Genetic Counseling in Sickle Cell Anemia: Experiences with Couples at Risk

FLORENCE NEAL-COOPER, MS
ROBERT B. SCOTT, MD

Ms. Neal-Cooper is Instructor in Human Genetics and Dr. Scott is Professor of Medicine at the Medical College of Virginia, Virginia Commonwealth University, Richmond. An earlier version of this paper was presented at the annual meeting of the Sickle Cell Centers, Boston, MA, on April 18, 1986.

Tearsheet requests to Florence Neal-Cooper, Virginia Sickle Cell Anemia Awareness Program, Box 158, Medical College of Virginia, Richmond, VA 23298.

Synopsis

Beginning in 1970, a cohort of 74 sickle trait-carrying couples was identified who risked producing children with sickle cell anemia or other serious hemoglobinopathies. They were counseled concerning the disease and their risk, and their initial reactions, their stated intentions concerning birth control and childbearing, and their subsequent childbearing histories were documented.

Initial responses to the risk information varied widely, and stated intentions of birth control or childbearing did not accurately predict subsequent childbearing. Among 25 couples for whom there were complete childbearing data, there were 31 pregnancies with 13 affected children prior to counseling. After counseling, there were 25 pregnancies resulting in 10 affected children. The majority of the subsequent pregnancies occurred in the group who had not borne children or an affected child before counseling. In young couples, concern for producing a child with sickle cell anemia is often offset by a strong desire to have children regardless of risk.

SICKLE CELL ANEMIA presently occurs in approximately 1 in 500 births among blacks in the United States. Parents frequently learn of their risk of bearing affected children only after a child is born with this severe illness. With the availability of an inexpensive and specific test (electrophoresis of hemoglobin) for the asymptomatic carriers of sickle trait, interest developed in mass screening programs to detect trait carriers prior to childbearing so that, through counseling and education, they might have the opportunity of making informed choices—whether or not to have children and risk producing children with sickle cell anemia.

A number of such screening, education, and counseling programs have been established in recent years, many federally funded initially. Most of these programs were aimed at persons who had not reached the childbearing years, and identification and followup of couples composed of two carriers of the sickle trait (risk couples) was not stressed.

Efforts were made from the beginning of the screening program at the Medical College of Virginia to identify couples at risk in order to offer counseling to enable them to make their own judgments about future childbearing. Testing premaritally or prenatally is not an ideal time to screen. However, the program offered the service to persons who had not had the opportunity to benefit previously. A number of couples at risk were identified, and provided counseling, screening, and education. Because these couples were identified in the early years of new public and professional interest in hemoglobinopathies, we documented their initial responses to the knowledge of their risks and collected information concerning their subsequent childbearing after education and counseling about their risk status.

Methods

The Virginia Sickle Cell Anemia Awareness Program (VaSCAP) is a program of the Departments of Medicine and Human Genetics at the Medical College of Virginia. Although most persons screened for hemoglobin genes by VaSCAP have been young persons who have not reached their peak childbearing years, premarital testing was a part of the program's effort in its early phases. Legislation in the State of Virginia was enacted in 1972 to encourage physicians to offer voluntary sickle testing at the premarital serology examination. Thus, the staff of VaSCAP was able

to reach physicians and health facilities that were heavily involved in premarital testing and obtained more than 12,000 such samples for testing. In addition, samples from persons who attended prenatal or family planning clinics were obtained and, when a test was positive, the test was offered to the prospective father.

Screening tests consisted of hemoglobin electrophoresis on cellulose acetate and a confirmatory sickling test (Sickledex) on all samples which migrated like hemoglobin S or hemoglobin C.

In all instances, the risk couples were contacted by counselors and encouraged to take part in education and counseling. The probabilities of producing children with either sickle trait or sickle cell anemia were discussed, and information was given concerning the clinical features of sickle cell anemia as distinguished from sickle trait. The education and counseling effort was carefully designed to be nondirective and noncoercive.

The responses to the counseling information were assessed by the counselor; they included expressed and apparent anxiety, level of understanding, and the couple's initial plans vis-a-vis pregnancies. At the conclusion of the 60-minute counseling session, a brief written posttest was administered to assess the degree of retention of the salient facts by the couple. The results were tabulated as excellent, very good, average, fair, and poor. Both members of only two couples scored fair or poor, and one member of a third couple scored fair. The remaining couples were scored average or above in retention.

Additional demographic information was collected from the counselees, and they were urged to take part in a followup collection of data.

Followup sessions with each couple were attempted every 2 years. Home visits were scheduled, and a followup questionnaire was administered. In the questionnaire their understanding of the salient facts of heredity of the sickle trait was tested, and the previous educational content was reinforced as needed. In addition to collecting data concerning pregnancies, the counselor recorded individual responses to the knowledge of the potential risk, the degree of anxiety experienced, and the impact, if any, that this anxiety had on their marital relationship.

Results

Between 1970 and 1982, a total of 74 couples at risk were detected. Of these, three had genotypes AC/AC and were excluded since the risk of homozygous C disease was not considered to be on the same order of severity as the severe sickling syndromes. Also excluded were 11 other couples for whom childbearing was not possible because of tubal ligation, hysterectomy, or menopause. Twelve couples were excluded because they were no longer together after detection of the sickling trait, and 13 couples moved away or were not located after detection. The remaining 35 couples were available for evaluation.

The genotypes of these risk couples were AS/AS for 25, AS/AC for 9, and 1 couple was AS/A beta thalassemia. Twenty-two (63 percent) of the couples were detected by premarital blood testing, three (9 percent) were found in prenatal or family planning clinics, eight (23 percent) were found after pediatricians had referred children with a hemoglobinopathy, and two (6 percent) were found at health fairs. Followup periods ranged from 1 to 120 months, with a median of 48 months.

At the time of detection, the median age for the men (23.5 years) was slightly older than that for the women (21 years). Ages of the women ranged from 15 to 45 years, and the ages of the men ranged from 16 to 45 years.

The median educational level of the women was completion of the 12th grade; the range was from completion of 8th grade to graduate level. For the men, the grade level achievement ranged from 3rd grade to graduate school with the median completion of 12th grade. Of the 33 women with grade levels recorded, only 4 were at the ninth grade level or below. Among the men with grade levels recorded, only 5 were at the ninth grade or below.

Socioeconomic status was measured roughly by dividing the couples into five broad groups. Those who performed primarily manual work without supervisory roles were considered "blue collar." "White collar" work included office workers as well as a few with managerial or professional roles. Other categories included students, those who did not work outside the home (homemaker), and unemployed.

Socioeconomic status, as measured by occupational status at the time of detection, was varied. Of the women, 4 of the 33 responding had "white collar" occupations, 10 were "blue collar," 15 were not employed outside the home, and 4 were students. Among the men, 5 of the 32 responding held "white collar" jobs, 21 were "blue collar" workers, 5 were students, and 1 was unemployed.

After counseling, eight couples (23 percent) were uncertain what impact this new information would

Pregnancy history before and after genetic counseling of 25 couples with both members carriers of the sickle cell trait

Reproductive history	Number of couples	Before counseling		After counseling		
		Pregnancies	Affected children	Couples with pregnancies	Pregnancies	Affected children
No previous pregnancy	11	0	0	10 of 11	16	7
Previous pregnancy:						
No affected child	5	7	0	4 of 5	4	1
Affected child	9	24	13	4 of 9	5	2
Total	25	31	13	18 of 25	25	10

have on their future childbearing intentions. Fourteen couples (40 percent) did *not* believe that the information concerning their risk of bearing children with sickle cell anemia would have any effect on their future childbearing plans. The remaining 13 couples (37 percent) believed that the information about their risk would have an impact on their future childbearing.

Of the 35 couples responding, 21 (60 percent) stated that they planned to use some method of birth control. Of the 13 couples who stated that the risk information could affect their childbearing plans, 3 did not plan to use birth control. Among 14 couples whose childbearing plans were *not* influenced by the risk information, 9 (62 percent) were *not* using birth control. Plans to use birth control were reported by 50 percent of the women with 9th grade education or less, 55 percent of those with high school education, and 71 percent of those with college education. Of those women over the age of 24 years, 9 (82 percent) claimed to use birth control.

Prior to learning that they were at risk, 19 of the 35 couples had experienced at least one pregnancy. These couples had 41 pregnancies among them and had 14 offspring with serious hemoglobinopathies. One couple had 11 offspring and 5 affected children, but they had no subsequent pregnancies during the study period.

The ultimate impact on childbearing after learning of their risk is known for 25 couples during the study period. These results are shown in the table. Eighteen of the 25 couples experienced at least one pregnancy following counseling.

Of the 11 couples who had no children before learning of their risk, 10 subsequently experienced at least one pregnancy. There was a total of 16 pregnancies in this group, and 7 of these children had a significant hemoglobinopathy.

Among five couples who had at least one child previously, but none with a serious hemoglobinopathy, four women subsequently became pregnant. This group produced a total of four children, one of whom had a serious hemoglobinopathy.

Of the nine couples with children born before entering the study and one or more child with a serious hemoglobinopathy, only four couples had pregnancies following education and counseling. This resulted in five offspring and two sickle cell anemia patients.

These 25 couples produced 13 offspring with serious hemoglobinopathies prior to learning of their risk, and 10 affected offspring after counseling. Seven of the 10 were born to couples who had no previous children.

Discussion

Screening of couples at risk of producing children with serious hemoglobinopathies is a relatively recent development. The Virginia Sickle Cell Anemia Awareness Program began identifying such couples as early as 1970. Ideally, a screening program to inform people about childbearing risks should be performed well in advance of childbearing age, or at least prior to choosing mates. Nevertheless, in 1970, there had been no previous screening, and it was important at that stage in the implementation of screening programs to try to identify couples at the time of marriage so that they might have the opportunity of making informed decisions about childbearing.

As a result of this early effort, the VaSCAP screening program has identified perhaps the largest cohort of such couples presently known and followed for any length of time. These couples, then, represent a group who entered childbearing age without the benefits of an in-school education concerning the sickle cell disorders. Awareness of the existence of sickle cell anemia, and certainly an understanding of its hereditary nature, were not widespread, as documented in our 1968 survey of the general public (*1*).

In the initial education and counseling sessions,

the variety of individual responses was striking. The majority appropriately expressed certain anxieties upon learning of the possibility of producing ill children. Allaying these anxieties was an important concern of the counselors. In the subsequent followup there were no known instances of serious psychological sequelae, although whether counseling was responsible for this absence cannot be determined from the data.

Initial reactions ran the gamut from essential disregard of the risk information to intense interest and concern. One couple felt that "fate" or "God's will" would determine whether or not they had children with sickle cell anemia. There were a few instances in which the husband was unaware of the use of birth control by the wife. One husband expressed a belief that all healthy wives should have babies, regardless of the risk of producing sick children. One husband refused to consider the option of adopting children, since he had sired children in a previous marriage and they were all healthy. There was at least one instance of paternal pressure on the couple to produce grandchildren despite the risk.

The childbearing history of this group subsequent to counseling shows that of 25 with adequate data on childbearing, 13 had experienced at least one pregnancy at the time of followup (see table). Stated intentions to use family planning seemed to have little bearing on future pregnancy. Pregnancies occurred in two couples who initially felt that their childbearing would be influenced by their risk status, and 19 pregnancies occurred in those who felt uninfluenced by the risk information.

The experience with this group of couples in the first decade of widespread screening and education may or may not be typical of responses among those who learn about sickle cell anemia and are tested at a younger age, well in advance of childbearing or choosing mates. These experiences do emphasize the intense desire of young couples to have children, often despite known risks of illness in the offspring. This fact, plus the relative ineffectiveness of family planning intentions, suggests that many couples will not greatly limit family size in an effort to minimize the chance of producing a child with sickle cell anemia.

Studies of genetic counseling of risk couples involved with other serious heritable diseases have demonstrated a relative lack of enthusiasm for limiting family size despite the childbearing risk (*2,3,5*). The relative lack of clear understanding of the inheritance of recessive traits and poor

'Stated intentions to use family planning seemed to have little bearing on future pregnancy. Pregnancies occurred in two couples who initially felt that their childbearing would be influenced by their risk status, and 19 pregnancies occurred in those who felt uninfluenced by the risk information.'

comprehension of the concepts of probability are common in the general public (*4*). A controlled study of parents with one child with Down's syndrome showed that their subsequent reproduction behavior was not significantly different whether the couples were counseled or not (*5*).

In regard to Tay-Sachs disease screening, the major emphasis has been on prenatal diagnosis of affected fetuses and selective termination of these pregnancies (*6*). In the Ashkenazi Jewish population this program has gained widespread acceptance and resulted in prevention of many instances of the disease. Some have advocated that similar emphasis be placed on prenatal screening of parents for sickling, followed by amniocentesis for diagnosis of the fetus in lieu of mass screening programs at an earlier age (*7*).

One couple in our study group utilized prenatal diagnosis on three occasions. In the first two pregnancies, an affected fetus was aborted. In the third pregnancy, prenatal testing was negative, and a healthy child was delivered.

One particularly encouraging impression which we developed during the study was that since screening, education, and counseling have become available, a more positive attitude towards consideration of inherited illness, especially sickling, can now be noted among blacks. In the early phases of the screening programs, there was intense sensitivity concerning an inherited abnormality among blacks. With far better public understanding of the widespread occurrence of possibly deleterious genes among all races and ethnic groups, the subject of sickle cell genes has been put into a more positive perspective.

As the public becomes more aware of various inherited health problems, and as screening tests for carriers of recessive genes become more commonplace, screening programs for sickling should become less separately identified and more fre-

quently a part of broader genetic programs for the entire public. This change will have the beneficial effect of not appearing to single out one ethnic group for special attention. With the recognition that significant numbers of patients with hereditary disorders such as sickling continue to be born, continued emphasis on research and support services leading to better patient care remains an important priority.

References

1. Lane, J. C., and Scott, R. B.: Awareness of sickle cell anemia among Negroes of Richmond, Va. Public Health Rep 84: 949-953, November 1969.
2. Steele, M. W.: Lessons from the American Tay-Sachs programme. Lancet 2: 914, Oct. 25, 1980.
3. Burns, J. K., Azen, C. G., Rouse, B., and Vespa, H.: Impact of PKU on the reproductive patterns in collaborative study families. Am J Med Genet 19: 515-524, November 1984.
4. Evers-Kieboom, G., and van den Berghe, H.: Impact of genetic counseling: A review of published followup studies. Clin Genetics 15: 465-474, June 1979.
5. Oetting, L. A., and Steele, M. W.: A controlled retrospective follow-up study of the impact of genetic counseling on parental reproduction following the birth of a Down syndrome child. Clin Genetics 21: 7-13, January 1982.
6. Kaback, M. M.: Heterozygote screening and prenatal diagnosis in Tay-Sachs disease: A worldwide update. *In* Lysosomes and lysosomal storage diseases, J. W. Callahan and J. A. Lowden, editors. Raven Press, New York, 1981, pp. 331-342.
7. Rucknagel, D. L.: A decade of screening in the hemoglobinopathies: Is a national program to prevent sickle cell anemia possible? Am J Pediatr Hematol Oncol 5: 373-377, winter 1983.

Factors Affecting the Supply of Minority Physicians in 2000

CLAY E. SIMPSON, Jr., PhD
REMY ARONOFF, MSc

The authors are or were with the Division of Disadvantaged Assistance, Health Resources and Services Administration, (HRSA), Public Health Service. Dr. Simpson is the Division Director. Mr. Aronoff is now Chief, Analysis and Operations Branch, Division of Organ Transplantation in the Bureau of Maternal and Child Health and Resources Development, HRSA.

Tearsheet requests to Division of Disadvantaged Assistance, HRSA, Rm. 8A-09, Parklawn Bldg., 5600 Fishers Lane, Rockville, MD 20857.

Synopsis

There was a substantial increase in the number of black and Hispanic physicians between 1970 and 1985. During the next 15 years, 1985 to 2000, the increase is projected to continue. The factors that will determine the size of the increase and the changes in physician to population ratios include black and Hispanic population increases, medical school costs, availability of student support, minority enrollment in undergraduate schools and the pool of these students who will be applicants to medical school, attrition during medical school, competition from other professions for talented minority students, and the effects of intervention programs such as the Health Careers Opportunity Program. The most likely outcome would seem to be that the increases in black and Hispanic physicians will continue to 2000 at 1985 levels because the data show neither strong positive nor strong negative net influences for the factors examined.

THE MOST RECENT PROJECTIONS of minority health professionals by the Bureau of Health Professions in the Health Resources and Services Administration show an 83 percent increase in black physicians and a 35 percent increase in Hispanic physicians between 1985 and 2000 (*1*). While these projected increases are sizable, the factors that can affect them are themselves subject to significant change between now and the end of the century. Those factors include the increase in the population of blacks and Hispanics, costs of a medical education, enrollment in undergraduate and medical schools, the minority student pool of potential medical school applicants, competition for minority students from other occupations, attrition during medical school, and intervention programs intended to recruit and prepare minority medical students for the health professions. In this article we will examine these factors and relate them and