

Prenatal Screening for Hemoglobinopathies. II. Evaluation of Counseling

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Summary

Learning during genetic counseling is often below expectations, especially in the context of genetic screening. In this report we describe learning as a result of genetic counseling of 298 pregnant women identified as hemoglobinopathy carriers, 234 with sickle cell trait and 64 with beta-thalassemia trait. Counseling was designed to provide the information needed in a simple, clear, and nondirective manner. A special videotape produced for this purpose provided dramatization and a role model illustrating an appropriate response. After viewing the videotape the counselee had an opportunity to question the counselor and to have any misconceptions corrected. Questionnaires revealed significantly increased knowledge as a result of counseling in each of the three hemoglobinopathy subject areas tested—namely, clinical manifestations, genetics, and prenatal diagnosis. Five factors correlated with higher knowledge scores after counseling—namely, a younger patient age, more years of education, knowledge of having trait before this identification, knowledge of the baby's father having trait before counseling, and having no prior children.

Introduction

There are both theoretical and practical reasons to evaluate genetic counseling. Theoretical reasons include analyzing factors affecting comprehension of genetic information, determining how genetic risks influence decision making, and characterizing patterns of adjustments to genetic burdens. Practical reasons include improving the quality of services to patients and both improving the training and evaluating the performance of genetic counselors.

Learning during genetic counseling often falls below expectations, as revealed by objective evaluation after counseling (Meyerowitz and Lipkin 1976; Shaw 1977; Evers-Kiebooms and van den Berghe 1979). A follow-up of parents who were provided genetic counseling for cystic fibrosis, phenylketonuria, and Down syndrome revealed that only about half the couples had the kind of comprehension that could make the information helpful to them (Leonard et al. 1972).

Many of the counselees had difficulty comprehending the information because they lacked an understanding of probability and of basic biological concepts. In a multicenter study in which counseling was evaluated by questionnaire before and after counseling, 53% of clients given a recurrence risk figure and 40% of clients given a diagnosis did not know these items after counseling (Sorenson et al. 1981).

The setting of genetic screening provides further impediments to learning during genetic counseling. Testing is often initiated by the provider rather than arising from a spontaneous question of the counselee. The counselee may be unfamiliar with the disease in question. Finally, the significance of being a carrier is usually not clear to the counselee in advance. Individuals screened and counseled for sickle cell trait frequently were unable to recall what they had been told (Whitten et al. 1981). Individuals tested for Tay-Sachs trait generally had deficient knowledge despite an educational program provided before testing (Childs et al. 1976).

A national committee has formulated recommendations for the evaluation of genetic counseling (Rosenstock and Shaw 1977). The patient population should be clearly defined so that bias in patient selection can be ascertained. Methods of counseling and of evalua-

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tion should be clearly defined in advance. Undesirable outcomes, as well as desired outcomes, should be assessed. Evaluation should be performed by someone not involved in the initial counseling. In our evaluation of the impact of prenatal screening for hemoglobinopathies, we have tried to follow these recommendations in evaluating genetic counseling.

Methods

The overall design of the study, the enlistment of prenatal care providers, the methods used for diagnosing hemoglobinopathies, the diagnoses made, and the characteristics of the counselees are detailed in the preceding paper in this issue (Rowley et al. 1991). The counselors (S.L., C.J.S., and M.W.) were genetic associates experienced in counseling for hemoglobinopathies. Prior to being counseled, patients were asked for informed consent to participate in the research portion of the study, which involved answering questionnaires; all agreed. The 234 sickle cell trait patients and 64 beta-thalassemia trait patients were asked to complete the questionnaires shown in figure 1 of the preceding paper in this issue; patients with other hemoglobin genotypes were asked to complete only the demographic questionnaire.

Information provided at counseling was intended to be simple, clear, and nondirective. An outline of the information presented to patients who had sickle cell trait is shown in table 1; information provided to pa-

tients who had beta-thalassemia trait was analogous. In the case of sickle cell trait and beta-thalassemia trait, this information was provided in the form of a specially produced color-sound videotape. All of the actors in the sickle cell trait tape were black, and all actors in the beta-thalassemia trait tape were white. Each tape shows an attractive pregnant woman in her physician's office, receiving an explanation of her carrier state. The physician provides a clear and readily understood message covering the points provided in table 1. The patient shows an appropriate level of concern and asks questions throughout. The principal message of the tape is that, if the patient wishes to know whether the baby is truly at risk for the disease in question, the baby's father must also be tested. The patient states that she wants him to be tested but doubts that she can get him to agree. At this point the physician shows her a taped sequence depicting another couple at home. The woman has just returned from her physician, who has told her that she is a hemoglobinopathy carrier. She tells her partner she would like him to be tested. Although initially reluctant to be tested ("There's nothing wrong with me!"), her partner finally agrees, to please his wife. Having seen this dramatization, the original patient then expresses confidence that she also can convince her partner to be tested. The tape ends with the physician emphasizing that, if the partner is negative, the couple will be reassured, whereas, if the partner is positive, the decision about whether to have prenatal diagnosis

Table 1

Information Provided at Counseling: Sickle Cell Trait

Type of Information	Content
Name of condition I have	Sickle cell trait
Characteristics of trait form	Detected by blood test Without symptoms Sickle gene inherited from one parent
Name of disease my baby could be at risk for	Sickle cell anemia
Characteristics of disease	Easy fatiguability Attacks of pain Susceptibility to infection Treatment unsatisfactory Sickle gene inherited from both parents
Risks to a child of a trait individual.....	By normal partner→50% normal + 50% trait By trait partner→25% normal + 50% trait + 25% disease
How to determine whether baby is at risk.....	Have baby's father tested
If baby is at risk, options.....	No further action Prenatal diagnosis
If prenatal diagnosis shows that baby has disease, options...	Continue pregnancy Terminate pregnancy

is entirely theirs. The tape lasts 20 min. (The script is available on request.)

Individual counseling was then conducted. After viewing the tape, the patient had an opportunity to ask the counselor questions. These questions sometimes revealed misconceptions, and, if so, the counselor corrected them. The counselor then asked whether the patient wanted to have her partner tested. Additional questionnaires were administered, including knowledge of the material presented. Finally, the patient was given a brochure containing a statement of the main points covered during counseling; handing the counselee a written account increases the proportion of counselees who correctly recall risk figures (Hsia and Silverberg 1973).

Results and Discussion

The characteristics of the 551 pregnancies in which the patient came for counseling, as well as the respects in which they differed from the 259 pregnancies in which the patient did not come for counseling, have been summarized in the preceding paper in this issue (Rowley et al. 1991). Of the 463 women counseled in their first pregnancy, learning was assessed by questionnaire only in the 234 with sickle cell trait and in the 64 with beta-thalassemia trait. This subgroup consisting of the sickle cell trait patients and beta-thalassemia trait patients differed from the total counseled group only with regard to having a higher percentage who, before our identification of them as carriers, were aware of their diagnosis (43% vs. 34%; $P < .0001$) and aware of their risk for having a child with a hemoglobinopathy (36% vs. 20%; $P < .0001$).

The effect that counseling had on the patient's knowledge in this subgroup is shown in table 2. Knowledge of hemoglobinopathy manifestations, of genetics, and of prenatal diagnosis each significantly increased after counseling ($P < .001$ for each).

To identify factors predictive of post-counseling knowledge, we analyzed the relationship between various counselee characteristics and postcounseling scores in each area of knowledge tested. At the beginning of the study we predicted that a patient would have a higher score on the knowledge questionnaire if she did not know the carrier status of her partner, had a gestational age less than 18 wk, had more years of education, and had no prior children. We thought her knowledge would be influenced also by prior awareness that she had trait and by her age.

The actual results obtained by univariate analysis are shown in table 3. With one exception, more years of education and awareness of having trait before our identification each predicted a higher score in each of the three areas.

The results of the multivariate analysis are shown in table 4. Patients who were younger, who had more years of education, who knew before being identified by this program that they had trait, or who knew before counseling that their partner had trait had greater postcounseling knowledge of the manifestations of the particular hemoglobinopathy. Patients who were younger or had more years of education had a better postcounseling knowledge of genetics. Finally, patients who knew before the identification by this program that they had trait, who had more years of education, or who had no prior children had a greater postcounseling knowledge of prenatal diagnosis. The patient's gestational age was not predictive of postcounseling knowledge.

It is not surprising that women with more education learn more from genetic counseling. However, we had not predicted that younger women would learn more, since many were adolescents—some only 12 or 13 years old. We have noticed that women in their first pregnancy are more anxious and hence perhaps more eager for information; this fact probably contributes to the correlation of learning with age. The fact that

Table 2

Effect of Counseling on Patient Knowledge

SUBJECT	MEAN SCORE (% correct)		P Value
	Before Counseling	After Counseling	
Knowledge of hemoglobinopathy manifestations (N = 265)	43	73	<.001
Knowledge of genetics (N = 264)	32	64	<.001
Knowledge of prenatal diagnosis (N = 264)	38	69	<.001

NOTE—A questionnaire with any unanswered item was omitted.

Table 3**Predictors of Patient's Postcounseling Knowledge: Univariate Analysis**

POSTCOUNSELING KNOWLEDGE OF	P VALUE FOR	
	Years of Education	Prior Awareness of Trait Diagnosis
Disease manifestations.....	<.0001	<.01
Genetics	<.0002	NS
Prenatal diagnosis.....	<.04	<.01

NOTE. — The relationship between counselee's postcounseling scores for each of the three areas of knowledge tested, and other counselee characteristics was determined by *t*-test. Gestational age, patient age, patient's knowledge of partner's trait status, and having prior children were not predictive. A questionnaire with any unanswered item was omitted.

Table 4**Predictors of Patient's Postcounseling Knowledge: Multivariate Analysis**

Predictor	P Value
Knowledge of hemoglobinopathy manifestations (N = 265):	
Knew before this identification that she had trait.....	<.003
More years of education	<.009
Younger	<.007
Knew before counseling that partner had trait	<.036
Knowledge of genetics (N = 264):	
More years of education	<.004
Younger	<.001
Knowledge of prenatal diagnosis (N = 264):	
Knew before this identification that she had trait.....	<.006
More years of education	<.03
No prior children	<.05

NOTE. — For each type of postcounseling knowledge listed, a multiple regression analysis was performed to assess the predictive value of six factors—namely, patient age, years of education, gestational age, patient's precounseling knowledge of her trait status, patient's precounseling knowledge of her partner's trait status, and having prior children. The factors making a significant contribution are listed in their order of entry. A questionnaire with any unanswered item was omitted.

the patient who previously had been aware of her carrier status learns more than women not previously aware may be due in part to the fact that a previously aware woman was less likely to come for counseling (see preceding paper in this issue); thus, among these previously aware, the counseled group may have been more highly self-selected for interest in obtaining information. Finally, the fact that the woman who knows that the baby's father is also a carrier learns more is consistent with her recognition that her baby is indeed at risk.

Our previous comparison of methods for presenting genetic information demonstrated that, for nonpregnant adults identified as having beta-thalassemia trait, presentation by videotape was as effective as a presen-

tation in person, in terms of immediate and late recall and of patient acceptability (Rowley et al. 1984). For purposes of presenting factual material during genetic counseling, a videotape presentation has four advantages. First, the dramatization employed attracts and holds the counselee's interest. Second, a videotape can provide a role model demonstrating the appropriate level of concern and illustrating how the information presented can be useful. Third, a videotape presentation can ensure completeness and uniformity of presentation. Finally, the videotape saves professional time and thus may be useful in the busy practitioner's office.

In planning this study, we were concerned that, because of the anxiety aroused, pregnancy might be an

unsuitable time for carrier detection. Therefore, we initially measured anxiety state before and after counseling, by using a brief questionnaire (Spielberger et al. 1968). However, little anxiety was reflected in the initial series of patients, and this testing was discontinued.

Our experience in this study has prompted us to develop two kinds of materials for prenatal providers offering hemoglobinopathy screening. One kind, intended for the provider, is a manual summarizing the manifestations, diagnosis, and treatment of hemoglobinopathies. The other kind is a pair of instructional booklets, one for sickle cell trait and one for beta-thalassemia trait, which present, in pictorial form suitable for the provider to review with his patient page-by-page, the information listed in table 1.

One method of assessing learning is determining whether subsequent attitudes and behavior are altered. Three types of follow-up data were obtained about counselees in this study. First, 55% of those counseled had their partner tested, and, of at-risk couples offered prenatal diagnosis, 47% wanted it, as detailed in the preceding paper in this issue (Rowley et al. 1991). Second, for patients offered prenatal diagnosis, retention of the information provided at counseling was ensured during a follow-up visit to their home, after conclusion of the pregnancy; this information will be reported separately. Third, when a counseled patient was reidentified in a subsequent pregnancy, we had an opportunity to assess longer-term retention. Generally these patients remembered the name of their condition and that a fetus of theirs might be at risk for a serious blood disease. However, some did not realize that, to determine the risk to a fetus, each different partner required testing, a fact which apparently requires emphasis at the initial counseling session.

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