
Reproductive Pasts Reproductive Futures

Genetic Counseling and Its Effectiveness

**James R. Sorenson
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March of Dimes
Birth Defects Foundation

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As part of our efforts to achieve these goals, we sponsor, or participate in, a variety of scientific meetings where all questions relating to birth defects are freely discussed. Through our professional education program we speed the dissemination of information by publishing the proceedings of these and other meetings. From time to time, we also reprint pertinent journal articles to help achieve our goal. Now and then, in the course of these articles or discussions, individual viewpoints may be expressed which go beyond the purely scientific and into controversial matters. It should be noted, therefore, that personal viewpoints about such matters will not be censored but this does not constitute an endorsement of them by the **March of Dimes Birth Defects Foundation**.

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*To our reproductive pasts
and their reproductive futures:*

Peter
Beth and Woody
Stephen, Ruth, and Kenneth
Meghan

List of Tables

2-1	Research Design	19
2-2	Reasons for Case Nonentry Into Study	23
2-3	Client Retention Rates	25
3-1	Institutional Location of Genetic Clinics	32
3-2	Professional Staff Reported to Regularly See Counseling Clients at Clinics	33
3-3	Diseases and Disorders for Which Counseling Is Provided at 47 Clinics	35
3-4	Services Clinics Report Providing Directly to Clients	36
3-5	Clients in Study	37
3-6	Professional Degrees of Counselor Staff	38
3-7	Areas of Specialization, by Type of Professional Degree	38
3-8	Training in Human Genetics, by Type of Professional Degree	39
3-9	Training in Counseling, by Type of Professional Degree	40
3-10	Average Number of Years of Counseling Experience, by Type of Professional Degree	41
3-11	Mean Estimated Time in Selected Professional Activities, by Counselor Degree	41
3-12	Importance of Six Counseling Goals to Counseling Staff	42
3-13	Importance of Seven Tasks in Genetic Counseling to Counseling Staff	43
3-14	Appropriateness of Five Counselor Decision-Making Strategies to Counseling Staff	44
3-15	Marital Status of Study Population	45
3-16	Age of Clients	45
3-17	Clients' Highest Level of Education	46
3-18	Reported Family Income	46
3-19	Clients' Religion	47
3-20	Reproductive Experience of Female Client Population	47
3-21	Problems and Disorders for Which Clients Were Most Frequently Counseled	49

xiv / List of Tables

4-1	Most Commonly Occurring Questions and Concerns	53
4-2	Percent of Female Clients Saying They Came to Counseling to Discuss Various Topics	54
4-3	Mean Number of Topics Client Wanted to Discuss, by Area and Client Reproductive Experience	56
4-4	Percent of Female Clients Wanting to Discuss a Topic Before Counseling Who Reported Having Discussed It in Some Depth During Counseling	58
4-5	Percent of Topics Clients Wanted to Discuss That Were Discussed, by Client Reproductive Experience	60
4-6	Agreement Between Topic Client Most Wanted to Discuss and Counselor's Assessment of What That Topic Was	61
4-7	Reasons Cited by Female Clients Why All Their Genetic- Medical Questions and Concerns Were Not Discussed	62
4-8	Reasons Cited by Female Clients Why All Their Socio- medical Questions and Concerns Were Not Discussed	62
4-9	Female Clients Scheduled for Additional Counseling Who Reported Counselor Had to Gather More Information	63
4-10	Distribution of Clinic Scores for Discussing With Clients the Genetic-Medical Topics Clients Wanted Discussed	66
4-11	Distribution of Clinic Scores for Discussing With Clients the Sociomedical Topics Clients Wanted Discussed	67
4-12	Relationship Between Estimated Length of Counseling Session and Discussion of Client Genetic-Medical and Sociomedical Questions and Concerns	69
4-13	Female Clients' Desire for More Genetic Counseling Six Months After Initial Counseling Session	74
4-14	Client Reasons for Wanting Additional Genetic Counseling	75
4-15	Topics Clients Sought Information About After Counseling	75
5-1	Female Clients' Knowledge of Diagnosis Before, Immediately After, and Six Months After Counseling	82
5-2	Comparison of Female Clients' Knowledge of Diagnosis Before and After Counseling	83
5-3	Female Clients' Knowledge of Risk Before, Immediately After, and Six Months After Counseling	85
5-4	Comparison of Female Clients' Knowledge of Risk Before and After Counseling	86
5-5	Magnitude of Errors in Risk Estimation by Female Clients Before and After Counseling	87
5-6	Comparison of Client and Counselor Attitudes About Clients' Risk	97

5-7	Changes in Clients' Attitudes About Numeric Risk Before and After Counseling	98
5-8	Female Client's Sense of Disorder Burden Before and After Counseling	100
6-1	Female Clients Rating Factors as Very Important in Their Reproductive Planning	108
6-2	Female Client Reproductive Considerations Rated as Very Important, by Pregnancy and Planned Pregnancy Status	110
6-3	Reproductive Considerations Rated Most Important, by Female Client Reproductive Experience	111
6-4	Female Client Interest in Counseling to Obtain Information to Make Reproductive Decision, by Reproductive Plans	113
6-5	Female Clients Seeking Reproductive Planning Information in Counseling, by Reproductive Experience	114
6-6	Profile of Questions and Concerns of Clients Seeking and Not Seeking Reproductive Planning Information in Their Counseling ..	115
6-7	Female Client Short- and Long-Term Reproductive Plans Prior to Genetic Counseling	116
6-8	Female Clients Pregnant at Entrance to Study	117
6-9	Female Client Intentions to Have a Child Within Two Years, by Reproductive Experience	118
6-10	Changes in Female Client Two-Year Reproductive Plans From Before to After Counseling	121
6-11	Changes in Female Client Two-Year Reproductive Plans From Before to Six Months After Counseling	122
6-12	Reproductive Plans Before and Six Months After Counseling for Clients Influenced by Their Counseling	123
6-13	Reproductive Plans Before and Six Months After Counseling for Clients Not Influenced by Their Counseling	124
6-14	Female Clients' Reproductive Plans Six Months After Counseling and Role of Counseling in Those Plans	125

Foreword

Although birth defects are as old as the human race, early advances in understanding human inheritance were limited. Beginning in 1910, with the rediscovery of the work of the great pioneer Gregor Mendel, there was steady but slow growth in knowledge of the mechanisms of inheritance and in understanding the causes of certain birth defects.

In the last three decades, however, the field of human genetics has virtually exploded. In terms of scientific understanding, research on DNA holds promise of remarkable developments. In addition, there has been significant growth in charting the role of genetics in more and more birth defects and in making increasingly precise estimates of the likelihood that such defects will occur in specific pregnancies. Perhaps the most spectacular technologic advances have involved the development of a wide array of prenatal diagnostic procedures.

This increased understanding of human inheritance, and associated technologies, is unfortunately clinically applicable today to but a minority of the population with and at risk for birth defects. As investigators continue to explore new frontiers, what lies ahead will undoubtedly be pertinent to many more people.

The field of applied human genetics rests on this foundation of rapidly expanding knowledge of inheritance and technologic developments. Clients seeking genetic or birth defects counseling confront many novel issues and concerns, as do the providers of these services. Genetic counseling, a hybrid field slightly more than 25 years old, is expected — by practitioners, clients, and society — to provide expert services with the same effectiveness as other, more developed areas of medicine.

Whatever its basis in knowledge and technology, whatever the expectations of practitioners, clients, and society, genetic counseling is an evolving and expanding service. It exists, it provides services — some better than others, and it touches the lives of thousands of individuals.

This book examines the nature of the services provided today from two points of view — providers' and recipients'. We examine the discrepancy in expectations and assessments of services by these two parties. We try to ascertain which services are good and which are lacking. We also include contextual factors and their influence on services. In our view, a major missing ingredient in the growth of genetic counseling has been a virtual absence of feedback to providers about the services and their effectiveness. In contrast with the laboratory situation, where investigators see with their own eyes the impact of varying procedures, in genetic counseling this important ingredient is often missing. Accordingly, genetic counselors cannot change ineffective practices, and genetic counseling cannot be modified for maximum effectiveness.

The study reported here is designed to provide some of this necessary information, so that providers may develop more effective genetic counseling and clients may find the genetics of the 21st century humane and useful to them in living with their reproductive pasts and achieving their varied reproductive futures.

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We would also like to acknowledge the support and encouragement of the March of Dimes in this research. The March of Dimes' role encompassed three activities. First, they provided funding through a series of grants to the research team. Second, they served as initial liaison between the genetic counseling clinics and the researchers, encouraging the participation of the clinics and their staffs in the research project. Third, they gave the research team a single mandate: to evaluate the effectiveness of genetic counseling in the clinics to which they were providing service funds.

Beyond this, the March of Dimes played no role in the design, conduct, or conclusions of the research. Determining what aspects of genetic counseling were to be studied, how they were to be studied, and what constituted effective genetic counseling was solely the responsibility of the research team, which consisted of N.A. Scotch, PhD, J.R. Sorenson, PhD, and J.P. Swazey, PhD, Co-Principal Investigators; D.B. Matthews, MD, PhD, Project Director; Carole M. Kavanagh, MS, Project Coordinator; and M. Griffin and C. Goodman, Research Assistants. Statistical consultation was provided by J. Barrett and computer assistance by Marc Mucatel.

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Contents

List of Tables	xiii
Foreword	xvii
Acknowledgments	xix
Chapter 1 Genetic Counseling: Definitions and Goals	
Introduction	1
Genetic Counseling: A Brief History	3
What Is Genetic Counseling?	5
Is Genetic Counseling Effective?	7
The Study	10
Organization of the Monograph	11
Summary	12
References	13
Chapter 2 The Study: Instrumentation, Design, and Methods	
Introduction	15
The Pilot Study	15
Instrumentation	16
Design	17
Procedures	20
Clinic Participation Rates	21
Client Participation Rates	22
Client Retention Rates	24
Counselor Participation Rates	26
Special Methodologic Considerations	26
Instrument Effects	26
Postcounseling Questionnaires	27
Representativeness of the Study Population	28
Summary	29
References	30
Chapter 3 Clinics, Counselors, and Clients	
Introduction	31
Clinic Settings	31
Staffing Patterns	33
Clinic Services	34
The Organization of Counseling Services	35

The Counselors	37
Specialization	37
Professional Training in Genetics and Counseling	39
Counseling as a Professional Activity	40
The Counselors' Objectives	41
Clients	45
Sociodemographic Characteristics	45
Reproductive History	46
Medical Concerns	48
Summary	48
References	50
Chapter 4 Effective Genetic Counseling: Discussing Client Questions and Concerns	
Introduction	51
Clients' Questions and Concerns	52
Does Counseling Address the Topics Clients Want to Discuss?	57
Meeting Client Needs: Clinic, Counseling Process, Client, and Counselor Factors	64
Clinics	65
Clinic Factors Associated With "Low" and "High" Clinic Scores	68
Counseling Process Factors	68
Client Characteristics	70
Counselor Factors	71
Clients' Six-Month Assessment of Their Genetic Counseling Experiences ..	73
Summary	76
References	77
Chapter 5 Effective Genetic Counseling: More Informed Clients	
Introduction	79
Client Diagnostic Knowledge	80
Client Knowledge of Risk	84
Factors Associated With Accurate Client Knowledge Postcounseling	88
Clinics	88
Counselors	91
Clients	93
Client Attitudinal Changes	96
Summary	101
References	104
Chapter 6 Genetic Counseling and Client Reproductive Plans	
Introduction	105
Client Considerations in Reproductive Planning	107
Client Reproductive Plans	112
Changes in Client Reproductive Plans	119
Genetic Counseling and Its Influence on Client Reproductive Plans	122
Summary	127
References	129

Chapter 7 Medical Genetics and Genetic Counseling

Introduction	131
Is Genetic Counseling Effective?	132
Toward More Effective Genetic Counseling	136
Teaching and Learning Medical-Genetic Information	136
The Provision of Counseling	137
Counselor Characteristics	138
Client Characteristics	139
Counseling Clients	140
Counseling and Medical Genetics	142
References	144
Appendix 1 Counselor Questionnaires	147
Appendix 2 Client Questionnaires	159
Appendix 3 Directors, at Time of Study, and Participating Clinics	183
Index	191

Chapter 1

Genetic Counseling: Definitions and Goals

INTRODUCTION

In 1981 approximately 3,300,000 babies will be born in the United States. Between 150,000 and 200,000 of these newborns will be diagnosed as having a birth defect, that is, a structural or metabolic disease or disorder that is genetically determined or the result of environmental influence during embryonic or fetal development [1, 2]. These birth defects will range from mild to severe to fatal, and involve symptoms which may be physical or mental, or both. Some defects will be present at birth; others will appear later in life. Some disorders will occur throughout the population, while others will be confined to certain ethnic or social groups. Some birth defects will be treatable, but most will impose a lifetime of limitation on the biologic, psychological, or intellectual functions of the affected individual. In all, in 1981 some 15,000,000 Americans of all ages will be living under varying levels of handicap due to one or more birth defects [1].

Statistics such as these, imperfect as they are and open to revision as knowledge changes, counter the notion that birth defects are not an important public health problem. Indeed, each individual type of birth defect is rare; but collectively birth defects have major public health significance. Birth defects are today a leading cause of mortality in this country in the early years of life [3]. In addition, because of their chronic nature, they account for sizable amounts of health expenditures [2]. In addition it has been estimated that birth defects, because of their usual early onset, account for a heavier loss or reduction in productive future years than other more widely recognized public health problems such as cardiovascular disease, cancer, and stroke [4].

A second perspective gained from such statistics is that birth defects are a recurring threat to a sizable proportion of each generation of children. While medical science has gained dramatic control over some major health problems, such as certain serious infectious diseases, it appears that knowledge of the causes, treatment, and prevention of birth defects has not progressed as much or as rapidly as needed.