Effectiveness of Genetic Counseling for Families with Craniofacial Anomalies

HILLARY BRODER, PH.D.
WILLIAM C. TRIER, M.D.

Questionnaire data about genetic counseling experiences are presented from 37 parents and 25 patients who were evaluated. The subjects' perceptions vary regarding the cause of cleft lip or palate or both. More positive feelings about the birth defect are expressed after the subjects receive counseling. However, 25 percent of the sample express negative feelings about the birth defect after counseling. Ninety percent of the subjects indicate that counseling should occur within the first 3 months of the birth of the infant with a cleft. The subjects agree significantly on the important components of genetic counseling. The authors conclude that effective counseling includes (1) providing facts, alleviating guilt, and dispelling misperceptions; (2) discussing decision making; and (3) facilitating the coping process. Implications for further research and a protocol for counseling are suggested.

Orofacial and craniofacial patients present with an array of problems that require evaluation and treatment by various specialists including plastic surgeons, dentists, speech pathologists, geneticists, and psychologists. Depending upon the size and philosophy of a craniofacial or orofacial team, more than one specialist may offer the patient counseling. The term "counseling" implies a two-way exchange between patient and professional, not just passive reception of information by the patient. A very important aspect of orofacial or craniofacial disorders is the possible inheritance of the disorder by subsequent siblings or by offspring of the patient. Therefore, in addition to providing accurate genetic information, the counselor must be sensitive to the patients' or the parents' emotional responses (Haan, 1979). Effective counseling must provide accurate medical information and help patients face future childbearing decisions and cope successfully within the family system.

Research reports documenting the effectiveness of genetic counseling often rely on retrospective accounts, and samples vary in regard to the type of birth defect (Reynolds et al, 1974). Although there is no simple profile of the client in a genetics clinic, barriers to successful counseling have been identified in the literature. These barriers include emotional responses such as guilt, anger, anxiety, and depression (Leonard et al, 1972), the timing of counseling (Ives et al, 1979), and the fear that future offspring will have similar birth defects (Lippman-Hand and Fraser 1979b; Tishler, 1981).

To better understand the effectiveness of genetic counseling with orofacial patients, the authors conducted a pilot study at the Oral-Facial and Communicative Disorders Program, a diagnostic and treatment center at the University of North Carolina in Chapel Hill. Patients responded to a mailed questionnaire. The specific aims were to have the respondents identify important components of genetic

The authors are affiliated with the Oral-Facial and Communicative Disorders Program at the University of North Carolina, Chapel Hill, NC. This paper was presented in part at the Convention of the American Cleft Palate Association in Seattle, Washington, in May 1984.

counseling and evaluate previous counseling experiences.

METHOD

Subjects

The subjects were patients or parents of patients from the data bank registry of the Oral-Facial and Communicative Disorders Program. The registry consisted of 839 patients. This pool of patients was divided into five age groups: 0 to 6 years, 6 to 12 years, 12 to 18 years, 18 to 30 years, and over 30 years.

After a proportional number of subjects from each age group was chosen, subjects were selected using a table of random numbers (Snedecor and Cochran, 1971). This selection allows representative samples of varied treatment periods which could have historical and clinical relevance. Because of the smaller number of patients in the over 30 age group, a higher proportion of this age group was selected.

Table 1 reveals the total number of patients, the number of patients returning the questionnaire, and the response rate for each age group.

The sample consisted of 62 subjects. The parents (N=37) were the subjects for all patients under 18 years of age; patients who were over 18 years of age (N=25) themselves were the subjects. Fifty-one percent of the subjects responded to the survey.

Approximately 53 subjects (85%) were parents or patients with a cleft lip with or without cleft palate; 9 subjects (15%) represented patients having speech or language disorders or both, sometimes accompanied by physical problems such as a high-arched palate or a neuromuscular deficiency.

Forty-two percent of the subjects were females and 58 percent were males. Ninety percent of the subjects were white, and 10 percent were black or American Indian.

TABLE 1. Distribution of Subjects by Age Group and Questionnaire Response Rate

Age Group	*OFCDP Patient Population*	*N (sample)**	*Response Rate***
0–6 years	114	9	56%
6 < 12 years	230	19	51%
12 < 18 years	229	9	31%
18 < 30 years	196	17	55%
≥ 30 years	70	8	66%

*N = 62

**Because of address changes, 25 patients were deleted from the study.

INSTRUMENTATION

Based on a literature review of genetic counseling and clinical observations of orofacial patients, the authors constructed a questionnaire and mailed it to the members of the study group.

The purpose of this instrument was to elicit retrospective accounts of the patients' or their parents' experiences with genetic counseling and to have patients identify factors that maximize the effectiveness of counseling. Examples of effectiveness of counseling are an increase in the retention of correct genetic information for the patient and the patient's family, and facilitation of successful family planning by the patient or the patient's family.

The questionnaire consisted of 14 open-ended or multiple-choice items which described and evaluated the subject's previous genetic counseling session(s). Using a 5-point Likert-like scale (Kerlinger, 1965), the subjects also rated the importance of the counseling factors. Additional comments were requested at the end of the questionnaire.

PROCEDURE

Each subject selected for inclusion in this study received a mailed questionnaire, a stamped return envelope, and a consent form. Approximately 3 weeks later, a second identical questionnaire was mailed to the subjects who did not return the initial questionnaire. The study was approved by the University of North Carolina Committee on Investigations Involving Human Subjects.

Frequency distributions and percentages were calculated to describe the data and to reveal any trends. The subjects'

"additional comments" were noted. The responses were divided by test item and age group to observe historical changes in genetic counseling and the Oral-Facial and Communicative Disorders Program.

RESULTS

One-third of the patients or their families recalled receiving genetic counseling. The majority of counseling was provided by the plastic surgeon, a speech pathologist, or a pediatrician. Patients who desired formal professional genetic counseling received this from the medical geneticist or genetic counselor. The majority of subjects who recalled receiving genetic counseling were parents of children in the 0 to 6 year age group and patients who were in the 18 to 30 year age group.

Subjects' estimates of risk of recurrence of the birth defect and the perceived level of risk are presented in Table 2. Approximately one-half of the sample reported the correct incidence of inheritance for cleft lip with or without cleft palate (2 to 5%). One-third of the subjects considered the risk of recurrence low; the remainder considered the risk medium to high. Although the perceptions did not appear to be associated with the degree of risk, this observation may depend on psychosocial factors that warrant further investigation. For example, a couple may be middle-aged, have offspring with other medical problems, or have financial difficulties; thus, they consider the 5 percent risk level high because of other stresses. On the other hand, a childless couple, desiring children may consider a 2 to 5 percent risk low.

Over 80 percent of the subjects reported no counseling prior to their evaluation and treatment at the University of North Carolina. One-half of the sample reported that both parents attended the session; 46 percent reported attendance by one parent (6 were single parents). Five percent of the subjects were adults or adolescents whose mothers accompanied them to the sessions. One-quarter of the sample had a follow-up genetic counseling session.

The timing of counseling recommended by the subjects is presented in Table 3. Ninety percent of patients or their families expressed a desire for counseling during the first few months after the birth of a child with a defect, and 66 percent recommended counseling as early as possible.

TABLE 2. Estimate of Recurrence of Birth Defect and Perceived Level of Risk

Estimated Recurrence Risks	*% of Sample*	*Risk Level Perceived By Respondents*	*% of Sample*
2–5%	48	Low	33
10–25%	32	Medium	42
50%	21	High	25

TABLE 3. Subjects' Recommended Timing of Counseling After Birth of Child with Defect

Timing Recommended	*N*	*%*
As soon as possible	30	66
1 Month–3 months	11	24
1 Year	2	4
Not at all	0	0
Uncertain	3	6

The subjects were given a list of possible causes associated with the birth defect and were permitted to make multiple entries. Table 4 summarizes their responses regarding etiology. The variety of responses by the subjects of the perceived causes of cleft lip with or without cleft palate suggest that subjects lack a clear understanding of the defect.

Table 5 reveals the clients' feelings about

TABLE 4. Perception of Causes of Birth Defects

Cause	*N*
Many factors	24
Family's genes (hereditary)	21
Drugs or alcohol	13
Unknown (no idea)	11
Environmental influence (such as x-rays)	8
God's will	7
Injury during pregnancy	5
Injury during delivery	4
Emotional problems	3
Diet	2
Other	2

TABLE 5. Subjects' Feelings About Their Birth Defect

Feeling	*Pre-Counseling*		*Post-Counseling*	
	N	*% of Sample*	*N*	*% of Sample*
Positive	12	34	19	56
Neutral	6	19	5	16
Negative	13	38	9	25
Can't remember	3	9	1	<1

the birth defect at the time of counseling and following counseling. Although 27 percent reported more positive feelings after counseling, 41 percent did not report positive feelings about the birth defect. Guilt, disappointment, anxiety, and depression were among the negative feelings expressed by the subjects.

Sixty-five percent of the respondents indicated that they did not have a follow-up session; 26 percent reported having a follow-up session. Three subjects reported that they could not remember.

Table 6 lists the factors that the subjects reported to be important in genetic counseling. The table includes the percentage of subjects rating the factor moderately (4) or very (5) important on a 5-point scale. The consistency of the responses is impressive. Ninety-six percent of the subjects reported that the professionals' factual written information is extremely useful to them. Treatment and emotional factors are also rated as important to these subjects.

TABLE 6. Important Factors in Genetic Counseling

Factors Rated Very Or Moderately Important	*% of Sample*
Letter summarizing the findings and recommendations	96
Follow-up meeting with counselor	88
Discussing client's feelings about birth defect	88
Appointments for treatment or consultations	88
Discussing (questions/feelings) family planning	88
Sympathetic/understanding attitude from counselor	84
Both parents attending session	76

In the "additional comments" section of the questionnaire, the subjects reviewed their feelings about treatment. Thirty-eight percent of the subjects who had not received genetic counseling either requested more genetic information on cleft lip, with or without cleft palate or genetic counseling. The majority of the requests for counseling were from subjects in the 18- to 30-year-old group.

Discussion

Counseling implies a meaningful exchange between the patients and the counselor. The exchange is meaningful only if the patients' needs are met and the genetic information is understood by them.

This study suggests that the protocol and experience of genetic counseling varies. Riccardi et al (1978) have noted that hospital records of genetic counseling are unsystematic. The patients' perceptions and feelings about the counseling experience also vary, a difference which may reflect a variety of psychosocial factors.

Contrary to the presumption that very early genetic counseling might increase the burden of guilt upon parents of a child with a congenital defect, it is clear that the majority of subjects believe that genetic counseling should be provided as early as possible. Our respondents suggest that the ideal time for counseling is "as soon as possible". This finding, which contradicts earlier reports in the literature, may be a product of poor recall or it may reflect differences between samples with different etiologies, e.g., chromosomal (Down's syndrome) versus multifactoral (cleft lip with or without cleft palate) or samples with a different potential for treatment (e.g., the cleft of the lip is usually repaired at 3 months of age). In any case, the timing of genetic counseling should be explored in further research.

Furthermore, to be certain that genetic information is provided, one or more members of the treatment team should always provide this information. Other members of the team should determine that genetic information has been provided and may reinforce information or correct mis-

understood impressions. No matter when or where a previous evaluation and treatment have been provided, it should never be assumed that genetic counseling has previously been provided or that the information has been understood.

"Why?" and "Could it happen again?" are the first questions patients in counseling ask (Reynolds et al, 1974). Since our results reveal that many patients do not understand the etiology of the defect, we question the effectiveness of the counseling experience. Clearly the patients have psychosocial barriers that need to be investigated by professionals serving craniofacial and orofacial patients. The data also reveal negative feelings about the defect by patients before and after counseling.

The results from the survey indicate that a follow-up to initial counseling should be provided. Counselors should reinforce the initial understanding of the patient or the patient's family and also discuss in detail the import of such genetic information. Simply providing a percentage probability for inheritance of a cleft lip or palate is not enough. The age of the parents, number of siblings, quality of correction of a defect, potential impact of the defect upon a child, parents' attitude toward the defect, and importance to the parents of having another child would all need to be weighed with the assistance of the counselor.

It is quite clear that a formal letter containing genetic and other information should always follow the evaluation, so that the patient or his or her family can refer to this information in the future and can share this information with others who might have a need to know it.

It appears that the 18- to 30-year-old age group, in particular, would find genetic counseling important in their family planning.

Finally, the retrospective pilot study on the efficacy of genetic counseling suggests the need for a further study in which the effectiveness of genetic counseling could be determined. The extent of the parents' knowledge regarding the incidence and causes of a particular defect and their feelings about the deformity resulting from the defect may serve as measurements of effective counseling.

CONCLUSION

From our clinical experience, reinforced by the questionnaire data, we conclude that providing genetic counseling to parents of children with craniofacial anomalies has three elements. The overall effectiveness of genetic counseling can depend on the success of each element.

Giving the Facts, Alleviating Guilt, and Dispelling Misconceptions. Genetic counseling informs parents of the cause of the child's malformation. Sometimes fear of the unknown may be related to the parents' inability or unwillingness to comprehend medical information. Fear may also distort parents' perceptions of the financial, psychological, and medical needs of the affected child which can constitute an additional burden (Leonard et al, 1972). Many parents inappropriately place blame on themselves (Lippman-Hand and Fraser, 1979a), which leads to lower self-esteem (Smith and Antley, 1979) and deficient communication. By providing parents with a medical explanation for their child's condition, professionals hope to lessen the burden of guilt, provide parents with useful information regarding their child's problem and treatment, and possibly refer them to other professionals.

Decision Making. Genetic counseling provides parents or patients with information about the risk of future children being affected with a similar condition. Based on the known risk information, parents' attitudes toward having more children and other options for parenting can be addressed. This issue is especially relevant to the genetic counseling session where couples make decisions based on their perceptions and the seriousness of the risk. For example, a 10 percent risk may be considered low by research geneticists (Lippman-Hand and Fraser, 1979b). Risk levels may be perceived differently by patients. For prospective parents who do not wish to face their genetic risks, adoption or artificial insemination may be alternatives. Prenatal diagnosis for certain con-

ditions is also available. Decision making based on accurate risk information may lessen the chance of bearing another child with a craniofacial malformation.

Coping Process. It is recognized that parents of a child with a malformation feel shock, guilt, denial, and other stress-related feelings (Dicker and Dicker, 1978). Our findings suggest that these feelings are lessened after counseling. However, persistent negative feelings for some parents can seriously impair the communication process and the family system.

Genetic counseling can give parents an avenue to express their feelings. Responses by parents and older patients reveal that their expression of feelings is as important as the medical information they receive in the counseling sessions. Follow-up counseling is also desired by patients, since misconceptions may persist which can inhibit their acceptance of the defective child and their communication with that child. Although the clients' issues may vary, the important factors in counseling are overwhelmingly consistent (see Table 6).

A consistent protocol for counseling patients would allow professionals a more reliable means to evaluate their counseling. The professionals (geneticist, speech pathologist, plastic surgeon) who provide this service also warrant further examination. A longitudinal study of the efficacy of genetic counseling for craniofacial anomalies has not been undertaken to date. A unique approach to genetic counseling may be appropriate for these families, since most of these disorders are visible at the time of the birth. Parents need information regarding the defect and immediate help in accepting their child's appearance and condition; their attitudes may affect the child's self-concept (Broder, 1980). Later genetic counseling can focus on patients' risks of having future children with birth defects and can correct any misperceptions that linger.

References

BRODER H. Perceptions of children with visible or invisible oral-facial defects (Doctoral dissertation, Duke University, 1980). Dissertation Abstracts International, 1980 (University Microfilms No. 81–01, 720).

DICKER M, DICKER L. Genetic counseling as an occupation specialty: A sociological perspective. Soc Biol 1978; 24:272.

HAAN NG. Psychosocial meanings of unfavorable medical forecasts. In: Stone GC, Cohen F, Adler NE, et al, eds. Health psychology–a handbook. San Francisco: Jossey-Bass Inc, Publishers, 1979.

IVES EJ, HENICK P, LEVERS MI. The malformed newborn–telling the parents. Birth Defects 1979; 15:223.

KERLINGER FN. Foundation of Behavioral Research. New York: Rinehart and Winston, 1965.

LEONARD CO, CHASE GA, CHILDS B. Genetic counseling: A consumer's view. N Engl J Med 1972; 287:433.

LIPPMAN-HAND A, FRASER FC. Genetic counseling: provision and reception of information. Am J Med Genet 1979a; 3:113.

LIPPMAN-HAND A, FRASER FC. Genetic counseling: The post-counseling period: II. Making reproductive choices. Am J Med Genet 1979b; 4:73.

REYNOLDS BV, PUCK MH, ROBINSON A. Genetic counseling: an appraisal. Clin Genet 1974; 5:177.

RICCARDI VM, COHEN A, CHEN MT. Genetic counseling as part of hospital care. Am J Public Health 1978; 68(7):652.

SMITH RW, ANTLEY RM. Anger: a significant obstacle to informed decision making in genetic counseling. Birth Defects 1979; 15:256.

SNEDECON GW, COCKRAN WG. Statistical Methods (6th ed). Ames: Iowa State University Press, 1971.

TISHLER CL. The psychological aspects of genetic counseling. Am J Nursing 1981; 4:733.