Emery, 1984).

Problems in the postcounselling reproductive decision-making process can be divided into 3 categories (a) experiencing the decision-making process as particularly difficult, (b) unresolved doubt about a decision once taken and (c) inability to make a decision. Factors related to difficulty with the decision-making process after genetic counselling have been assessed only occasionally. The absence of a healthy child, the inability to abdicate the responsibility for the decision with others, and the fear of not being able to cope with an affected child complicated the decision-making process (Lippman-Hand and Fraser, 1979a), while others found that the availability of prenatal diagnosis facilitated the decision-making process (Laurence and Morris, 1981; Evers-Kiebooms et al., 1988). A few studies listed factors associated with persistent reproductive uncertainty after genetic counselling. Lubs (1979) found that the higher the level of genetic risk, the more likely it was that a couple remained undecided. Others reported that it was the perception of the risk and burden of the disorder as high that increased the chance of persisting uncertainty after genetic counselling (Abramovsky et al., 1980; Wertz et al., 1984). Couples more likely to remain undecided were those who had an affected child (Wertz et al., 1984; Sorenson et al., 1981) or those who were uncertain about reproductive plans before genetic counselling (Sorenson et al., 1981.

The study reported here is part of a larger one concerning the decision making-process after genetic counselling. Other aspects included analysis of single factors or a combination factors influencing the reproductive decision (Frets et al., 1990a). A model was developed capable of identifying the reproductive decision correctly in more than 90% of the cases (Frets et al., 1990b). In the present study we investigated which factors were associated with postcounselling (a) difficulty in making the reproductive decision, (b) unresolved doubts after the decision has been made, or (c) persistent reproductive

uncertainty.

Insight into the factors which are related to problems with the decision-making process might indicate which couples would benefit from additional counselling.

POPULATION AND PROCEDURE

Study population

The study population comprised 500 couples seen for genetic counselling at the Department of Clinical Genetics in 1984 most of whom had complex family histories ofmental and/or physical handicaps. This counselling entailed diagnostic work-up, family history taking, estimation of the risk level, and discussion of various options to prevent the birth of an affected child.

Only those couples were entered into the study that had requested genetic counselling for their own offspring (n=421). The following couples were excluded from the study because of (a) a history of genetic counselling at another clinical genetics centre (n=63), (b) uncompleted genetic counselling (n=30), (c) separation of the spouses (n=13), (d) insufficient command of Dutch (n=5), (e) personal circumstances (n=6) and (f) assessment in the pilot study to test the questionnaire or to train the research assistants (n=13).

Three levels of genetic risk were distinguished. Category I involved a genetic risk of less than 5%, category II entailed a genetic risk of 5 to 15% and category III consisted of a genetic risk higher than 15%. The lowest risk category was overrepresented in the whole population. Therefore, couples from this risk category were randomly selected to achieve equal representation of the 3 risk categories, whereby 101 couples were excluded.

Of 190 couples eligible for the study, 16 refused to participate because they were not interested (n=8) or because the subject was too emotional (n=8). Five couples could not be traced and 3 couples were excluded because the husband would not participate. Two couples failed to understand the questions and were therefore excluded. Altogether, 164 couples were enroled in the study. A detailed description of the educational level and religious affiliation of the study population has been reported elsewhere (Frets et al., 1990a).

Procedure

Between 1986 and 1987, the couples under study were interviewed at home by a psychologist or one of three senior medical students, trained for this purpose.

A questionnaire was constructed listing 91 items, partly multiple-choice and partly open-ended. The questionnaire inquired whether the couple had been worried in the months before genetic counselling, whether they expected their risk to be high or low and how they felt shortly after genetic counselling, e.g. relief, worry, disappointment etc. They were also asked about their recollection of the risk level given by the counsellor indicated as the recalled risk. Risk recall was considered correct if this fell in

the correct category, with both spouses giving the correct answer.

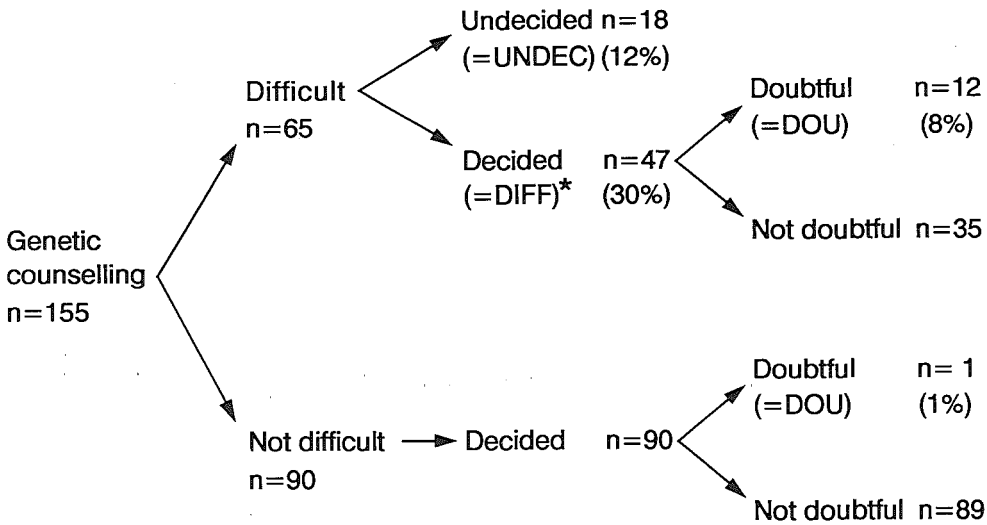
Couples were also asked whether they had subsequently come to a reproductive decision or had remained undecided (=UNDEC). If they had made a decision they were asked whether they had experienced particular difficulty in making this decision or whether it had taken a great deal of deliberation or both (=DIFF). Whether the decision once made by the couples had left unresolved doubts was based on the answers to the following questions: (a) does your decision bother you or (b) have you been wondering recently whether you made the right decision? Unresolved doubt (=DOU) was indicated when either one of these questions was answered in the affirmative.

The questions about difficulty with the decision-making process and subsequent doubtfulness had to be answered with yes or no. Difficulty with the decision-making process or postdecision doubt were considered to be present when the consultand had experienced these feelings more than once following genetic counselling. In 2 cases consultands did not give unequivocal answers regarding unresolved doubts, requiring a senior psychologist's judgement.

Figure 1 is a schematic presentation of the variable outcome of the postcounselling decision-making process in relation to problems experienced in making that decision. DIFF and DOU couples are classed separately because unresolved doubt about such an important decision was considered more serious than difficulty experienced in making a decision that leaves consultands at peace. Factors that contributed to both difficulty in the decision-making process and postdecision doubt were not assessed separately.

Both husband and wife were required to participate in the follow-up study. In case of disagreement between spouses, the answer which might indicate worries concerning the health of a future child was selected. For example, if one spouse had not felt relieved after counselling, this would become a concern for the couple in their decision-making process. Thus the couple would be considered to have felt no relief after genetic counselling even if one spouse had felt relieved. Interpersonal differences might also appear in respect to other factors relevant to problems in the decision-making process. Evaluation of the influence of disagreement between spouses on problems in making the reproductive decision will be investigated at a later date.

FIGURE 1. Problems in making the reproductive decision after genetic counselling



* The DIFF couples are those who had made a decision and have experienced the decision-making process as difficult

DIFF couples were compared with those who did not experience the decision-making process as difficult. Similarly, DOU couples were compared with those who had no doubts about the decision they had made. Couples who were undecided were compared with those that had decided to have children. The latter group is described in detail elsewhere (Frets et al., 1990a; 1990b).

A previous publication lists the disorders involved in the study (Frets et al., 1990a).

Statistical analysis

To identify differences, the relative risk (RR) was estimated by the odds ratio with the levels of significance (p values) being two-tailed. In case of trichotomies such as the genetic and recalled risk level, the Chi-square test for trend was applied (Breslow and Day, 1980). If cell entries in the table equalled zero, 0.5 was added to each cell to estimate the odds ratio (Woolf, 1955).

From the analysis of single factors significantly associated with the criteria (DIFF, DOU or UNDEC) no insight can be obtained into the overlap of the significant single factors on the criteria. Therefore, stepwise logistic regression analysis was used to identify those factors independently and significantly associated with the criteria (DIFF, DOU or UNDEC). This analysis allows for associations with multiple factors simultaneously. It selects each factor in terms of its strength of independent association with the criteria (DIFF, DOU and UNDEC), while controlling other associated factors. Furthermore, it provides an estimate of the odds of the association between each factor and the criteria. The factors selected in the final analysis are presented with the unstandardized regression coefficient, standard errors and the coefficient divided by the standard error. This latter measure indicates the statistical significance of the association between the factors and the criteria (a value of > 2.0 implies a statistical significant level of $p < .05$). This analysis excludes those subjects who left at least one item of the questionnaire unanswered. To enable comparison between single factors in relation to the criteria and the results of the logistic regression analysis, those couples who left at least one question unanswered were excluded from the statistical analysis.

RESULTS

Of the 164 couples under study, 137 had made a reproductive decision: 109 (66%) had decided to have (more) children, while 28 (17%) had decided to refrain from having children. Eighteen couples (12%)[1] were undecided at the time of the follow-up. The remaining 9 couples were excluded from the statistical analysis - 6 cases because of failure to answer all questions (all being couples who had decided to have children) and the other 3 cases because the reproductive decision was basically different: 2 couples opted for artificial insemination by donor, and one (1%) couple waited for prenatal diagnosis to become available in the near future. Thus, the statistical analysis applied to 155 couples.

Figure 1 shows that 47 couples (30%) who had made a decision were DIFF couples. Twelve (8%) of those were also DOU couples. One DOU couple (1%) did not experience the decision-making process as difficult. Thus, of those who had made a decision 13 (12+1=9%) were DOU couples. Eighteen couples (12%) had not made a decision (=UNDEC). Of all 155 couples eligible for the study, 43% ((47 DIFF + 1

[1] The seemingly higher proportion of couples that remained undecided in comparison with the results of Chapter II (12% vs 11%) is due to the smaller number of couples involved (155 vs 164)

DOU + 18 UNDEC)/155)[2] had problems in making the reproductive decision.

Table I lists the factors which were significantly related to DIFF, DOU or UNDEC. The strength of the relationships is indicated as the relative risk (RR). The more the relative risk deviates from 1.0, the stronger the association with the criteria.For example, a couple anticipating a high risk level prior to counselling was 2.67 times more likely to become a DIFF couple than when the risk level was expected to be low (RR 2.67). Couples who did not feel relieved after genetic counselling were more likely to become a DIFF, DOU or UNDEC couple than those who felt relieved (RR 8.02, 5.10 and 14.36, respectively).

The higher the genetic risk the stronger the association with the criteria (DIFF, DOU, UNDEC). Couples with a genetic or recalled risk > 15% were most likely to become DOU couples (RR 9.09 and 19.92, respectively). Of the couples that had decided to have children, those who were eligible for prenatal diagnosis were more likely to become DIFF or DOU couples than couples for whom this was not available (RR 2.68 and 6.70, respectively). Of the couples that decided to have children, 45% of those who were eligible for prenatal diagnosis became DIFF couples and 12% DOU couples compared with 23% and 2% of those for whom prenatal diagnosis was not available. Couples with an affected child (irrespective whether this child had died or survived) were more likely to become DIFF or DOU couples than those without an affected child (RR 2.23 and 6.67, respectively). Couples that refrained from having children were more likely to become DOU couples than those opting to have children (RR 5.72). Couples whose relatives disapproved of their decision were more likely to become a DIFF couple than those whose relatives did not meet disapproval (RR 5.35).

No significant associations were found regarding DIFF, DOU or UNDEC couples and the following single factors: death or survival of an affected child, the presence of healthy and affected children or only healthy child(ren), the presence or absence of mental retardation in the disorder, the type of and perceived severity of the disorder, parental age and whether the couple was at risk for a chromosomal abnormality or a disorder with a multifactorial mode of inheritance. This latter could only be assessed in couples at risk for *one* disorder.

Any unresolved doubts experienced by couples that had decided to undertake a pregnancy after genetic counselling were not related to the subsequent health of this child.

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[2] In the 43% experiencing problems in the decision-making process the overlap of DIFF and DOU couples was excluded.

TABLE I. Problems in making the reproductive decision in 155 couples: factors significantly related to difficulty in the decision-making process (n = 47), unresolved doubts (n = 13), or persisting reproductive uncertainty (n = 18).

	Difficult*	Doubtful*	Undecided**
	RR***	RR	RR
Anticipated high risk level	2.67[c]	ns	ns
No postcounselling relief	8.02[a]	5.10[c]	14.36[a]
Genetic risk: 5-15%	3.38[c]	8.33[d]	5.82[d]
>15%	4.89[c]	9.09[d]	6.86[d]
Recalled risk: 5-15%	3.55[a]	9.67[d]	.83[c]
>15%	9.67[a]	19.92[c]	5.08[c]
Risk interpreted as high	3.10[c]	5.55[c]	4.86[c]
Couples opting to have children: Prenatal diagnosis available	2.68[d]	6.70[d]	-
Had child(ren) during genetic counselling	ns	4.55[d]	ns
Presence of affected child	2.23[d]	6.67[c]	ns
Personal acquaintance with disorder	10.71[c]	ns	ns
Decided not to have children	ns	5.72[c]	-
Relatives' disapproval of decision	5.35[a]	ns	

[a] $p << .001$ [b] $p < .001$ [c] $p < .01$ [d] $p < .05$; ns = not significant ;

* 137 couples had made a decision;
** 109 couples opted to have children and were compared with those who remained undecided;
*** RR = relative risk estimated by odds ratio's

Stepwise Logistic regression analysis

All factors that had a statistically significant relationship with DIFF, DOU or UNDEC were entered into the stepwise logistic regression analysis.

Table II shows the factors independently and significantly associated with problems in the decision-making process: no postcounselling relief (1), anticipation of a high risk level (2), relatives' disapproval of the decision (3), having decided against having children (4) and the presence of an affected child (5).

TABLE II. Stepwise logistic regression for couples experiencing difficulty with the decision-making process or subsequent doubts

	Coefficient	Standard error	Coeff/s.e.*	exp (coefficient)
DIFFICULTY				
1. Relieved by counselling	-2.11	0.45	-4.71	0.12
2. Anticipated high risk level	0.91	0.44	2.04	2.48
3. Relatives disapproving of the decision	1.96	0.62	3.16	7.07
constant	-0.44	0.40	-1.10	0.64
DOUBTFULNESS				
4. Decided not to have children	-1.58	0.63	-2.49	0.21
5. Presence of affected child	1.73	0.65	2.64	5.64
constant	-2.02	0.63	-3.23	0.13

* Coefficient divided by standard error For all factors: 1 = yes and 0 = no

Couples who felt relieved after genetic counselling were less likely (8.33 times = 1/0.12) to become a DIFF couple than those who did not feel relieved (1). Twenty-three of the 35 DIFF couples stated that counselling had not brought relief because (a) the risk was perceived to be high (n=13) or (b) genetic counselling had not provided a definitive mode of inheritance in their case in the absence of a postnatal diagnosis (n=10). Couples that anticipated a high risk were more likely (2.48 times) to become a DIFF couple than those anticipating a low risk level (2). Couples whose relatives subsequently disapproved of their decision were more likely (7.07 times) to become a DIFF couple than those who did not meet disapproval (3).

Couples that decided against having a(nother) child were more likely (4.76 times = 1/0.21) to become a DOU couple than those who decided to have children (4). Seven DOU couples decided not to have children, in 4 cases because they felt they had no choice. Reasons given were: "we did not want another affected child, therefore we could not undertake another pregnancy" and "rationally we know that we have made the right decision, but emotionally it is very difficult to accept".

Couples who had an affected child were more likely (5.64 times) to become a DOU couple than those who did not have an affected child (5). Couples with an affected child that decided against a subsequent pregnancy had found the care of their affected child very demanding.

The anticipation of a high risk level in DIFF couples was not related to (a) precounselling worries or postcounselling relief, (b) genetic or recalled risk category or (c) risk interpretation.

Couples feeling relieved after genetic counselling were less likely (14.36 times) to remain undecided than those who had not experienced relief. This appeared to be the only factor that contributed significantly to the differentiation between couples that remained undecided and those opting to have children.

DISCUSSION

Altogether, 43% of the study population had experienced problems in the postcounselling reproductive decision. Our finding that 12% of the couples remained undecided after counselling was in agreement with the literature after correction for variations in the follow-up intervals. Only Emery et al. (1979) found no reproductive uncertainty at 2 years' follow-up. This may be because at that stage consultands had been assessed 3 times, which might have provided additional support in the decision-making process.

The postcounselling decision is never easy to make (Lippman-Hand and Fraser,

1979a) but some couples seem to experience more difficulty than others. The logistic regression analysis revealed several factors which were independently and significantly associated with problems in the decision-making process. These were no postcounselling relief, anticipation of a high risk level, relatives' disapproval of decision, having decided against having children and the presence of an affected child. Jointly these factors can substantially differentiate couples that will develop problems in the decision-making process from those who will not.

Where genetic counselling had not brought relief due to the unavailability of a precise diagnosis, couples tended to experience additional difficulty in the decision-making process. Kessler et al. (1984) pointed out that feelings of guilt and self-blame tend to increase when the circumstances surrounding the cause and nature of the defect are ambiguous. Because of this ambiguity the couple may be preoccupied with the guilt feelings so that their decision-making is shrouded in emotions (Kessler, 1979; 1984). Therefore it is important to explore the rational as well as emotional aspects of the reproductive decision during counselling. Such exploration might facilitate the decision-making process.

The absence of postcounselling relief appeared to be the only factor contributing to the inability to make a decision. The reasons given for remaining undecided showed some similarity with the reasons given by couples who had decided not to have children, such as high risk interpretation or fear of not being able to cope with another affected child (Frets et al., 1990b).

It is possible that the desire to have children was stronger for couples who remained undecided than for those who had decided against having children. At any rate the desire was not strong enough for the latter group to counterbalance the fears of having a(nother) affected child.

Couples who expected their risk to be high tended to experience additional difficulty in the decision-making process. The reason for this remains unclear. At the very beginning the genetic counsellor needs to find out whether the consultand is anxious about an anticipated high risk. Talking about this might bring relief, even if the counsellor can not provide a risk level at that stage.

Couples whose relatives disapproved of the decision tended to experience difficulty in the decision-making process, as if they had sensed that disapproval would be forthcoming. This may not have occurred in the consultand's consciousness. The influence of the reaction of other people on the decision-making process was also stressed by others (Rangell, 1976; Janis and Mann, 1976; Lippman-hand and Fraser, 1979b). It is therefore important to find out during counselling whether the consultands anticipate disapproval of any decision from relatives.

Couples who decided to refrain from having children tended to have unresolved doubts about their decision. If genetic counselling seems to leave in the perception of the consultands no other option than to refrain from having children additional counselling may be beneficial. In couples refraining from having children, one or both parents might have experienced the birth of their affected child as a punishment. Taking the entire responsibility for the affected child, this spouse may be bolstering his/her self-esteem by denying him/herself the pleasure of having another child (Kessler, et al., 1984).

Wertz et al. (1984) and Sorenson (1981) found that couples who had an affected child tended to remain undecided. Our findings of unresolved doubt in the presence of an affected child are similar.

Couples with unresolved doubts have been analyzed as a separate group, even though nearly all these couples had experienced the decision-making process as difficult. The factors associated with postdecision doubtfulness are different from those related to experiencing the decision-making process as difficult.

Some single factors need to be mentioned, even though these were not independently and significantly related to problems in the decision-making process. Interestingly, the availability of prenatal diagnosis appeared to increase rather than decrease difficulties and doubtfulness for those couples opting to have children. This contradicts the notion that prenatal diagnosis would provide the easy way out. Difficulty experienced by these couples eligible for prenatal diagnosis might be due to the uncertainty of their test result at the time. In half the couples only extreme fetal abnormalities could detected by fetal ultrasound or by sex-determination in couples at risk for an X-linked disorder. Other investigators stressed the period of anxiety while awaiting the result of prenatal testing (Donnai et al., 1981) and the burden of selective abortion in the case of fetal abnormality (Blumberg et al., 1975; Donnai et al., 1981; Lloyd and Laurence, 1985). The availability of prenatal diagnosis of a specific disorder such as cystic fibrosis or neural tube defect appeared to facilitate the reproductive decision-making process (Laurence and Morris, 1981; Evers-Kiebooms et al., 1988) because of the widened scope of choices. However, these workers did not include the burden of selective abortion in their assessment and as such their findings are not really comparable with our results. Difficulty experienced by the couples eligible for prenatal diagnosis in our study might be partly due to the technical limitations of prenatal diagnosis at that time. Sex-determination by amniocentesis or detection of major fetal abnormalities by ultrasound could only provide a restricted answer whether the fetus was affected with the disorder in the family.

In agreement with the literature we found that couples who interpreted their risk as high tended to experience the decision-making process as difficult (Lippman-Hand and Fraser, 1979a) or to remain undecided (Abramovsky et al., 1980; Wertz et al., 1984).

Due to the retrospective design of this study, no firm conclusions can be drawn from the reports of precounselling feelings. The recollection might have been distorted by the outcome of genetic counselling or the birth of an affected child after counselling, a mechanism called anchoring (Pearn, 1973; Tversky and Kahneman, 1974; Wertz et al., 1984). In our study, however, couples who perceived the disorder in their family as severe, seemed to have a particularly vivid memory of the period before and immediately after genetic counselling. Considering the fact that this group did not differ from the entire study population in other respects, we feel that the recollection of the precounselling emotions would be similar for the whole study population. A prospective study is required to substantiate this premise.

The emotional impact of the factors which can substantially differentiate couples who are developping problems in the decision-making process from those who will not, can be discussed during counselling. Not all couples experiencing problems in the decision-making process will need counselling provided by a psychologist or psychosocial worker familiar with their specific problems. Talking about these subjects might facilitate the decision-making process. However, the emotional impact of these factors might only become apparent after counselling rather than in the course of it. Therefore, a second questionnaire was designed based on these factors, to be used 3-6 months after genetic counselling for couples requesting genetic counselling for own offspring. This questionnaire will be tested in a forthcoming study. This second questionnaire also gauges the need for additional support, although not all couples that need support will accept the offer.

When consultands are still undecided 9 months after genetic counselling, they may benefit from counselling provided by a psychologist or psychosocial worker familiar with their specific problems.

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CHAPTER VI.
CHARACTERISTICS OF THE POSTCOUNSELING REPRODUCTIVE DECISION-MAKING PROCESS: AN EXPLORATIVE STUDY

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ABSTRACT

An in-depth, recorded interview of 30 couples 2-3 years after genetic counseling explored the characteristics of the postcounseling decision-making process, including the role of guilt. The study concerned couples with an affected child, sibling or spouse. Results were evaluated by two to four judges. In contrast to other studies a generally unstructured decision-making process was found whereby guilt feelings played a significant role in more than half the couples. Guilt feelings were more predominant in couples with an affected sibling than in those with an affected spouse. Lack of structure did not seem to complicate the decision-making process. Therefore, authors do not advocate promotion of structuring the decision-making process. Genetic counselors might focus on understanding counselees' feelings concerning the reproductive decision. Acceptance of apparently irrational considerations is particularly important, because these feelings indicate the influence of unconscious motives. Another important aspect of supporting counselees is to understand the role played by guilt feelings towards parents or an affected sibling.

KEY WORDS: family planning, follow-up, genetic counseling, qualitative analysis, reproductive decision-making, risk interpretation.

INTRODUCTION

One of the main goals of genetic counseling is to provide information counselees can use to make a reproductive decision allowing for personal and social considerations (Emery, 1984).

In the last decade many studies have been carried out to assess factors influencing the reproductive decision after genetic counseling using questionnaires or semi-structured interviews applying quantitative analysis (Kessler, 1989; Frets et al., 1990a). However, the qualitative characteristics of this decision-making process have rarely been studied. Lipmann-Hand and Fraser (1979a; 1979b) found that all counselees used scenarios in their postcounseling decision-making process to neutralize the uncertainty about the outcome of various options. Scenarios are defined as a sequence of consequences the counselees imagine as outcome of a decision. One of these scenarios is "trying out the worst", for example trying to imagine that they had an affected child and wondering if they could cope with this. It is assumed that parents who feel that they could cope with a severely affected child may decide to have children. This way of deciding is a cognitive process[1].

The present study was undertaken to characterize the process by which reproductive decisions are made. Answers were sought to six questions. First, do couples follow the sequential steps that are prerequisites for a "high quality" decision: (a) consider the available options with all pros and cons, (b) collect all available information, (c) assimilate new information without bias, (d) plan thoroughly how to implement the decision and (e) adhere to the decision in the long run (Janis and Mann, 1976)? Second, do all couples use scenarios before they make their reproductive decision (Lippman-Hand and Fraser, 1979b)? Third, do counselees feel they have been influenced in their final decision by the attitude of relatives? Fourth, do guilt feelings towards the proband play a role in the decision-making process? Fifth, do the decision-making characteristics differ comparing couples with an affected child, with an affected sibling, or with an affected spouse? Sixth, do feelings concerning the proband's disorder influence the decision-making characteristics?

In the present study we focused on the decision-making process of couples opting for or against having children of their own.

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[1] Cognition is a general term covering all the various modes of knowledge - perceiving, remembering, imagining, judging and reasoning (Drever, 1976).

POPULATION AND PROCEDURE

Study population

During 1984, the department of Clinical Genetics counseled some 500 couples, most of them with complex family histories of mental and/or physical handicaps. Counseling included a diagnostic work-up, family history, estimation of the risk level, and discussion of various options to prevent the birth of an affected child. A large follow-up study involving 164 couples was carried out 2-3 years later to assess reproductive planning after genetic counseling (Frets et al., 1990b; 1990c; 1990d). Only those couples who had requested genetic counseling for their own offspring were entered into the study.

Three levels of genetic risk were distinguished: a "low" genetic risk (< 5%), a "moderate" risk (5-15%) and a "high" risk (>15%). The "low"-risk group being the largest, only a random selection of couples from this group was enrolled in the study to ensure equal representation of each category. A detailed description of the exclusion criteria has been reported elsewhere (Frets et al., 1990b).

The sample of 164 couples was divided into 4 subsets: (1) couples with an affected child (n=77), (2) couples with an affected sibling (n=28), (3) couples with an affected (distant) relative, other than sibling (n=22), and (4) couples of whom either spouse was affected (n=37). Couples with an affected spouse as well as an affected child were assigned to group 1 because the affected child was the dominant reason for counseling (11 of 77). Couples with an affected spouse as well as an affected sibling were assigned to group 4 because the affected spouse was the dominant reason for genetic counseling (18 of 37).

The present study explored the degree of acceptance of the proband's disorder. To obtain a reasonable basis for comparison of feelings concerning the proband, only counselees from group 1,2 and 4 were studied. The reason for excluding group 3 was that these counselees may not have been as personally acquainted with the proband, e.g. a second cousin as those in group 1,2 or 4. Ten couples were randomly selected from each of these 3 groups.

In view of the explorative nature of the study a sample of 30 couples was considered sufficient.

Regarding the genetic risk level estimated by the genetic counselor there were 11 couples with a risk under 5%, 11 fell in category 5-15% and the remaining 8 couples had a genetic risk over 15%.

The classification of Ekwo et al. (1987) was used for the type of disorder. For the couples with an affected child the disorders were postnatal death (n=6); mental retardation (n=3); and chronic incapacitating illness (n=1). For the couples with an affected sibling the disorders were postnatal death (n=1); mental retardation (n=6); chronic incapacitating illness (n=2); and facial abnormalities (n=1). For the couples

with an affected spouse the disorders were postnatal death (n=2), because one of the spouses was a carrier of a balanced chromosomal translocation; chronic incapacitating illness (n=5); skeletal problems (n=2); and facial abnormalities (n=1).

The educational level of the study population was significantly below that of the general Dutch population[2] (Chi-square 15.90, df=6, $p < .05$ and 28.63, df=6, $p < .001$).

Procedure

Between 1986 and 1987, the couples under study were interviewed at home by a psychologist (PGF). The in-depth interviews were registered on audiotape and lasted 2 to 3 hours. Subsequently, the interviews were randomly divided into three subgroups of ten each. The results were evaluated independently by four judges. The judges were either psychotherapists or psychoanalysts with at least 10 years of professional experience. Two judges looked at the results of one subgroup, the two other looked at the second subgroup, and all four judges evaluated the third subgroup.

The first part of the interview explored the characteristics of the reproductive decision-making process, focusing on the period after genetic counseling.

The first objective concerned the "quality" of the decision. Counselees were asked whether they felt they had collected all additional relevant information, thus apart from the genetic counseling information, such as genetic publications from libraries or other sources. They were also asked whether they felt they had been receptive to all this information. Counselees were asked whether they had considered all available options, weighing the pros and cons of each option. Did their choice of option include implementation of the consequences? For example, did the decision to have a(nother) child include plans for a thorough medical work-up once the child was born? Counselees were also asked whether the decision had left any unresolved doubts. Finally, counselees were asked whether they had experienced the decision-making process as difficult. The judges had to ascertain whether counselees had been receptive to the genetic information, consciously as well as unconsciously. The judges had to determine the presence of doubt on a conscious as well as an unconscious level. The question about doubtfulness was asked to assess adherence to the decision in the long run.

The second objective was to determine whether counselees used scenarios in their decision-making process. The couples were asked whether they had imagined the consequences of at least some of the available options, e.g. to have a child that may be

[2]Dutch Office for Statistics, Voorburg, The Netherlands

affected, to refrain from having any (more) child(ren), artificial insemination by donor (=AID), prenatal diagnosis with selective abortion, adoption, to raise a foster child, etc.. Specifically, they were also asked whether they had imagined the consequences of having a child that may be affected.

The third objective focused on the consideration given to attitudes and opinions of relatives in counselees' final decision.

Hereditary disorders are often associated with feelings of guilt. The judges had to determine to what extent conscious and/or unconscious guilt feelings had played a role in the decision-making process.

The second part of the interview focused on the degree of acceptance of the proband's disorder. Counselees' feelings concerning the disorder(s) affecting their child, sibling or spouse were explored. Any negative effect of the disorder on counselees' self-esteem was explored by indirect questions, such as "Do you ever feel offended by the fact that a member of your family is affected by a disorder?" or "Do you ever take it personally that a disorder occurs in *your* family?". An affirmative answer to either of these two questions was considered to indicate a lowered self-esteem. Counselees were also asked whether they had felt ashamed of the proband's disorder, tempting them to hide this disorder. The judges were asked to ascertain whether the proband's disorder had had a negative effect on counselees' self-esteem and whether the counselees felt ashamed, both on a conscious and unconscious level. In the same context, the judges had to look for signs of guilt feelings towards the proband on both a conscious and unconscious level. Finally the judges were asked to determine the degree of acceptance of the proband's disorder.

Both husband and wife were required to participate in the study. The answers given by either spouse were evaluated separately. If the answers differed, the answer used was the one that might impede the decision-making process. For example, the couples were marked as not having considered all available options even if one of the spouses had done so, because failure to do so by the other spouse would impede the joint decision-making. Likewise, if one spouse had not been able to accept the disorder this might result in preoccupation with the disorder, regardless of the fact that the other spouse had accepted it. Preoccupation with the disorder would result in increased anxiety regarding a future child being affected.

The judgments of the degree of acceptance of the proband's disorder was graded on a 10-point-scale, all remaining items on a 5 point-scale. Only the 2 extremes were labeled on these scales veering from "not at all" on the left to "to a great extent" on the right. To achieve acceptable final scores with such semi-quantitative data the following guidelines were established.

In case of evaluation by two judges only:

- if the difference in grading amounted to no more than 2 points on the 5 point-scale or 3 points on the 10 point- scale, the average was taken as final score. With a greater divergence there was no final score.

In case of evaluation by four judges:

- the average became the final score with the following exceptions: if one of the individual gradings showed a divergence from the average of 1.5 or more on the 5 point-scale and 2.0 or more on the 10 point-scale, this figure was eliminated and a new average was taken from the remaining three. If this divergence applied to more than one individual grading there was no final score.

All items involving conscious as well as unconscious levels, would result in two final scores. In case of divergence the maximum score applied.

The final scores on the 5 point-scale were divided into three categories of equal range categories: < 2.33 = "not at all", 2.33- 3.67 = "to a certain extent", > 3.67 = "to a great extent". On the 10 point-scale the range for the same categories became $<$ 4.0, 4.0-7.0 and > 7.0.

An *item* was excluded from the statistical analysis if no final score could be established regarding this item for six couples or more. To identify differences, the relative risk (RR) was estimated by the odds ratios ($p < 0.05$; two-tailed). If cell entries in the table equalled zero, 0.5 was added to each cell to estimate the odds ratio (Woolf, 1955). Regarding the relation between feelings towards proband's disorder and the decision-making characteristics or the proband category and these characteristics the scores "not at all" and "to a certain extent" were combined.

RESULTS

The results of 5 couples were excluded from the analysis. One spouse of two of these couples appeared unresponsive to the interview, in one case due to alcoholism; one couple proved to have an infertility problem which dominated the interview; one couple had resorted to AID and one couple was still undecided. The latter two couples were excluded because this study focused on couples that had made a decision for or against having children of their own.

Of the remaining 25 couples, 22 had decided for and 3 against having children. Seven couples had an affected child, 10 an affected sibling and 8 an affected spouse. The inter-rater-reliability did not differ for the number of judges, as was shown by a similar proportion of absent final scores for couples evaluated by all four judges (4%) and for those evaluated by only two judges (6%).

Table I shows the distribution of the characteristics of the postcounseling decision-making process. The numbers for the evaluation refer to the couples, including a division

TABLE I. Characteristics of the postcounseling reproductive decision-making process (n = 25)

	No N (C,S,P)	To a certain extent N (C,S,P)	Yes N (C,S,P)	Total N (C,S,P)
A. Collected relevant information	*3* (0,2,1)	*11* (3,3,5)	*10* (4,5,1)	*24* (7,10,7)
B. Receptive to new information	*3* (2,0,1)	*4* (3,0,1)	*16* (2,10,4)	*23* (7,10,6)
C. Considered available options	*13* (4,5,4)	*7* (2,4,1)	*5* (1,1,3)	*25* (7,10,8)
D. Weighed pros and cons	*15* (4,6,5)	*6* (1,3,2)	*3* (1,1,1)	*24* (6,10,8)
E. Imagined the consequences of various options	*7* (2,2,3)	*12* (4,5,3)	*4* (1,2,1)	*23* (7,9,7)
F. Decision influenced by relatives	*20* (5,8,7)	*3* (2,1,0)	*2* (0,1,1)	*25* (7,10,8)
G. Unresolved doubt	*22* (6,8,8)	*2* (1,1,0)	*0*	*24* (7,9,8)
H. Difficulty in the decision making process	*11* (4,4,3)	*5* (1,1,3)	*8* (2,4,2)	*24* (7,9,8)
I. Guilt feelings played a role	*5* (1,1,3)	*6* (2,1,3)	*14* (4,8,2)	*25* (7,10,8)

C (= child); S (= sibling); P = partner (=spouse) affected

per subset. The total number of couples was different for each item due to the absence of final scores ranging from 0 to 2.

Most couples (16/23) appeared receptive to relevant information (B), whereby couples with an affected sibling were more likely to be receptive than those with an affected child (Relative risk 46.2, $p < 0.005$).

It appeared that 4 of 23 couples had imagined the consequences of various options (E); of the remaining 19 couples, 6 had limited their choice to having children of their own or not at all, while 5 did not feel the need to consider other options because of low risk interpretation. The item "imagining the consequences of having an affected child" was excluded, because no final score could be established for 8 couples.

Most couples (20/25) claimed that the attitudes of relatives had not influenced their decision (F). Likewise, the majority (22/24) claimed that their decision had left no unresolved doubts (G).

For more than half the couples (14/25) guilt feelings had clearly played a role in the decision-making process, in all 14 cases unconsciously as well as consciously (I). Guilt feelings were significantly more likely to play a role in the decision-making process for couples with an affected sibling than for those with an affected spouse (Relative risk 12, $p < 0.05$).

Table II gives the incidence of lowered self-esteem, feelings of shame, or acceptance of the proband's disorder. For the first two items the incidence appeared low (2/23 and 4/25). Eight of 24 couples had accepted proband's disorder, 6 of them with an affected sibling. Feelings towards proband's disorder were not significantly associated with the characteristics of the decision-making process.

TABLE II. Incidence of lowered self-esteem, feelings of shame, or acceptance of the proband's disorder (n = 25).

	No N (C,S,P)	To a certain extent N (C,S,P)	Yes N (C,S,P)	Total N (C,S,P)
Lowered self-esteem	*11* (3,6,2)	*10* (3,4,3)	*2* (0,0,2)	*23* (6,10,7)
Sense of shame	*10* (4,4,2)	*11* (2,5,4)	*4* (1,1,2)	*25* (7,10,8)
Acceptance of disorder	*2* (0,0,2)	*14* (6,4,4)	*8* (1,6,1)	*24* (7,10,7)

C = child affected; S = sibling affected; P = partner (=spouse) affected

DISCUSSION

This explorative study showed that in most couples the decision-making process was unstructured because these couples did not follow the sequential steps of a balanced decision-making process as described by Janis and Mann (1976) and did not use scenarios as defined by Lippmann-Hand and Fraser (1979a; 1979b). The lack of structure is understandable because the postcounseling reproductive decision is an emotional one. Moreover, the motives underlying the desire for parenthood are in most instances not in the person's consciousness (Kessler, 1979a). The use of cognitions may even serve as a defense against the often ambivalent emotions involved in the reproductive decision-making process (Kessler, 1979b).

The decision analysis theory favors structuring the postcounseling reproductive decision-making process (Humphreys and Berkeley, 1987; Pauker and Pauker, 1987). Studies on structuring the postcounseling decision-making process of people confronted with a hypothetical genetic risk and the subsequent reproductive decision revealed inconsistency on the usefulness of structuring this process, as perceived by these people (d'Ydewalle and Evers-Kiebooms, 1987; Pitz, 1987). The results of the present study seemed to indicate that a lack of structure did not unduly complicate the decision-making process.

Only in a minority of the couples cognitions played a role in the decision-making process. Four of our couples structured their decision-making process by imagining the consequences of various options prior to making a decision (=scenarios). These four couples were slightly better educated than most and might therefore be more capable of expressing their thoughts. Comparing these four couples with the remainder did not reveal any difference as to difficulty in the decision-making process.

The unstructured decision-making process might be partly due to the fact that the strong desire to have children overrules any consideration of other options. The decision-making process was unstructured for six of seven couples with a very strong desire to have children as determined in a previous study (Frets et al., 1990c). The small number does not allow for a definite conclusion.

In contrast to our study, Lippman-Hand and Fraser (1979a; 1979b) found that all counselees use scenarios in the postcounseling reproductive decision-making process. The discrepancy in findings might be due to differences in: methods of genetic counseling (e.g. with or without promotion of scenarios); study population; phrasing of the follow-up questions; or methods of analysis.

The results of the present study indicate that the genetic counselor does not necessarily have to promote structuring of the decision-making process (e.g. the use of scenarios). The genetic counselor can use another approach to support the counselees in their decision-making process. All aspects should be open to discussion, while the feelings and opinions of counselees should be respected. The reproductive decision may be based on unconscious motives and feelings and as such appear irrational. By accepting irrational considerations, the counselor may encourage a discussion of these considerations which will help clarifying the influence of unconscious emotions. For example, a couple with a 50% risk of having an affected child may be convinced that their child will be healthy. The inability to imagine having an affected child may ward off extreme anxieties about this happening. Bringing this anxiety into the open might bring relief and help counselees to face realities.

Janis and Mann (1976) postulated that a "high quality" decision has a better chance of being adhered to in the long run. The majority of our study population had made a "low quality" decision while only two couples had unresolved doubts.

More than half the couples were receptive to relevant information. In this respect, the literature shows that interpretation of such information is more important for the reproductive decision than the genetic facts (Frets et al., 1990a; Kessler, 1989). Surprisingly, couples with an affected sibling tended to be more receptive to relevant information than those with an affected child. Couples with an affected sibling may have had more opportunity to distantiate themselves from the disorder emotionally than parents

of an affected child. This is supported by the finding that six of ten couples with an affected sibling had accepted the disorder emotionally compared with only one of seven with an affected child.

Almost all couples claimed to have made the decision by themselves. This seems to be in contrast with previous findings from a questionnaire study among these counselees, that disapproval of relatives complicated the decision-making process (Frets et al., 1990d). The findings from the questionnaire study may be more relevant as these were based on an indirect approach. The latter method is more suitable to reveal unconscious feelings than the direct approach of the in-depth interview concerning this item. Janis and Mann (1976) found that the attitudes of significant others did influence the final decision. They assessed a variety of decisions in which more conscious considerations are involved, whereas the reproductive decision can be heavily influenced by unconscious motives (Kessler, 1979a).

The present study showed clearly that guilt feelings played a role in the reproductive decision-making process, but the exact role was not revealed. Guilt feelings did not seem to make the decision-making process more difficult.

Guilt feelings were more frequently observed in couples with an affected sibling (80%) than in those with an affected child (57%) or spouse (25%). Studies assessing the influence of an affected child on the healthy siblings showed that healthy siblings may be at increased risk for psychological disturbance (Drotar and Crauford, 1985; Gath et al., 1989; Eiser et al., 1990). Healthy siblings of a mentally retarded child were more likely to develop behavioral problems if the mental deficiency was associated with behavior, than when this was not the case (Gath and Gumley, 1987). Buchanan (1979) found some degree of emotional distress in 60% of healthy siblings of a patient with Duchenne muscular dystrophy. Fishman (1979) described how comprehension of the genetic nature of cystic fibrosis led healthy siblings of the affected child to fantasize why they themselves were spared, the so-called "survivor" guilt. Guilt feelings might also be engendered in healthy children for feeling angry with their parents or affected sibling (Seligman, 1987). Thus, growing-up with an affected sibling can leave its mark on the healthy children.

Guilt feelings towards an affected sibling may be due to the following mechanisms. First, a couple with a good chance of having healthy offspring may feel guilty towards his or her parents because the latter had to cope with an affected child, while they themselves do not have to face such a disruption of ideals (Fishman, 1979). Second, the affected sibling may trigger "survivor" guilt feelings in couples faced with a reproductive decision. Nearly all the affected siblings in this study population were unlikely ever to become self-supporting, let alone have a family of their own.

In couples with an affected child, guilt feelings may be triggered by the desire to have a healthy child. Five of the seven affected children in this sample had died very young. The desire for a healthy child may have been an (unconscious) attempt to undo the past, a denial that the affected child had ever existed. Guilt feelings may also be triggered by the availability of prenatal diagnosis with selective abortion of a fetus with the disorder of their affected child.

Some approaches to genetic counseling may help to relieve guilt feelings in the decision-making process. The genetic counselor could explain that these guilt feelings are a normal phenomenon. The counselor could discuss that they now know their risk of having an affected child and that they can make their own decision whether to take the risk with or without the use of prenatal diagnosis or to abstain from having (more) children. Another approach is to support counselees by emphasizing the positive rather than the negative aspects of the reproductive decision. The counselor could discuss that the counselees have had an emotionally burdensome time after the discovery of the disorder in their child or family. Subsequently, the counselor could stress the fact that counselees deserve admiration for the courage it takes to make such a difficult decision.

In contrast to the literature (Antley et al., 1973; McCollum and Silverberg, 1979; Corgan, 1979; Targum, 1981; Kessler et al., 1984; Broder and Trier, 1985; Messner and Smith, 1986; Miller et al., 1986/87) hardly any couple clearly experienced feelings of lowered self-esteem or shame concerning the proband. These feelings only occurred in half the couples to a certain extent. The discrepancy with the literature may be due to the small study population.

CONCLUSION

This explorative study showed that couples make their postcounseling reproductive decision in an unstructured way and that this lack of structure did not seem to have a detrimental influence on the decision-making process. Genetic counseling may focus on understanding counselees' feelings concerning the reproductive decision. Acceptance of apparently irrational considerations is particularly important, because these feelings indicate the influence of unconscious motives. A major aspect of supporting counselees is to understand the role played by guilt feelings towards parents or an affected sibling.

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CHAPTER VII.
GENERAL DISCUSSION

RESULTS OF PRESENT STUDY IN COMPARISON WITH OTHER FOLLOW-UP STUDIES

The present study attempted to monitor the transfer of information during genetic counseling. Furthermore, the adequacy of the existing strategies for supporting counselees in their decision-making process was investigated by comprehensive assessment of various factors influencing the reproductive decision as well as the problems experienced in the decision-making process.

In agreement with other studies (Black, 1979; Springer and Steele, 1980; Abramovsky et al., 1980; Seidenfeld and Antley, 1981; Swerts, 1987; Somer et al., 1988) our study showed that over two-thirds of couples (69%) correctly recalled the genetic risk range. Eighty-five per cent correctly recalled their eligibility for prenatal diagnosis. Thus, the transfer of information was highly satisfactory.

Counselees were satisfied with the way genetic counseling was provided. Almost all counselees (90%) thought that the genetic counselor gave the information in a clear way. The majority of the counselees ($\pm$ 80%) thought the comprehensive, written summary of the information presented and discussed during counseling was clear and useful. Most counselees said they had benefited from genetic counseling in that this enabled them to make an informed decision. However, these data on the degree of satisfaction may be somewhat distorted by the tendency of people to give socially desirable responses (Kessler, 1989).

Most follow-up studies focus on single factors influencing the reproductive decision after genetic counseling (=univariate studies). A good number of these studies were carried out by Evers-Kiebooms et al. (1980; 1984; 1988). Univariate studies demonstrated that the genetic risk was only of relative importance for the reproductive decision (Bocsknov, 1979; Emery et al., 1979; Abramovsky et al., 1980; Czeizel et al., 1981; Kessler and Levine, 1987). In our study the strength of the desire to have children was one of the single factors which was most strongly related to the reproductive decision (Chapter III). Before our study was initiated this desire was found to be merely one of the factors associated with the reproductive decision (Bocsknov, 1979; Emery et al., 1979; Springer and Steele, 1980; Burns et al., 1984; Evers-Kiebooms et al., 1984). However, comparison of results of various follow-up studies after genetic counseling is complicated by insufficient information about the study population and procedure of the

studies (Kessler, 1989).

There seem to be no cross-cultural differences concerning the basic factors influencing the reproductive decision. However, there may be important cross-cultural differences in acceptance and implementation of genetic services and carrier screening programmes among different cultural societies. Modell et al. (1982; 1984; 1988) clearly showed the difference in acceptance of carrier screening for thalassemia and prenatal diagnosis in couples at risk between various ethnic immigrant populations.

A multivariate assessment, investigating a number of factors jointly, seems to be a more realistic approach than assessing the influence of single factors only, because counselees are not able to isolate one factor at a time in the course of their decision-making process. Multivariate assessment also provides insight into the importance of factors for the reproductive decision (Chapter IV). The model showed the importance of the desire to have children (Sissine et al., 1981; Sorenson et al., 1987), the interpretation of the risk (Lippman-Hand and Fraser, 1979c; Sorenson et al., 1987), and the significance of the availability of prenatal diagnosis for the individual couple.

The paramountcy of the desire to have children might be due to the specificity of a genetic counseling population. In clinical practice, there may be an interaction between physicians and patients whereby referral to the geneticist is limited to couples who are considering a(nother) pregnancy. However, the desire to learn about the nature and cause of the disorder in the family might well be an equally important, albeit separate, indication for counseling. This is frequently overlooked by primary health care physicians, the parents themselves and other relatives. At present, only a minority of couples request genetic counseling specifically for this reason. The couples that do request genetic counseling may be those with an exceptionally strong desire to have children. They may see the risk of having an affected child as an impediment to becoming pregnant. This apparent impediment caused them to realize that they very much desire to have children. Without the impediment this realization was not necessary, because the desire could then be fulfilled. It is also possible that the desire to have children is not strong enough to counterbalance the anxiety for a future affected child. These couples decide to refrain from (further) childbearing. This latter decision was made by a considerable number of couples at risk for cystic fibrosis (Kaback et al., 1984), thalassemia (Modell and Mouzouras, 1982) and Duchenne Muscular Dystrophy (Lubs, 1979) before prenatal diagnosis became available.

The multivariate assessment enabled a correct identification of the reproductive decision in 91% of the cases (Chapter IV). Sissine et al. (1981) correctly classified the reproductive decision in 79% of cases. In their qualitative study Lippman-Hand and Fraser (1979a; 1979b) did not provide figures on correct classification of the

reproductive decision. Sorenson et al. (1987) carried out a large prospective study, but unfortunately did not present figures on the proportion of correctly predicted reproductive decisions. These three groups did not develop a quantitative model, but merely indicated which factors were most important for the reproductive decision. The results of the various multivariate studies were inconsistent concerning the importance of (a) the perceived burden of the disorder, (b) the reproductive history, and (c) the influence of the consequences of the diagnosis.

THE AVAILABILITY OF PRENATAL DIAGNOSIS

Prenatal diagnosis as an option for the reproductive decision dates from the early seventies. The present study showed that in the absence of prenatal diagnosis, the genetic risk clearly influenced the reproductive decision when this risk was above 15% (Chapter III). This concurs with findings reported in the literature for specific disorders such as cystic fibrosis or thalassemia (Modell and Mouzouras, 1982; Kaback et al., 1984; Cao et al., 1987). Our study was the first to show the influence of prenatal diagnosis on the reproductive decision comparing couples eligible and not eligible for prenatal diagnosis at risk for various disorders.

Other studies have shown that for couples with a genetic risk of 5% or less of having a child with Down's syndrome or a neural tube defect, prenatal diagnosis is also important: two-thirds of couples eligible for prenatal diagnosis used this option (Laurence and Morris, 1981; Evers-Kiebooms et al., 1984; Adams et al., 1984; Swerts, 1987). Women at risk for chromosome abnormality due to advanced age are not considered here because the latter differs from couples requesting genetic counseling for a disorder in the family. They did not experience the (possible) presence of a genetic disorder in the family which could occur or recur in a future child, irrespective of the maternal age.

Unfortunately, in some circles the emotional burden of prenatal diagnosis with the possibility of selective abortion is erroneously viewed as the easy way out. It has raised concerns about the "slippery slope" (abortions asked because of "less severe disorders" in a fetus). It has been proposed that society may justifiably place limits on the types of conditions for which prenatal diagnosis will be provided (King's Fund Forum Discussion, 1987; Consensus Conference on the Application of Knowledge Gained from Mapping the Human Genome, 1989). We found that the availability of prenatal diagnosis had complicated rather than facilitated the reproductive decision-making process for couples who eventually decided to have children (Chapter V). Thus, the present study revealed that the technology of prenatal diagnosis did *not* provide the easy way out. This clearly demonstrates that parents are aware of their responsibilities. It also shows that there are

no clear indications that parents will seek prenatal diagnosis for so called "less severe conditions", since the couples who seek genetic counseling face the conflict between their desire to have children and their genetic risk with all its implications. Termination of a pregnancy for these couples means the loss of a wanted child.

Others found that the decision to have children appeared to facilitated by the availability of prenatal diagnosis in couples at risk for cystic fibrosis or neural tube defect (Laurence and Morris, 1981; Evers-Kiebooms et al., 1988). This facilitation might be due to the widened scope of choices. However, having to make a decision whether or not to undergo prenatal diagnosis raises fundamental questions, e.g. whether to terminate a pregnancy when fetal abnormality is found. The psychological burden of these considerations did not seem to be incorporated in the studies of Laurence and Morris (1981) and Evers-Kiebooms et al. (1988) and as such their findings are not really comparable with our results.

THE REPRODUCTIVE DECISION-MAKING PROCESS

The present study (Chapter V) revealed that problems in the decision-making process were experienced frequently (43%). This study was the first to show that couples experienced a variety of problems in the decision-making process such as (a) difficulty in the decision-making process, or (b) doubtfulness about the decision they had made, or (c) inability to make a decision. Various factors were significantly and independently related to these problems: (1) no postcounseling relief; (2) anticipation of a high risk level; (3) relatives' disapproval of decision; (4) the decision not to have children; and (5) the presence of an affected child (Chapter V). These issues merit attention for postcounseling follow-up, which is clearly more often indicated than provided in daily practice. Our study was the first to show the importance of no postcounseling relief and anticipation of a high risk level for problems experienced in the decision-making process. The absence of relief was due to the lack of a precise diagnosis in two-thirds of cases. This lack of a precise diagnosis may evoke guilt feelings (Kessler, 1979a; 1984). These feelings may in turn have complicated the decision-making process. This result underlines the importance of a precise diagnosis.

The analysis of problems experienced in the decision-making process (Chapter V) indicated how couples who are likely to experience problems, could be identified and supported. Individual problems specific to a couple can only be revealed during in-depth psychological counseling, exploring the feelings of the couple concerning the 5 factors mentioned above.

A remarkable finding was that the perceived severity of the disorder was not

significantly associated with problems experienced in the decision-making process. This factor may well have had a strong influence in a subgroup, e.g. couples with an affected child, but this may have been overruled by the adverse influence of this factor in a complementary subgroup, e.g. couples without an affected child. The statistical analysis included only single factors, which meant that the influence of the various factors in subgroups could not be identified with this analysis.

The in-depth analysis of how the reproductive decision was made showed the importance of conscious and unconscious affective issues (Chapter VI). Affective aspects are important because unconscious motives are likely to play a major role in the decision-making process (Kessler, 1979b). Interviewing couples with an affected child, sibling or spouse demonstrated that guilt feelings clearly played a role in the decision-making process for more than half the study population. A three-way division of couples based on affected child, sibling, or spouse turned out to be useful. In couples with an affected sibling, guilt feelings were significantly more likely to play a role in the decision-making process than in those with an affected spouse (Chapter VI).

Our questionnaire study (Chapter V) revealed that disapproval of relatives complicated the decision-making process. However, during the in-depth study almost all couples claimed to have made the decision by themselves (Chapter VI). The findings from the questionnaire study may be more relevant as these were based on an indirect approach. The latter method is more suitable to reveal unconscious feelings than the direct approach of the in-depth interview concerning this item. Our impression is that while couples claim to have made their own decision of their own free will, they may unconsciously have been influenced by the anticipated disapproval of their relatives.

COMPARISON OF THE REPRODUCTIVE DECISION OF COUNSELEES AND THAT OF PATIENTS CONCERNING THEIR OWN MEDICAL TREATMENT

The reproductive decision and patients' decisions concerning their own medical treatment are far-reaching and may have long-ranging consequences. However, there are also differences between these decisions. The reproductive decision is intrinsically more complicated as this involves individual and joint objectives and desires, responsibility towards a future child as well as the goals and functioning of the entire family. In view of the similarity, we compared the ways in which these decisions were made. Increased patient participation concerning decisions about their treatment have necessitated guidelines for ways to support patients in their decision. Decision-analysis is a quantitative method to measure patients' preferences and to use these values to reach a rational decision. The outcome with the highest expected utility will become the decision

of choice (Hogarth, 1980). The advocates of decision-analysis claim that it enhances effective decision-making (Pauker 1976; Humphreys and McFadden, 1980; Howard, 1980; Keeney, 1982). Research shows, however, that patients often do not make the decision with the highest expected utility (Slovic et al., 1977; Tversky and Kahneman, 1981; Eraker and Politzer, 1982).

Patients may be biased in their preference of medical decisions due to the way in which the information is presented (Tversky and Kahneman, 1981; Eraker and Sox, 1981). For instance, if it is a choice of two drugs, the patient tends to make a different decision when the physician emphasizes the therapeutic effects of these drugs rather than if the adverse effects were compared. Also, a certain outcome overrules an uncertain one. People prefer the drug with a guaranteed, moderately therapeutic effect over a gamble on either a very good or a very poor result (McNeil et al., 1978). Patients' preferences may be distorted by this so-called 'certainty' effect (Tversky and Kahneman, 1981). Another possible bias of preferences may be the availability effect. This refers to the tendency of people to judge the likelihood of an event by the ease with which this event can be brought to mind (Tversky and Kahneman, 1974). For example, a man refuses to be admitted to hospital although he needs treatment for malignant hypertension, "because" his brother has recently died in a hospital.

Janis and Mann (1977) reviewed a great number of studies on health-related decisions which showed the influence of emotional factors - not necessarily conscious - on the decision-making process. The emotional factors, such as anxiety, can interfere with information processing, lead to overemphasis of arguments in favor of preferred alternatives, or prevent a search for new alternatives.

Patients' decisions concerning medical treatment are influenced by emotional experiences rather than the highest expected utility. This is in agreement with our finding that guilt feelings play an important role in the reproductive decision-making process. How these guilt feelings play a role in the decision-making process is yet unknown.

Rationality and emotions are often in conflict with each other. It is generally very difficult to integrate these two functions. The conflict between rationality and emotions is often seen in counselees who have a very strong desire to have children but know that they are at risk of having an affected child. Being at risk "forces" the couple to consider the situation rationally, which in itself triggers the urge not to do so and thus react emotionally without the ability to integrate these functions.

PRACTICAL APPLICATION OF THE RESULTS

The present study lists the most important factors for the reproductive decision after genetic counseling and the problems and characteristics of the decision-making process. These results were adapted for clinical application.

The flow-chart model which identified the reproductive decision may prove useful in clinical practice in the future (Chapter IV). When a couple has difficulty reaching a decision, following the flow-chart will help to determine which issues are relevant or important for the couple. These issues are: (a) the desire to have children, (b) the interpretation of the risk, and (c) the significance of the availability of prenatal diagnosis for the individual couple. If uncertainty about an issue emerges from the flow-chart model, the genetic counselor may provide help by stimulating discussion of that particular topic. If requested by the counselees, genetic counselors could use the flow-chart model to show what other couples in similar circumstances have decided and for what reasons. Genetic counselors should be aware of the danger of couples feeling that the model dictates what counselees should decide. It is strongly emphasized that this is *not* the purpose of the model.

Analysis of problems in the decision-making process (Chapter V) revealed the factors or issues which require special attention during genetic counseling such as (1) no postcounseling relief; (2) anticipation of a high risk level; (3) relatives' disapproval of decision; (4) the decision not to have children; and (5) the presence of an affected child. One of the important strategies for supporting counselees in their decision-making process is to pay attention to these issues. This would serve to identify couples who are likely to experience problems in the decision-making process. Moreover, discussion of the emotional impact of these factors might facilitate the decision-making process.

The decision analysis theory favors structuring the postcounseling reproductive decision-making process (Humphreys and Berkeley, 1987; Pauker and Pauker, 1987). The results of the studies on this approach in people confronted with a hypothetical genetic risk were inconsistent on its usefulness showing very large differences in individual's ratings (d'Ydewalle and Evers-Kiebooms, 1987; Pitz, 1987). For our study population the lack of structure did not seem to complicate the decision-making process. Therefore, promotion of structuring this process is not advocated. Genetic counselors might rather focus on understanding counselees' feelings concerning the reproductive decision. Acceptance of apparently irrational considerations is particularly important, because these feelings indicate the influence of unconscious motives. These unconscious motives may be in conflict with each other or with counselees' conscious ideas. Another important aspect of supporting counselees in their decision-making process is to

understand the role of conscious as well as unconscious guilt feelings towards parents and affected siblings.

Not all couples will be able to make a decision, despite the support of the genetic counselor. The undecided couples might benefit from support by a psychologist or psychosocial worker familiar with the specific problems in the postcounseling decision-making process. When the couple desires support the psychologist or psychosocial worker needs to identify the psychological conflicts that obstruct the decision-making process, which might be related to problems in accepting the situation. Support requires identification of the defense mechanisms used unconsciously as well as the need for any particular defense mechanism. The function of defense mechanisms is to ward off feelings that would provoke overwhelming anxiety if these feelings would rise to counselees' consciousness. Problems in the decision-making process and/or acceptance of their situation may be related to pre-existing emotional problems of a general nature, which were triggered during genetic counseling. If this becomes apparent, counselees are referred for long-term psychological support.

In some cases, the defense mechanisms are so essential that counselees will become overwhelmed by anxieties if these are torn down. For example, one woman was referred for psychological counseling within our department, because she had difficulty accepting the death of her affected child. This woman had lost her mother when she was 12 and had an abortion at 18 because her then boyfriend did not want the child. She was very reluctant to talk about her emotions concerning these three losses. She herself clearly indicates that she want to suppress her feelings. If she had been forced to experience all these emotions, her vital protection would have been taken from her by force possibly resulting in a total breakdown. Thus, this woman was considered to be better off with her defenses intact.

Hardly any couple who experienced difficulty in the decision-making process, requested additional supportive counseling even though this had been offered during genetic counseling. The small number of couples who had contacted the center again had done so because they wanted to have additional information. Couples who experienced no postcounseling relief were more likely to experience difficulty in the decision-making process. These couples may have felt that the clinical geneticist gave them the 'bad news' which might (unconsciously) have evoked anger in the counselees. Moreover, many couples may feel that they have to accept their problems without the considering the possibility that psychological support might be beneficial for them. It seems of utmost importance to carry out a structured follow-up, 3-6 months after genetic counseling, by using a questionnaire exploring the factors associated with problems experienced in the decision-making process. This is another strategy recommended for supporting counselees

in their decision-making process. This follow-up would identify couples that would benefit from additional support (Chapter V). Obviously, counselees have to make their own decision whether or not they accept the offer for additional supportive counseling.

FUTURE RESEARCH

In the present study factors influencing the reproductive decision and its process were investigated. It seemed best to assess the influence of these factors retrospectively, even though such design may have disadvantages. For example, the precounseling feelings may be distorted by the outcome of genetic counseling, a mechanism called anchoring (Tversky and Kahneman, 1974). In the present study no control group was used. This control group might have consisted of couples who had not received genetic counseling and who desired future offspring even though this was indicated for the same reasons as in the study population. However, such a group was not readily available.

Future research based on the results of the present study

This study was successful in resolving some of the inconsistencies concerning factors that influence the reproductive decision and its process but, more importantly, it has paved the way for a prospective multivariate study. It would involve interviewing couples just before and shortly after genetic counseling and again 6 months later. The flow-chart model designed in the retrospective study could be tested on its merits to predict the reproductive decision. This would substantiate the importance of the factors incorporated in the model. This knowledge will be very helpful for the genetic counselor who will then be able to focus on these factors during counseling.

Another prospective study might serve to predict which couples will experience problems during the decision-making process. Based on the retrospective study, we designed a questionnaire to determine the predictive factors. The factors found in the present study are rather global. The future study should try to identify more specific factors. The designed questionnaire also explores whether all couples that experience problems desire additional support. From a psychological perspective it is important to understand the resistance against psychological support after genetic counseling. Obviously, couples that do not fall into the "vulnerable" category may still want extra support.

This retrospective study provided an outline of the factors associated with the inability to make a decision. More insight is needed regarding the dilemma confronting couples that remain undecided, including their conscious and unconscious, ambivalent feelings. An in-depth interview of these couples might provide such insight. The in-

depth interview should also explore whether these couples have problems in other areas of decision-making.

Future studies concerning low attendance of and referral for genetic counseling

Another avenue to explore would concern reasons why couples do not request genetic counseling even though this would be indicated to them. In the Netherlands, the number of requests for genetic counseling have stabilized at approximately 2500 per year (Galjaard, 1988). The Dutch Health Council has estimated that 15,000 of all couples annually contemplating a pregnancy, are eligible for genetic counseling (Galjaard, 1990). This includes questions about genetics put to the family doctor or medical specialists. One would expect that with the improved genetic knowledge of risks for future offspring the number of requests for genetic counseling at a clinical genetics center would increase. However, this is not the case. Very little is known about the reasons for the discrepancy between the number of couples eligible for genetic counseling and those who seek it. Obviously, there are couples who do not desire genetic counseling because they will accept any risk for a future child.

A study carried by our Department around 1981 showed a 20% nationwide utilization of prenatal diagnosis in women of 38 years and over. A much higher utilization (80-90%) occurred in the patients of a University Hospital where information on prenatal diagnosis was routinely provided (Thomassen-Brepols et al., 1982). Recently the nationwide utilization of women of 36 years and over rose to approximately 40% (Personal communication Dr. H. Brandenburg). Eurocat registration of congenital malformations showed that in 17-37% of the parents of children with a congenital malformation (due to chromosomal abnormality, multifactorial or mendelian cause), the risk factor had not been identified prior to the conception of these children (ten Kate, 1989). Thus, medical knowledge and the transfer of information by physicians remain important factors in view of low attendance of and referral for genetic counseling.

Fear of social consequences of genetic counseling seem to be a factor which strongly influences its utilization. An interview concerning the attitude towards utilization of genetic counseling with people randomly selected from the general Dutch population demonstrated that 85% would request genetic counseling when this would be indicated to them (Interview KRO, 1989). This figure dropped to 36% when the outcome of genetic counseling could lead to a high chance of being excluded from a job. In contrast, neither educational level, religious affiliation or personal acquaintance with a genetic disorder seemed to influence the utilization of genetic counseling. However, this survey has some methodological shortcomings. It is unknown whether the interviewees understood what genetic counseling entailed or whether they would actually do what they have said if they

would become eligible for genetic counseling.

From a psychological perspective the discrepancy in the number of couples eligible and seeking genetic counseling may be due to resistance against genetic counseling on the part of the patients or the physicians (Frets et al., 1988).

Low attendance of genetic counseling may be due to a resistance against knowing the risk of having an affected child. Couples resisting referral may desire a pregnancy, but it may seem psychologically advantageous not to know the risk of having an affected child. That might relieve the couple of any responsibility for their decision to undertake a pregnancy and from feelings of guilt when the child turned out to be affected. Alternatively, couples may resist genetic counseling because confrontation with the hereditary disorder may negatively affect their self-esteem. The helplessness and hopelessness due to the absence of a cure for a disorder may make a genetic problem extra burdensome.

Resistance in physicians to refer for genetic counseling may be caused by a lack of knowledge or because they want to safeguard their patients against guilt feelings and lowered self-esteem evoked by a genetic disorder (Frets et al., 1988). Physicians may consider that these feelings are too difficult to cope with for their patients. Furthermore, evoking these uneasy feelings in patients may confront the physician with his or her own (unbearable) feeling of powerlessness. These and other factors might be subject for a study comparing couples with an affected child that seek genetic counseling with those that do not. The reason for not seeking counseling might be due to a lack of referral or a difference in personality structure, e.g. internal or external locus of control: either believing that one's course of life is regulated by influences which are under personal control or the opposite. Alternatively, the reasons might be totally unrelated to their personality structure. Moreover, the study also needs to explore the reasons why physicians do not refer patients who are eligible for genetic counseling to the geneticist.

Future studies in respect to the impact of future genetic technological developments

Due to rapid technological advancement, particularly concerning DNA analysis, more people will be confronted with the option of presymptomatic diagnosis of genetic disorders with late onset. Furthermore, in common complex disorders, such as cancer and arteriosclerosis, more will become known about the interaction of genetics and environment; this may lead to the identification of genetic risk factors for the effects of habits or life-styles. In both fields we will have to learn about motivation to utilize this knowledge, and the need for support and understanding when people are confronted with such knowledge. Areas for future research in respect to the psychological aspects of

genetic counseling include: the social consequences of genetic counseling, the possibility of presymptomatic testing, and the integration of genetic, ethical, legal, social, and psychological aspects of genetic counseling. This discussion provides only a global perspective on future studies in this rapidly developing field.

The first area of research concerns the social consequences of genetic counseling: the extent to which genetic data influence individual eligibility for a job or a life insurance. The privacy of the individual may be threatened when insurance companies are allowed to request any information about genetic status (Galjaard, 1988). Others fear enforcement of risk reducing life-styles for those at risk for disorders such as cancer. These social consequences may become a reason for couples not to seek or not to complete genetic counseling when there is a clear indication, e.g. when the couple has an affected child (Interview KRO, 1989; Report Dutch Health Council, 1989). The uncertainty about the genetic risk remains and might lead to problems in the decision-making process.

Screening for the carrier status of autosomal recessive diseases, such as Tay Sachs disease, or the hemoglobinopathies, is actually addressed at specific populations. The introduction of carrier screening for cystic fibrosis would lead to large scale testing. If so, information strategies will have to be developed carefully. Carrier screening for an autosomal recessive disorder provides the opportunity for prospective parents to find out whether they are both carriers and subsequently at risk for an affected child. If so, they can decide to refrain from childbearing, accept the risk, and/or undergo prenatal diagnosis. Thalassemia major, an autosomal recessive severe hemoglobinopathy, has a high prevalence on the islands of Sardinia and Cyprus as well as in regions of Greece (one in seven is carrier). The results of carrier screening and prenatal diagnosis are impressive. On the island of Sardinia, this program resulted in a decline in thalassemia major births of 90% (Cao et al., 1989). However, learning that one is a healthy carrier of an autosomal recessive disorder or a balanced chromosomal translocation, etc. might also evoke fear of carrier stigmatization, which also might evoke resistance against genetic counseling (Keenen, 1978; Kaback, 1982; Wilfond and Fost, 1990). Such carrier stigmatization can affect a person's self-esteem or can have social implications when it results in discrimination, by denying e.g. sickle cell carriers health and life insurance (Culliton, 1972).

The second area concerns a qualitative study of counselees' psychological reactions to a presymptomatic diagnosis of a genetic disorder with a late onset. Presymptomatic testing will become available for an increasing number of disorders. A great number of questions have to be answered, e.g. will a precocious diagnosis lead to preoccupation with symptoms of the future disease? Will this inhibit normal development of personality? Are relationships with the partner, children, relatives, etc. going to be

affected by the threat of the illness? (Galjaard, 1990) Are there differences in psychological reaction to various genetic disorders with a late onset due to the high variability of these disorders, e.g. in expression, in age at onset, in course etc. Utilization of presymptomatic testing seems to be more likely when the disorder is less severe and the chance of effective treatment of the disorder is bigger (Sujanski et al., 1990). Furthermore we need to know whether specific supportive strategies are needed and if these have to be developed for different disorders.

Some workers have reported the psychological reactions to presymptomatic testing in individuals at risk for Huntington's disease (Evers-Kiebooms et al., 1987; Kessler et al., 1987; Meissen et al., 1988; Tibben et al., 1990). Planning for the future, which might include children, was the common reason for seeking the test. The decision to apply for the test may have enormous psychological and social consequences for the test-candidates, their partners, children and other relatives. All these parties need attention before, during and after the presymptomatic test procedure (Tibben et al., 1990).

The third area concerns the genetic, ethical, legal, social, and psychological aspects of genetic counseling. These aspects have usually been investigated separately. These aspects need to be assessed jointly for a specific population, such as those at risk for Huntington's disease, Myotonic Dystrophy, etc. This assessment should include any interrelationship of these factors. Such investigation would provide a more integrated view of what genetic counseling means for the counselees and what counselees may expect from society in answer to their problems.

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SUMMARY

Genetic and congenital disorders are now the prime cause of infant mortality and morbidity in the western industrialized countries. This is mainly due to a reduction in the incidence of infectious diseases and malnutrition brought about by economic developments, improved hygienic conditions, vaccination procedures, and the availability of antibiotics. Consequently, there is a growing need for exact diagnosis of genetic malformations as well as genetic counseling. An exact diagnosis is a major goal of genetic counseling as well as an informed reproductive decision-making process, which should always allow for personal and social considerations. To facilitate this reproductive decision-making process it is important that the genetic counselor is aware of the factors that influence the reproductive decision and its process.

A psychological follow-up study was carried out at the Erasmus University Department of Clinical Genetics and the University Hospital Dijkzigt in Rotterdam, involving interviews with 164 couples who had undergone genetic counseling 2-3 years previously. The couples were interviewed at home by a psychologist or a competent interviewer by means of either a questionnaire of 91 items specifically constructed for this study and a semi-structured in-depth interview.

The aims of this study were to monitor the transfer of information during genetic counseling and to investigate the adequacy of existing strategies for supporting counselees in their decision-making process and, if necessary, to devise new strategies. The focus was on the psychological aspects of the postcounseling decision-making process. Only those couples who had requested genetic counseling for their own offspring were eligible for the study. The objective of this study was to assess (a) which factors influenced the reproductive decision; (b) whether it was possible to identify the reproductive decision with a limited number of factors; (c) what kind of problems counselees experienced in their decision-making process (d) which factors were associated with these problems; and (e) how counselees eventually came to a reproductive decision. The results of this study were adapted for clinical application.

This is the first reported follow-up study after genetic counseling carried out in the Netherlands and as such will enable cross-cultural comparison with similar studies carried out in other countries. A review of the literature (Chapter II) summarizes *a variety of factors influencing the reproductive decision and the frequently contradictory conclusions.* The combination of the high burden of the disorder in the family and the factual genetic risk (= the risk that a future child is affected) is often seen as dominant

for the reproductive decision; others find that the interpretation of this risk, as high or low, is decisive. The availability of prenatal diagnosis influences the decision, particularly when the risk is over 15%. Our study was the first to show the influence of prenatal diagnosis on the reproductive decision, comparing couples that were eligible for prenatal diagnosis with those that were not concerning various disorders. In agreement with other follow-up studies carried out in the last decade, our results demonstrate that the desire to have children is more important for the reproductive decision than the magnitude of the genetic risk. In other words counselees tend to take high risks.

Of 164 couples in our follow-up study involving the questionnaire (Chapter III), 115 (70%) opted for having children, 28 (17%) decided not to have any (more) children, and 18 (11%) were still undecided. Two couples (1%) opted for artificial insemination by donor and one (1%) decided to wait for prenatal diagnosis to become available for them in the near future. Analysis of single factors or various combinations of two of these factors revealed *the desire to have children and the absence of personal experience with the disorder (a distant relative being affected) as dominant factors for the reproductive decision.*

The magnitude of the genetic risk was of relative importance only. Seventy per cent of the couples with a high genetic risk (>15%) opted for having children. When the disorder was perceived as severe and the risk was interpreted as high, 72% opted for having children. The availability of prenatal diagnosis only became important when there was a high genetic risk (>15%). Forty-seven per cent of couples with a high genetic risk opted for having children when prenatal diagnosis was not available. In the absence of prenatal diagnosis, couples who already had an affected child were more hesitant about having a(onther) child than those who did not - 50% decided to abstain versus 14% of the latter group.

To assess the *identifiability of reproductive planning* after genetic counseling, a model was designed to study relevant factors influencing reproductive decisions after genetic counseling (Chapter IV). A flow chart was developed whereby the dominant factors, totalling eight, were reduced to four main groups (a) *reproductive history;* (b) *desire to have children;* (c) *interpretation of risk;* (d) *significance of the availability of prenatal diagnosis for the individual couple.* The model documented the strength of the desire to have children and the interpretation of information provided at genetic counseling. The reproductive decision was identified correctly in 91% of the cases. This model may prove helpful in clinical practice. When a couple has difficulty reaching a decision, the flow-chart would determine which issues are important or

relevant for that particular couple.

Analysis of the *problems experienced in the reproductive decision-making process revealed that 43% had experienced such problems* (Chapter V). These couples had (a) experienced the decision-making process as difficult, (b) suffered doubts about the decision once made, or (c) been unable to make a decision. Logistic regression analysis revealed that the following factors were independently and significantly associated with problems experienced in the decision-making process: (1) *no postcounseling relief;* (2) *anticipation of a high risk level;* (3) *relatives' disapproval of the decision;* (4) *deciding not to have a(nother) child; and* (5) *presence of an affected child.* Interestingly, 45% of couples eligible for prenatal diagnosis who decided to have children experienced the decision-making process as difficult against 23% of couples deciding to have children while prenatal diagnosis was not available ($p < .05$). This contradicts the notion that prenatal diagnosis provides the easy way out for couples with an increased risk of having an affected child. Problems in the decision-making process might only become apparent after genetic counseling rather than in the course of it. Therefore, we suggest a structured follow-up 3-6 months after genetic counseling, to identify couples in need of additional, supportive counseling.

An *in-depth,* recorded *interview* of 30 couples randomly selected from the study population of 164 couples explored the characteristics of the postcounseling decision-making process, including the role of guilt feelings in this process (Chapter VI). This part of the study concerned couples with an affected child, sibling or spouse. Results were evaluated by two to four judges. In contrast to other studies, *the decision-making process generally appeared unstructured,* whereby guilt feelings played a significant role in more than half the couples. Guilt feelings were more predominant in couples with an affected sibling than in those with an affected spouse ($p < .05$). Lack of structure did not seem to complicate the decision-making process. Therefore, forcing counselees to structure their decision-making process, as proposed by those who favor the theory of decision analysis, is not advocated. It seems better for genetic counselors to focus on understanding counselees' feelings concerning the reproductive decision. Acceptance of apparently irrational considerations is particularly important, because such considerations represent the influence of unconscious motives. These unconscious motives may be in conflict with each other or with counselees' conscious ideas. Another important aspect of supporting counselees in their decision-making process is to understand the role played by guilt feelings towards parents or an affected sibling.

Monitoring the *transfer of information* during genetic counseling showed that the *proportion of counselees correctly recalling this information was high* and that counselees were *satisfied with the amount of information given during genetic counseling and the way genetic counseling was provided* (Chapter VII).

A *prospective study* is envisaged which would focus on testing the stability of abovementioned model, investigating its potential for predicting the reproductive decision (Chapter VII). Structuring the follow-up by means of a newly developed questionnaire might serve to predict during genetic counseling which couples will develop problems in the decision-making process.

In the future, people's attitude towards genetic counseling could be assessed in relation to the social consequences of genetic counseling.

New methods of DNA analysis will enable presymptomatic testing for an increasing number of genetic disorders with a late onset. Therefore, it is important to assess the psychological reaction to this kind of testing. The results may indicate how best to support these counselees.

The genetical, ethical, legal, social and psychological aspects of genetic counseling have been assessed separately up till now. Future studies are needed to integrate these aspects and should include any interrelationship of these factors.

SAMENVATTING

Erfelijke en aangeboren afwijkingen zijn thans de voornaamste oorzaak van kinderziekte-en sterfte in de westerse geïndustrialiseerde landen. Dit wordt met name veroorzaakt door de afname van infectieziekten en voedseltekorten ten gevolge van de economische ontwikkelingen, verbeteringen in de hygiënische omstandigheden, inentingsprogramma's en de beschikbaarheid van antibiotica. Derhalve is er een toenemende behoefte aan een exacte diagnose van genetische aandoeningen en aan erfelijkheidsadvies. Eén van de hoofddoelen van erfelijkheidsadvies is het stellen van de precieze diagnose, maar ook het voor de adviesvragers vergemakkelijken van een geïnformeerd beslissingsproces over het al dan niet krijgen van kinderen. Hierbij dient altijd ruimte te blijven voor persoonlijke en sociale overwegingen. Om hulp te bieden bij het beslissingsproces is het belangrijk dat de klinisch geneticus zich bewust is van de factoren die de beslissing en het beslissingsproces beïnvloeden.

Bij 164 echtparen die 2-3 jaar tevoren erfelijkheidsadvies gekregen hadden op de Afdeling Klinische Genetica, Erasmus Universiteit en Academisch Ziekenhuis, Rotterdam deden wij een psychologisch follow-up onderzoek. De echtparen werden thuis geïnterviewd door een psycholoog of een daartoe opgeleide interviewster, waarbij gebruik werd gemaakt van een speciaal voor dit onderzoek ontwikkelde vragenlijst met 91 vragen alsmede een semi-gestructureerd diepte-interview.

Het doel van dit onderzoek was om de informatie-overdracht van het erfelijkheidsadvies na te gaan en om te onderzoeken of de bestaande wijze van ondersteuning door de klinische geneticus bij het beslissingsproces voldoende was. Bovendien werd nagegaan of het nodig was om de manier van ondersteuning aan te passen. In dit onderzoek werd de nadruk gelegd op de psychologische aspecten van het beslissingsproces na erfelijkheidsadvies. Alleen die adviesvragers werden opgenomen die erfelijkheidsadvies hadden gevraagd voor toekomstig nageslacht. De specifieke vragen van het onderzoek waren (a) welke factoren de beslissing over het al dan niet krijgen van kinderen na erfelijkheidsadvies beïnvloedden; (b) of het mogelijk was om de beslissing over het al dan niet krijgen van kinderen te identificeren met een beperkt aantal factoren; (c) wat voor soort problemen de adviesvragers ervoeren in hun beslissingsproces; (d) welke factoren geassocieerd waren met deze problemen; en (e) hoe de adviesvragers tot hun beslissing over het al dan niet krijgen van kinderen waren gekomen. Voorts werd aangegeven hoe de resultaten van dit onderzoek toegepast kunnen worden in de klinische praktijk.

Dit is het eerste gepubliceerde follow-up onderzoek na erfelijkheidsadvies in Nederland en als zodanig is het mogelijk om een cross-culturele vergelijking te maken met soortgelijke studies uitgevoerd in andere landen. In een overzicht van de literatuur (Hoofdstuk II) wordt een *verscheidenheid van factoren samengevat welke de beslissing over het al dan niet krijgen van kinderen na erfelijkheidsadvies beïnvloeden met nogal eens tegenstrijdige conclusies*. De combinatie van de belasting van de aandoening in de familie en het feitelijke genetische risico, dat wil zeggen de kans dat een toekomstig kind aangedaan is, wordt vaak gezien als zeer belangrijk voor de beslissing over het al dan niet krijgen van kinderen; anderen vinden dat de interpretatie van het risico, als hoog of laag, beslissend is. De mogelijkheid van prenatale diagnostiek beïnvloedt de beslissing met name als het risico groter is dan 15%. In dit onderzoek tonen wij als eerste de invloed aan van prenatale diagnostiek op de beslissing omtrent nageslacht bij echtparen met een risico op verschillende aandoeningen. Wij vergeleken echtparen die wel en die niet voor prenatale diagnostiek in aanmerking kwamen. In overeenstemming met andere follow-up onderzoeken van de laatste 10 jaar komt ook uit ons onderzoek naar voren dat de wens om kinderen te krijgen belangrijker is voor de beslissing omtrent nageslacht na erfelijkheidsadvies dan de hoogte van het genetische risico. Met andere woorden, adviesvragers hebben de neiging hoge risico's te nemen.

Van de 164 echtparen in ons onderzoek, waarbij de vragenlijst werd gebruikt (Hoofdstuk III), kozen 115 (70%) voor (verder) nageslacht, 28 (17%) zagen van het krijgen van (meer) kinderen af en 18 (11%) hadden nog geen beslissing genomen. Twee echtparen (1%) kozen voor kunstmatige inseminatie met donor sperma en één (1%) wilde wachten tot prenatale diagnostiek voor hen beschikbaar zou komen. Wanneer wij de factoren die van belang zijn voor de beslissing over het al dan niet krijgen van kinderen na erfelijkheidsadvies enkel of in combinatie van 2 analyseerden kwamen 2 factoren duidelijk naar voren als zijnde heel belangrijk, namelijk *de wens om kinderen te krijgen en het niet bekend zijn met de aandoening, bijvoorbeeld wanneer een ver familielid is aangedaan.*

De grootte van het genetische risico was slechts van relatief gering belang voor de beslissing. Zeventig procent van de echtparen met een hoog genetisch risico (> 15%) koos voor (verder) nageslacht. Van de echtparen die de aandoening als ernstig ervoeren en het risico hoog vonden, koos 72% voor een (volgende) zwangerschap. De mogelijkheid van prenatale diagnostiek werd alleen belangrijk in combinatie met een hoog genetisch risico (> 15%). Zevenenveertig procent koos voor (verder) nageslacht als prenatale diagnostiek voor hen niet mogelijk was. Indien prenatale diagnostiek niet

mogelijk was waren de echtparen die reeds een aangedaan kind hadden veel terughoudender om het nog een keer te proberen dan degenen die geen aangedaan kind hadden - 50% zag af van verder nageslacht in tegenstelling tot 14% van de laatste groep.

Om de *identificeerbaarheid van de beslissing over het al dan niet krijgen van kinderen na erfelijkheidsadvies* te onderzoeken, hebben wij een model ontworpen om de relevante factoren te bestuderen welke deze beslissing beïnvloeden (Hoofdstuk IV). Wij ontwikkelden een stroomdiagram waarbij de 8 belangrijkste factoren werden teruggebracht tot 4 groepen: factoren samenhangend met (a) *de obstetrische voorgeschiedenis,* (b) *de kinderwens,* (c) *de interpretatie van het risico,* (d) *de betekenis van de beschikbaarheid van prenatale diagnostiek voor het individuele echtpaar.* Het ontworpen model gaf aan dat de sterkte van de kinderwens en de interpretatie van de informatie gegeven tijdens het erfelijkheidsadvies zeer belangrijk waren voor de beslissing na erfelijkheidsadvies. Het bleek mogelijk om met het model de beslissingen van 91% van de echtparen correct te identificeren. Het model kan zinvol zijn in de klinische praktijk. Als een echtpaar moeite heeft om tot een beslissing te komen is het mogelijk om, door het volgen van het stroomdiagram, te bepalen welke aspecten belangrijk of relevant zijn voor de beslissing van een bepaald echtpaar.

Uit de analyse van de *problemen welke ervaren werden bij het nemen van de beslissing over (verder) nageslacht kwam naar voren dat 43% van de echtparen deze problemen ervaren had* (Hoofdstuk V). Deze echtparen (a) hadden het nemen van de beslissing als moeilijk ervaren, of (b) twijfelden aan de juistheid van de door hen genomen beslissing, of (c) waren nog niet in staat geweest een beslissing te nemen. Uit de logistische regressie analyse kwam naar voren dat de volgende factoren onafhankelijk en significant geassocieerd waren met het ervaren van problemen in het beslissingsproces: (1) *geen geruststelling na het erfelijkheidsadvies;* (2) *anticipatie van een hoog risico;* (3) *afkeuring van de beslissing van het echtpaar door familieleden;* (4) *de beslissing om af te zien van (verder) nageslacht; en* (5) *de aanwezigheid van een aangedaan kind.* Een interessante bevinding was dat 45% van de echtparen die in aanmerking kwamen voor prenatale diagnostiek en die besloten hadden om kinderen te krijgen het nemen van deze beslissing moeilijk hadden gevonden, in vergelijking met 23% van de echtparen die kozen voor het krijgen van kinderen terwijl prenatale diagnostiek voor hen niet mogelijk was ($p < 0.05$). Deze bevinding is in tegenspraak met de opvatting dat prenatale diagnostiek een makkelijke uitweg is voor echtparen met een verhoogd risico op een kind met een aandoening. Soms kunnen problemen, ervaren in het beslissingsproces, pas duidelijk worden na, in plaats van

tijdens, het erfelijkheidsadvies. Daarom raden wij een gestructureerde follow-up aan, 3-6 maanden na erfelijkheidsadvies, om zo echtparen te kunnen identificeren die aanvullende ondersteunende begeleiding nodig hebben.

In een *diepte-interview,* vastgelegd op geluidsband, bij 30 echtparen, willekeurig geselecteerd uit de studie populatie van 164 echtparen, werden de kenmerken van het beslissingsproces na erfelijkheidsadvies nagegaan met speciale aandacht voor de rol van schuldgevoelens in dit proces (Hoofdstuk VI). Dit deel van het onderzoek richtte zich op echtparen met een aangedaan kind, broer/zuster of partner. De resultaten werden geëvalueerd door 2 of 4 beoordelaars. In tegenstelling tot andere onderzoekers, vonden wij een over het algemeen *ongestructureerd beslissingsproces.* Bij meer dan de helft van de echtparen speelden schuldgevoelens een belangrijke rol. Schuldgevoelens kwamen vaker voor bij echtparen met een aangedane broer of zuster dan bij de echtparen bij wie één van de partners was aangedaan ($p < 0.05$). Gebrek aan structuur leek het beslissingsproces niet te compliceren. Daarom lijkt het niet zinvol om adviesvragers te forceren hun beslissingsproces te structureren zoals wordt voorgesteld door de aanhangers van de beslissingsanalyse-theorie. Het lijkt beter dat klinisch genetici zich richten op begrip voor gevoelens van de adviesvragers ten aanzien van de beslissing over het al dan niet krijgen van kinderen. Acceptatie van schijnbare irrationele overwegingen is met name belangrijk, omdat deze gevoelens kunnen wijzen op de invloed van onbewuste motieven. Deze onbewuste motieven kunnen in conflict zijn met elkaar of met de bewuste ideeën van de adviesvragers. Een ander belangrijk aspect bij het ondersteunen van adviesvragers in hun beslissingsproces is oog te hebben voor de rol welke schuldgevoelens ten opzichte van ouders of een aangedane broer/zuster kunnen spelen.

Onderzoek naar de informatieoverdracht van het erfelijkheidsadvies liet zien dat een *groot deel van de adviesvragers deze informatie correct onthouden had* en dat zij *tevreden waren met de hoeveelheid informatie en de manier waarop deze gegeven werd tijdens het erfelijkheidsadvies* (Hoofdstuk VII).

Een *toekomstig prospectief onderzoek* zou zich kunnen richten op het testen van de stabiliteit van het eerder genoemde model, waarbij de mogelijkheid van het voorspellen van de beslissing onderzocht kan worden (Hoofdstuk VII). Structureren van de follow-up met behulp van een nieuw ontwikkelde vragenlijst zou ervoor kunnen dienen om tijdens het erfelijkheidsadvies te kunnen voorspellen welke echtparen problemen zullen gaan krijgen in hun beslissingsproces.

In de toekomst kan de houding van mensen ten opzichte van erfelijkheidsadvies

onderzocht worden in relatie tot de maatschappelijke gevolgen van erfelijkheidsadvies.

Nieuwe methoden van DNA analyse zal presymptomatische diagnostiek mogelijk maken voor een toenemend aantal later in het leven optredende erfelijke aandoeningen. Daarom is het belangrijk om de psychologische reacties op dit soort diagnostiek te onderzoeken. De resultaten kunnen een indicatie geven hoe deze adviesvragers het beste ondersteund kunnen worden.

Tot nu toe zijn de genetische, juridische, maatschappelijke en psychologische aspecten van erfelijkheidsadvies steeds apart bestudeerd. In de toekomst is onderzoek nodig waarin deze aspecten worden geïntegreerd waarbij nagegaan kan worden of en hoe deze aspecten onderling gerelateerd zijn.

APPENDIX I. QUESTIONNAIRE

INSTRUCTIONS

I would like to ask you some questions about the genetic counseling you received at the Department of Clinical Genetics in Rotterdam.

There are several answers for each question. Please check the answer that applies.

Some questions allow for more than one answer; this will be clearly indicated.

When questions mention an affected child, this means that the child is affected by the disorder occurring in your family.

All information will be treated as confidential and kept secret; the Hyppocratic oath guarantees secrecy.

Date interview ..... Patient no. 84... Interviewer ...

---

1. What is your date of birth ?
   man .....
   woman .....

2. What is your marital status ?

   ☐ single
   ☐ cohabiting, since .....
   ☐ married, since .....

3. What is your religious affiliation ?

man	woman	
☐	☐	Reformed (Protestant)
☐	☐	Roman Catholic
☐	☐	Dutch Reformed (Orthodox Protestant)
☐	☐	Other (Please explain).....
☐	☐	None

4. What schooling did you complete ? (Please check all answers that apply)

man	woman	
☐	☐	Elementary school
☐	☐	Lower vocational school (home economics, lower technical school)
☐	☐	Secondary school (O-levels, A-levels)
☐	☐	Secondary vocational school (technical, practical, nursing, economics)
☐	☐	Higher vocational education (Polytechnic)
☐	☐	University or College

5. What is/was your profession ?

man .....
woman .....

6. Are/were you self-employed ?

man	woman	
☐	☐	Yes
☐	☐	No

7. If yes (ref. question 6) do/did you have any employees ?

man	woman	
☐	☐	Yes
☐	☐	No

8. If yes (ref. question 7) how many employees do/did you have ?

man	woman	
☐	☐	1-25
☐	☐	more than 26

9. If no (ref. question 6) do/did you preside over a department or division ?

man	woman	
☐	☐	Yes
☐	☐	No

10. How many children of your own do/did you have ?

Number ..... (of whom ..... died)
Number of miscarriages .....

11. What is the date of birth of your youngest child ?

.....

12. When did you last visit the Department of Clinical Genetics in Rotterdam?

man ..... month.....year
woman ..... month.....year

13. How many times did you come for consultation ?

man ..... times
woman ..... times

14. Were you/was your partner pregnant when you consulted the Department of Clinical Genetics for the first time ?

☐ Yes
☐ No

15. If no, did you/your partner use the pill/IUD or any other birth-control device ?

☐ Yes
☐ No

16. If yes (ref. question 14), was it a planned pregnancy ?

☐ Yes
☐ No

17. Did you undertake a(nother) pregnancy after your last visit to the Department of Clinical Genetics ?

☐ Yes
☐ No

18. If yes, how many times ?

..... times.

THE NEXT QUESTIONS (19-28) APPLY TO THE TIME *BEFORE* YOU CAME FOR GENETIC COUNSELING

19. Concerning which disorder/disease(s)/handicap(s) did you seek counseling and which member(s) of your family was/were affected ? (just a description is enough)

disorder ..... in .....
disorder ..... in .....
disorder ..... in .....

20. Before coming for genetic counseling, did you think your risk of having an affected/handicapped child was high ?

man	woman	
☐	☐	Yes
☐	☐	No
☐	☐	I didn't know

21. Did this worry you ?

man	woman	
☐	☐	Yes, why?.....
☐	☐	No .....

22. Where did you hear about the Department of Clinical Genetics in Rotterdam ? (Please check all answers that apply).

man	woman	
☐	☐	through an advertisement
☐	☐	from television, radio
☐	☐	from patient support group
☐	☐	from our general practitioner
☐	☐	from our medical specialist
☐	☐	from friends
☐	☐	other (Please explain).....

23. When you decided to seek genetic counseling, how long did it take for you to contact the Department of Clinical Genetics ?

man	woman	
☐	☐	Immediately through, e.g., specialist/g.p./midwife
☐	☐	After ..... weeks

24. What did you feel about going to the Department of Clinical Genetics ?

man	woman	
☐	☐	I dreaded it
☐	☐	I was a little anxious
☐	☐	I didn't mind

25. If you dreaded it, why did you ?

man: .....
.....

woman: .....
.....

26. Do you feel you could have gone sooner ?

man	woman	
☐	☐	Yes
☐	☐	No
☐	☐	don't know

27. If yes, why didn't you come sooner ?

man: .....
.....
woman: .....
.....

28. Who supported your visit to the Department of Clinical Genetics and who did not ? (Please check the appropriate box for each person)

	did support		did not support		no communication about this	
	man	woman	man	woman	man	woman
partner	☐	☐	☐	☐	☐	☐
g.p.	☐	☐	☐	☐	☐	☐
specialist	☐	☐	☐	☐	☐	☐
relatives	☐	☐	☐	☐	☐	☐
friends	☐	☐	☐	☐	☐	☐
priest/minister	☐	☐	☐	☐	☐	☐
other:.....	☐	☐	☐	☐	☐	☐

THE NEXT QUESTIONS (29-40) APPLY TO THE TIME AFTER YOU CAME FOR GENETIC COUNSELING.

29. Have you had a(nother) child(ren) after your last visit to the Department of Clinical Genetics ?

☐ Yes
☐ No

30. If yes how many children were born and what was the date of birth ?

..... children
birthdate(s) .....
.....

31. Is this/are these child(ren) healthy ?

☐ Yes
☐ No

32. If no, what is the matter with this/these child(ren) ?

.....
.....

33. How big would you say the risk is for any set of parents to have a child with a mental and/or physical handicap ?

man ..... %
woman ..... %

34. If the two of you had a(nother) child, what would you say the risk is for the child to be affected with the disorder that runs in your family ?

man ..... %
woman ..... %

35. Was the risk level an estimate, or was it possible for the clinical geneticist to determine the exact figure ?

man	woman	
☐	☐	It was an estimate
☐	☐	Exact figure could be determined
☐	☐	I don't know

36. Do you think this is a big risk ?

man	woman	
☐	☐	Yes
☐	☐	No

37. Do you feel that the disorder that concerns you (your family) is a serious one ?

man	woman	
☐	☐	Yes, very serious
☐	☐	Rather serious
☐	☐	Not very serious
☐	☐	No, absolutely not

38. Are you eligible for chorionic villus sampling/amniocentesis/ultra-sound should your you/your partner become pregnant ?

man	woman	
☐	☐	Yes
☐	☐	No
☐	☐	I don't know

39. Would the risk of occurrence/recurrence be smaller with artificial insemination by donor ?

man	woman	
☐	☐	Yes
☐	☐	No
☐	☐	I don't know

40. Can you tell where you got this information (ref. questions 33-35,38, 39) ? (Please check all answers that apply)

man	woman	
☐	☐	from genetic counseling
☐	☐	from an advertisement
☐	☐	from television/radio programme
☐	☐	from a magazine
☐	☐	other (Please explain).....
☐	☐	I don't know

41. Was any co-operation required from your family for the clinical geneticist to determine your risk level ?

man	woman	
☐	☐	Yes
☐	☐	No

42. If yes, did that cause problems ?

man	woman	
☐	☐	Yes
☐	☐	No

43. What kind of co-operation was required ?
(Please check all answers that apply)

man	woman	
☐	☐	permission for divulging information
☐	☐	permission for blood tests
☐	☐	family members or relatives had to travel to Rotterdam or somewhere else for tests
☐	☐	participation in decision-making
☐	☐	other (please explain).....

44. How did you feel after your last visit to the Department of Clinical Genetics ?

man	woman	
☐	☐	I felt relieved about our risk of having an affected child
☐	☐	I was a little anxious
☐	☐	I was very anxious
☐	☐	Other (Please explain).....
		.....

45. Why was that ?

man .....

woman .....

46. What did you think of what the clinical geneticist told you ? (Please check all answers that apply)

man	woman	
☐	☐	I thought it was clear
☐	☐	I didn't think it was clear
☐	☐	I thought it was too much at once
☐	☐	I thought it was sufficient
☐	☐	I thought it was insufficient
☐	☐	Other (Please explain).....

Additional comments .....

47. Did you understand what you were told ?

man	woman	
☐	☐	Yes
☐	☐	A little
☐	☐	No

48. If no, why didn't you ?

man .....

woman .....

49. What did you think of the letter that summarized the consultation ? (Please check all answers that apply)

man	woman	
☐	☐	Clear
☐	☐	Not clear
☐	☐	Too long
☐	☐	The right length
☐	☐	Too short
☐	☐	Superfluous
☐	☐	Useful
☐	☐	I did not read the letter
☐	☐	I did not receive the letter
☐	☐	I don't know
☐	☐	Other (Please explain).....

Additional comments .....

50. Did the clinical geneticist give you a copy of the booklet "Disease and Genetics" ?

man	woman	
☐	☐	Yes
☐	☐	No

51. If yes, what did you think of it ?
(Please check all answers that apply)

man	woman	
☐	☐	Clear
☐	☐	Not clear
☐	☐	Too much information
☐	☐	Sufficient information
☐	☐	Insufficient information
☐	☐	Superfluous
☐	☐	Useful
☐	☐	I did not read it
☐	☐	I don't know
☐	☐	Other (Please explain).....

Additional comments .....

IF THE COUPLE HAS DECIDED WHETHER OR NOT TO HAVE (MORE) CHILDREN, PROCEED WITH THE NEXT QUESTIONS

IF THE COUPLE HAS NOT MADE A DECISION, GO ON TO QUESTION # 70

52. What did you finally decide after hearing how high the risk was for the two of you to have an affected/handicapped child?

☐ We decided to have a(nother) child
☐ We decided not to have a(nother) child
☐ Other (Please explain).....

53. Could you explain why you decided this ?

man .....
woman .....

54. How long after the last consultation did it take for you to make a definitive decision?

man	.....	weeks	.....	months	.....	years
woman	.....	weeks	.....	months	.....	years

55. Did it take a long time to reach a decision ?

man	woman	
☐	☐	Yes
☐	☐	No

56. Was it a difficult decision ?

man	woman	
☐	☐	Yes
☐	☐	No

57. Did the two of you talk about the decision ?

man	woman	
☐	☐	Yes, a lot
☐	☐	Rather a lot
☐	☐	Not so much
☐	☐	Hardly
☐	☐	Other (Please explain).....

Additional comments .....

58. Who talked about it most ?

man	woman	
☐	☐	Man
☐	☐	Woman
☐	☐	No difference

59. Did you think your partner talked too much or too little ?

man	woman	
☐	☐	Much too much
☐	☐	A bit too much
☐	☐	Just right
☐	☐	Not quite enough

60. Do you ever think that the decision would have been easier to make if you had not had genetic counseling ?

man	woman	
☐	☐	Yes
☐	☐	No

61. Who does/did not support your decision ?

	does support		does not support		no communication about this	
	man	woman	man	woman	man	woman
partner	☐	☐	☐	☐	☐	☐
g.p.	☐	☐	☐	☐	☐	☐
specialist	☐	☐	☐	☐	☐	☐
relatives	☐	☐	☐	☐	☐	☐
friends	☐	☐	☐	☐	☐	☐
priest/minister	☐	☐	☐	☐	☐	☐
other:.....	☐	☐	☐	☐	☐	☐

62. Did the opinion of others influence your decision for or against having (further) children ?

man	woman	
☐	☐	Yes
☐	☐	A little
☐	☐	No

63. If yes, who were these others ?

man .....

woman .....

64. Was the information supplied by the clinical geneticist important for your decision ?

man	woman	
☐	☐	Yes, very important
☐	☐	Not so important
☐	☐	No, not important at all
☐	☐	Other (Please explain).....

65. Would you have made another decision if you had not had genetic counseling ?

man	woman	
☐	☐	Yes
☐	☐	No

66. Does your decision ever bother you ?

man	woman	
☐	☐	Yes
☐	☐	No

67. Have you been wondering recently whether you made the right decision ?

man	woman	
☐	☐	Yes
☐	☐	No

68. Are you both satisfied with your decision to have or not to have a(nother) child ?

man	woman	
☐	☐	Yes
☐	☐	No

69. If no, why not ?

man .....

woman .....

THE FOLLOWING QUESTIONS (70-74) SHOULD ONLY BE PUT TO THOSE COUNSELEES WHO HAVE NOT YET MADE A DECISION WHETHER OR NOT TO HAVE A(NOTHER) CHILD.

FOR ALL OTHERS PROCEED TO QUESTION 75.

70. What do you find the most difficult aspect of having to decide ?

man .....
woman .....

71. Did genetic counseling raise more doubts about your reproductive decision ?

man	woman	
☐	☐	Yes
☐	☐	No

72. If yes, can you indicate why ?

man .....
woman .....

73. Do you often talk about it together ?

man	woman	
☐	☐	Often
☐	☐	Sometimes
☐	☐	Hardly ever
☐	☐	Other (Please explain) .....

74. If you don't discuss it why don't you ?

man .....
woman .....

FROM HERE ON THE QUESTIONS ONCE AGAIN APPLY TO EVERYONE

75. How do you feel about another pregnancy ?

man	woman	
☐	☐	I dread the thought
☐	☐	I am a little anxious about it
☐	☐	I am not very anxious about it
☐	☐	It does not worry me at all

76. Are you personally acquainted with the disorder or handicap that runs in your family? (Please check all answers that apply)

man	woman	
☐	☐	from my own family
☐	☐	from relatives
☐	☐	from immediate surroundings
☐	☐	professionally
☐	☐	not personally acquainted at all
☐	☐	other (Please explain) .....

77. Did genetic counseling reveal a certain risk for you personally ?

man	woman	
☐	☐	Yes
☐	☐	No
☐	☐	I don't know

78. If yes, was that totally unexpected ?

man	woman	
☐	☐	Yes
☐	☐	No

79. Did you consider beforehand whether you would want to know the risk ?

man	woman	
☐	☐	Yes
☐	☐	No

80. Under what condition would you resort to abortion ?
(Please check all answers that apply)

man	woman	
☐	☐	if the child's life is in danger
☐	☐	if the mother's life is in danger
☐	☐	if the pregnancy is unwanted
☐	☐	if the child has a serious congenital defect
☐	☐	other (Please explain).....

81. Did the clinical geneticist take enough time for you ?

man	woman	
☐	☐	Yes
☐	☐	No

82. In retrospect, would you have wanted additional genetic counseling ?

man	woman	
☐	☐	Yes
☐	☐	No

83. If yes, what would you have wanted to know ?

man .....
woman .....

84. Do you talk about the genetical aspects of the disorder in your family more often with your children or relatives since you had genetic counseling ?

man	woman	
☐	☐	Yes, I talk about it more often
☐	☐	I talk about it the same as before
☐	☐	No, I talk about it less than before
☐	☐	I don't talk about it
☐	☐	No
☐	☐	other (Please explain).....

85. If friends of yours were in the same situation, would you recommend genetic counseling ?

man	woman	
☐	☐	Yes
☐	☐	No
☐	☐	I don't know

86. Do you feel that genetic counseling was of much help ?

man	woman	
☐	☐	Yes
☐	☐	No
☐	☐	I don't know

87. If yes, in what way ?

man .....

woman .....

88. If you were the clinical geneticist would you have conducted the counseling in the same way ?

man	woman	
☐	☐	I would have done it the same way
☐	☐	I would have done it differently (Please explain).....
		.....

QUESTION 89 AND 90 ONLY APPLY TO COUNSELEES WHO ARE RELIGIOUS, ELSE PROCEED TO QUESTION 91

89. Do you generally allow your religious belief to determine your life ?

man	woman	
☐	☐	Yes, completely
☐	☐	Partly
☐	☐	No, not at all

90. Did your religious belief contribute significantly to your reproductive decision ?

man	woman	
☐	☐	Yes, completely
☐	☐	Partly
☐	☐	No, not at all

91. Do you have any comments ?

man .....
woman .....

THANK YOU VERY MUCH FOR YOUR CO-OPERATION

THE FOLLOWING QUESTIONS HAVE TO BE FILLED OUT BY THE INTERVIEWER

A. Did the counselees use the letter that summarized genetic counseling to answer the questions 33-35, 38 or 39 ?

man	woman	
☐	☐	Yes
☐	☐	No

DATA FROM THE COUPLES' FILE TO BE COMPARED WITH COUNSELEES' ANSWERS:

B. Concerning which disorder/disease(s)/handicap(s) did the couple seek genetic counseling and which member(s) of the family was/were affected ?

disorder ..... in .....
disorder ..... in .....
disorder ..... in .....

C. If the couple had a(nother) child, what is the risk for the child to be affected with the disorder that runs in their family (=genetic risk)

disorder ..... genetic risk ..... %
disorder ..... genetic risk ..... %
disorder ..... genetic risk ..... %

D. Is the couple eligible for prenatal diagnosis ?

☐ Yes
☐ No
☐ Unknown

E. Would the risk of occurrence/recurrence be smaller with artificial insemination by donor ?

- ☐ Yes
- ☐ No
- ☐ Unknown

F. Did genetic counseling reveal a certain risk for one or both counselees personally ?

- ☐ Yes
- ☐ No
- ☐ Unknown

G. If yes for which disorder ?

disorder ..... in .....
disorder ..... in .....

H. Was the genetic risk an estimate ?

- ☐ Yes
- ☐ No
- ☐ Unknown

I. Was the woman pregnant at the first consultation ?

- ☐ Yes
- ☐ No

J. If no was any kind of birth-control device used ?

- ☐ Yes
- ☐ No
- ☐ Unknown

K. How many times did the counselees come for consultation ?

man ..... times
woman ..... times

L. When did the counselees last visit the Department of Clinical Genetics in Rotterdam?

man ..... month .....year
woman ..... month .....year

APPENDIX II. IN-DEPTH INTERVIEW

IN-DEPTH INTERVIEW CONCERNING THE REPRODUCTIVE DECISION-MAKING PROCESS AFTER GENETIC COUNSELING AND FEELINGS CONCERNING THE PROBAND

The characteristics of the decision-making process

Concept assessed	Related questions
"Quality" of the decision	
Information collecting	1. Do you feel that you have collected all additional relevant information, thus apart from the genetic counseling information, such as genetic publications from libraries or other sources ?
Receptive to information	2. Do you feel you have been receptive to all this information or were there things you rather not hear such as a high risk level, uncertainty about the risk of occurrence / recurrence.
Consider all options	3. Did you consider all the options e.g. to have a child that may be affected, to refrain from having (more) children, artificial insemination by donor, prenatal diagnosis, adoption, to raise a foster child.
	4. Did you weigh all pros and cons of each of these options ?

Implementation of the decision	5. Did your choice of option include implementation the consequences of your decision, e.g. did the decision to have a(nother) child include plans for a thorough medical check-up once the child was born ?
Scenarios	
Imagining situations	6. Did you imagine the consequences of at all situations before least some of the available options, e.g. to have a child that may be affected, to refrain from having (more) children, artificial insemination by donor, prenatal diagnosis, adoption, to raise a foster child.
	7. Did you specifically imagine the consequences of having a child that may be affected ?
Influence of attitude significant others	
Reactions from outsiders	8. Was it important for you what others (e.g. parents /in-laws) would have to say about your decision ?
Burden of the process	9. Was it a very difficult decision for you decision-making to make ? Can you tell me why ?
	10. Do you ever wonder whether you made the right decision ?
Additional comments	11. Were there other things that influenced your decision ?

Feelings concerning the proband

Concept assessed	Related questions
Narcissistic attitude	12. Did you ever stop and think that a child of yours/sibling/you yourself/your partnermight have a physical or mental handicap?
	13. Do you ever feel offended by the fact that .....[1] is affected ?
	14. Do you find it hard to accept that ....is affected ?
	15. Do you ever take it personally that a disorder occurs in *your* family ?
Sense of shame	16. Do you ever feel tempted to hide the disorder of .....?
	17. Do you ever avoid the subject if others bring it up ?
	These questions apply to counselees with an affected child
	18. Do you mind taking ..... out in public/ into the street ?
	19. Do you mind taking ..... with you when you go visiting ?

Thank you very much for your co-operation

[1] During the interview the name of the proband was used in this place

APPENDIX III. GRADING LIST OF THE IN-DEPTH INTERVIEW

GRADING LIST IN-DEPTH INTERVIEW CONCERNING THE REPRODUCTIVE DECISION-MAKING PROCESS AFTER GENETIC COUNSELING AND FEELINGS CONCERNING THE PROBAND

Date of Grading: ..... Patient no. 84 ...

Judge: .....

The characteristics of the decision-making process

"Quality of the decision

1. Do the counselees feel they have collected all additional relevant information, thus apart from the genetic counseling information, such as genetic publications from libraries or other sources ?

man	not at all	*---*---*---*---*---*	to a great extent
woman	not at all	*---*---*---*---*---*	to a great extent

2. Were the counselees receptive to all this information ?

On a conscious level

man	not at all	*---*---*---*---*---*	yes completely
woman	not at all	*---*---*---*---*---*	yes completely

On an unconscious level

man	not at all	*---*---*---*---*---*	yes completely
woman	not at all	*---*---*---*---*---*	yes completely

3. Did the counselees consider all the options (e.g. to have a child that may be affected, to refrain from having (more) children, artificial insemination by donor, prenatal diagnosis, adoption, to raise a foster child) before they made their decision ?

man	not at all	*---*---*---*---*---*	yes completely
woman	not at all	*---*---*---*---*---*	yes completely

4. Did the counselees weigh all pros and cons of each of these options ?

man	not at all	*---*---*---*---*---*	yes completely
woman	not at all	*---*---*---*---*---*	yes completely

5. Did counselees' choice of option include implementation of the consequences of their decision, e.g. did the decision to have a(nother) child include plans for a thorough medical check-up once the child was born ?

man	not at all	*---*---*---*---*---*	to a great extent
woman	not at all	*---*---*---*---*---*	to a great extent

Scenarios

6. Is it your impression that the counselees imagined the consequences of at least some of the available options ?

man	not at all	*---*---*---*---*---*	yes completely
woman	not at all	*---*---*---*---*---*	yes completely

7. Is it your impression that the counselees specifically imagined the consequences of having a child that may be affected ?

man	not at all	*---*---*---*---*---*	yes completely
woman	not at all	*---*---*---*---*---*	yes completely

Reactions from significant others

8. Was it important for the counselees what significant others (e.g. parents /in-laws) would have to say about their decision ?

man	not at all	*---*---*---*---*---*	to a great extent
woman	not at all	*---*---*---*---*---*	to a great extent

Burden of the decision-making process

9. Did the counselees experience the decision-making process as difficult ?

On a conscious level

man	not at all	*---*---*---*---*---*	to a great extent
woman	not at all	*---*---*---*---*---*	to a great extent

On an unconscious level

man	not at all	*---*---*---*---*---*	to a great extent
woman	not at all	*---*---*---*---*---*	to a great extent

10. Did the reproductive decision leave any unresolved doubts in the counselees ?

On a conscious level

man	not at all	*---*---*---*---*---*	to a great extent
woman	not at all	*---*---*---*---*---*	to a great extent

On an unconscious level

man	not at all	*---*---*---*---*---*	to a great extent
woman	not at all	*---*---*---*---*---*	to a great extent

Guilt feelings

11. What is your impression that guilt feelings played a role in the decision-making process ?

On a conscious level

man	not at all	*---*---*---*---*---*	to a great extent
woman	not at all	*---*---*---*---*---*	to a great extent

On an unconscious level

man	not at all	*---*---*---*---*---*	to a great extent
woman	not at all	*---*---*---*---*---*	to a great extent

Feelings concerning the proband

Lowered self-esteem

12.Did the disorder of the proband negatively affect counselees' self-esteem ?

On a conscious level

man	not at all	*---*---*---*---*---*	to a great extent
woman	not at all	*---*---*---*---*---*	to a great extent

On an unconscious level

man	not at all	*---*---*---*---*---*	to a great extent
woman	not at all	*---*---*---*---*---*	to a great extent

Guilt feelings

13.Do counselees experience guilt feelings towards the proband ?

On a conscious level

man	not at all	*---*---*---*---*---*	to a great extent
woman	not at all	*---*---*---*---*---*	to a great extent

On an unconscious level

man	not at all	*---*---*---*---*---*	to a great extent
woman	not at all	*---*---*---*---*---*	to a great extent

Sense of shame

14.Do counselees feel tempted to hide the proband's disorder ?

On a conscious level

man	not at all	*---*---*---*---*---*	to a great extent
woman	not at all	*---*---*---*---*---*	to a great extent

On an unconscious level

man	not at all	*---*---*---*---*---*	to a great extent
woman	not at all	*---*---*---*---*---*	to a great extent

Degree of acceptance

15.To what extent have the counselees accepted the proband's disorder ?

mann accepted	not at all	*---*---*---*---*---*	completely accepted
woman accepted	not at all	*---*---*---*---*---*	completely accepted

Thank you very much for your co-operation

APPENDIX IV. LETTER TO INVITE COUPLES TO PARTICIPATE IN THE STUDY

ERASMUS UNIVERSITEIT ROTTERDAM

FACULTEIT DER
GENEESKUNDE
Afdeling Klinische Genetica

onze referentie MFN/PGF/mh uw referentie rotterdam, september 1986

doorkiesnr.463 4410

onderwerp

Zeer geachte heer en mevrouw,

In 1984 bezocht u de afdeling Klinische Genetica van het Academisch Ziekenhuis Dijkzigt te Rotterdam met de vraag naar een erfelijkheidsadvies.

Op het ogenblik onderzoeken wij de ervaringen van mensen, die erfelijkheidsinformatie en -voorlichting kregen.

Het onderzoek is bedoeld om van uw ervaringen te leren, zodat wij desnoods de erfelijkheidsvoorlichting kunnen verbeteren.
Mevr. Peters, assistente bij ons op de afdeling Klinische Genetica, zou u graag thuis bezoeken voor het afnemen van een vragenlijst bij u en uw partner. Wij zouden het op prijs stellen als u haar zou willen ontvangen. Zij belt u voor het maken van een afspraak. Overigens worden alle antwoorden geheel anoniem (dus zonder uw naam) behandeld. Ze vallen onder het medisch beroepsgeheim. De antwoorden, die u geeft worden dus niet bekend aan anderen, ook niet aan artsen.

Voor alle mogelijke vragen bent u welkom om inlichtigen te vragen bij: Mevr.Drs. P.G. Frets, tel. 010-4634410 (tussen 9.00 en 9.30 uur), die de leiding heeft over het onderzoek.

We danken u bij voorbaat voor uw medewerking en besluiten met vriendelijke groeten,

Hoogachtend,

Prof.Dr. M.F. Niermeijer

APPENDIX V: LETTER SEND TO THE COUPLES WITH THE SUMMARY OF THE RESULTS OF THE STUDY

ERASMUS UNIVERSITEIT ROTTERDAM

FACULTEIT DER
GENEESKUNDE
dr. Molewaterplein 50

Mw.Drs. P.G. Frets, psychologe
Afdeling Klinische Genetica

Uw brief

Ons kenmerk PGF/ms

Datum
februari 1988

Onderwerp follow-up onderzoek

Doorkiesnummer
408 7215

Zeer geachte heer en mevrouw,

Het is al weer enige tijd geleden dat u bezocht werd door een assistente van de afdeling Klinische Genetica Rotterdam. Zij stelde u vragen over uw ervaringen met het erfelijkheidsadvies zoals dat in 1984 op onze afdeling gegeven werd. We beloofden om u over de resultaten van het onderzoek nog iets te vertellen.

164 vragenlijsten zijn tot nu geanaliseerd. Ongeveer 78% van de ondervraagden gaf aan veel aan het erfelijkheidsadvies gehad te hebben. De voornaamste redenen hiervoor waren: gerustgesteld te zijn en weten waar men aan toe was. Ontevredenheid bij 18% werd met name veroorzaakt doordat de oorzaak van de aandoening onbekend was gebleven of dat men niet veel nieuws gehoord had.
Ongeveer 80% van de mensen waren tevreden over de manier waarop het gesprek gevoerd werd. Sommigen vonden de gesprekken te formeel of dat er te weinig werd ingegaan op emoties. Volgens 89% was de inhoud van het gesprek duidelijk. De afsluitende brief werd door 82% voldoende duidelijk gevonden.
Van de echtparen die het boekje "Ziekte en erfelijkheid" meekregen, vonden 65% dit duidelijk. Sommigen vonden echter, dat er te weinig instond over de eigen situatie van het betreffende echtpaar. Na het erfelijkheidsadvies voelde 53% van de echtparen zich gerustgesteld. De niet-gerustgestelden voelden zich teleurgesteld of bezorgd over hetgeen zij te horen hadden gekregen.
11% had meer gesprekken willen hebben op de afdeling, met

name over de ontwikkelingen op medisch gebied bijv. de mogelijkheden van prenatale diagnostiek (= onderzoek tijdens de zwangerschap).

Algemene opmerkingen waren:
1. Er is meer publiciteit nodig rond erfelijkheidsadvies voor het publiek en de artsen.
2. De wens om van te voren te weten wat er in het erfelijkheidsadvies gaat gebeuren.
Meer publiciteit is o.a. verzorgd door de Samenwerkende Ouder- en Patientenverenigingen en de SIRE reclames over erfelijkheidsvoorlichting en -advies. Echtparen die zich aanmelden voor een erfelijkheidsadvies ontvangen tegenwoordig een brief met een beschrijving van wat er gebeuren gaat.
Voor dit algemene overzicht hebben we enkel de meeste algemene opmerkingen weergegeven. De specifieke opmerkingen zijn echter net zo belangrijk als de algemene maar het voert hier te ver om daarop in te gaan.

Wij hebben dus al veel uit het onderzoek geleerd en waar mogelijk proberen we de genoemde bezwaren te verhelpen.
Wij willen u nogmaals heel hartelijk danken voor de medewerking aan het onderzoek.
Voor verdere vragen over het onderzoek bent u altijd welkom bij mij. U kunt mij bereiken via tel. 010-408 7215.

Met vriendelijke groeten,

Mw.Drs. P.G. Frets, psychologe

DANKWOORD

Allen die hun bijdrage hebben geleverd aan de totstandkoming van dit proefschrift wil ik bedanken. Een aantal mensen wil ik in het bijzonder noemen:

Mijn dank gaat allereerst uit naar mijn promotor Prof. Dr. M.F. Niermijer voor zijn deskundige en consciëntieuze begeleiding bij het onderzoek. Bovendien ben ik hem zeer erkentelijk voor zijn heldere analyses, zijn constructieve commentaar en het nauwgezet lezen van de manuscripten. Tevens waren zijn niet aflatend enthousiasme en zijn vertrouwen in mij een heel belangrijke stimulans.

Mijn promotor Prof. Dr. F. Verhage dank ik voor zijn goede adviezen, voor zijn steun bij de opzet en uitwerking van het onderzoek en de vele inspirerende discussies over de psychologische interpretatie van de resultaten.

Prof. Dr. H. Galjaard ben ik zeer erkentelijk voor zijn stimulerende invloed in alle fasen van het onderzoek en voor zijn prikkelende commentaar op het proefschrift.

De Stichting Klinische Genetica Regio Rotterdam ben ik zeer veel dank verschuldigd voor de bijzondere wijze waarop zij in verschillende fasen van het onderzoek gezorgd heeft voor stimulering en begeleiding.

Prof. Dr. R.W. Trijsburg wil ik bedanken voor zijn bereidheid zitting te nemen in de promotiecommissie.

De accurate en enthousiaste werkwijze van de interviewsters Sophie van de Berge, Eva Ketzer en Tineke Peters-Romeyn bij de organisatie en afname van de vragenlijsten waren van groot belang voor de kwaliteit van het onderzoek; hiervoor mijn hartelijke dank.

Benno Bonke, Ruud Erdman, Jan Out en Frans Verhage wil ik bedanken voor het beoordelen van de diepte-interviews.

Jeanette Hoogeboom, Daniëlle Majoor-Krakauer, Martinus Niermeijer en Lodewijk Sandkuyl hebben de patiënten die aan het onderzoek hebben deelgenomen erfelijkheidsadvies gegeven. Hun klinische gegevens hebben de basis gevormd voor dit onderzoek.

Graag wil ik de patiënten bedanken voor hun bereidheid en hun openheid om over voor hen vaak zeer emotionele onderwerpen te praten.

Voorts wil ik de huidige genetic counselors van de Afdeling Klinische Genetica, Erasmus Universiteit en Academisch Ziekenhuis Dijkzigt Rotterdam, Jeanette Hoogeboom, Dick Lindhout, Eveline Wesby-van Swaay bedanken voor hun constructieve commentaar op de manuscripten.

De hulp bij de statistische bewerking door Hugo Duivenvoorden was onmisbaar. Zijn bereidheid om de gebruikte statistische methoden aan anderen voor te leggen heb ik erg

gewaardeerd.

Alice Ribbink-Goslinga ben ik veel dank verschuldigd voor haar zeer deskundige hulp bij het redigeren van dit proefschrift en haar bereidheid om altijd tijd voor mij in te ruimen, ook als die er eigenlijk niet was.

Alle medewerkers van de Afdeling Medische Psychologie en Psychotherapie Erasmus Universiteit Rotterdam wil ik bedanken voor hun collegialiteit en persoonlijke steun.

Mirko Kuit van de Afdeling Fotografie Erasmus Universiteit bedank ik voor zijn toewijding en nauwgezetheid bij het maken van de illustraties.

Graag wil ik de huidige en voormalige administratieve staf van de Afdeling Klinische Genetica, Erasmus Universiteit en Academisch Ziekenhuis Dijkzigt Rotterdam en van de Afdeling Medische Psychologie en Psychotherapie Erasmus Universiteit bedanken voor hun ondersteuning.

In het bijzonder ben ik Jacqueline Drost-van der Linden veel dank verschuldigd voor haar bereidheid en inzet om in zeer korte tijd de lay-out van dit proefschrift te voltooien.

Alexander dank voor je sportiviteit om de PC ten alle tijden aan mij af te staan zodat ik dit "proefwerk" kon afmaken.

Lieve Sten, dank voor je onmisbare steun en geduld. Met zeer veel belangstelling heb je mijn werkzaamheden gevolgd en mij daarmee gestimuleerd. Ook in de minder voorspoedige periodes van het onderzoek wist je mij steeds op juiste en constructieve wijze te steunen en te stimuleren. We hebben samen deze belangrijke fase afgesloten en ik kijk ernaar uit om met jou een volgende fase in te gaan.

CURRICULUM VITAE

5 mei 1957	Geboren
1969 - 1976	Eindexamen Rijnlands Lyceum te Oegstgeest Atheneum A
1976	Aanvang studie Psychologie aan de Vrije Universiteit te Amsterdam
1979	Kandidaatsexamen
sept. 1983 - juni 1984	Stage Psychotherapeutische Gemeenschap "the Ingrebourne Centre" te Londen
25-1-1985	Doctoraal examen Klinische Psychologie
1-3-1985 tot heden	Aanvang aanstelling op de Afdeling Klinische Genetica Erasmus Universiteit en het Academisch Ziekenhuis Dijkzigt Rotterdam met een honoraire aanstelling bij de Afdeling Medische Psychologie en Psychotherapie Erasmus Universiteit Rotterdam. Per 1-3-1989 in vaste dienst.

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