age separation of all urinary amino acids to detect any excesses. Although the yield of newly diagnosed inborn errors by this screening program is small, those few cases detected are important because they may lead to a specific diagnosis that permits a prognosis, a recurrence risk for other family members, and counseling about the possibility of treatment or prenatal diagnosis for future pregnancies.

Clearly, genetic screening programs are an important component of the primary care physician's efforts in preventive medicine. Participation in and support of these programs is therefore strongly urged.

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The Virginia Sickle Cell Anemia Awareness Program (VaSCAP): Education, Screening, and Counseling

FLORENCE N. COOPER AND ROBERT B. SCOTT, M.D., *Department of Medicine*

In 1968, a program of screening for sickle trait carriers was begun as part of the work of the Hematology Division, Department of Medicine, at the Medical College of Virginia. It was felt that sickle cell anemia was more of a public health problem than was generally recognized, and in addition to instituting screening and education programs, data were collected to document the relative neglect of the problem.

A survey of the City of Richmond was conducted to ascertain the level of awareness among

Correspondence and reprint requests to Robert B. Scott, M.D., Box 214, Richmond, Virginia 23298.

black adults of sickle cell anemia, a severe chronic disease affecting one of every 500 black babies. The results[1] showed that only 3 of every 10 black adults had ever heard of this disease, and of those who recognized it, few understood the nature of the illness. Additional background information was collected which further showed the lack of professional and public understanding of the importance of sickle cell anemia as a public health problem. These data,[2,3] which attracted widespread attention, showed that sickle cell anemia research received far less support than other much less common chronic illnesses.

Public attention was also focused on sickle cell anemia by publicity concerning a new "test tube" test for sickling, and claims for therapeutic benefit from intravenous urea solutions. The heightened interest in sickle cell disease finally resulted in the Sickle Cell Anemia Prevention Act of 1972, which included a series of sickle cell research and treatment centers plus screening, education, and counseling clinics throughout the nation. At that time VaSCAP competed successfully for a contract under the national program, and expanded from a small screening effort to a larger, well-staffed clinic which could extend its effects state-wide.

Although the ability to detect healthy carriers of the recessive sickle trait had existed for years, no widespread screening and genetic counseling had taken place prior to these new programs. Another factor which made mass screening economically feasible was the development of rapid, simple instruments for performing hemoglobin electrophoresis. Electrophoresis has distinct advantages over the classical sickle cell "prep" or the newer test tube tests since it can distinguish sickle trait from the homozygous state, sickle cell anemia, and can also detect numerous other hemoglobin variants.

The purpose of screening for abnormal hemoglobins is to give young people an opportunity, not previously available, to know in advance of childbearing whether they run the risk of having children with sickle cell anemia. The frequency of sickle trait is about 8% of the black population, or 1 in every 12 blacks. The chance, then, that both potential parents in a couple are trait carriers is 1 in 144; thus slightly less than 1% of black couples are composed of two trait carriers and are at risk for bearing children with sickle cell anemia. The chance is 1 in 4 *with each pregnancy* that a child will be born with this incurable illness.

Screening should not be performed without adequate prior *education.* To implement this principle, VaSCAP has developed a number of audiovisual programs and written materials suitable for a wide variety of audiences. Included in these are the *Sickle Cell Anemia Fact Book,*[4] a booklet for teachers, nurses, and other professionals, and *Our Sickle Cell Story Fact Book,*[5] for elementary and middle school students. These, as well as brochures, posters, and fact sheets are distributed through the Virginia State Department of Health, to many areas of Virginia and throughout the nation. Since its inception, VaSCAP has provided educational sessions to over 200,000 individuals in groups of various sizes. It has also reached many more through programs, spot announcements, and structured educational models for specific target groups with the result that screening for hemoglobin genes has become an acceptable procedure among the general population. At present over 42,000 persons have been screened by hemoglobin electrophoresis and 3,951 have been found to have hemoglobin variants.

The ultimate result of education and screening is *counseling* those found to have sickle or other mutant hemoglobin genes. Counseling is aimed at educating the individual so that he or she can know the nature of the sickle gene, understand what sickle cell anemia is, understand the probabilities of transmitting the genes, and then be able to make informed decisions about childbearing.There is a set of slides available which has been developed by the VaSCAP staff. These explain the probabilities of inheritance for each specific hemoglobin variant. Individuals newly diagnosed with sickle cell anemia or one of its variants have been referred to a family physician or to the Hematology Clinic at MCV.

Approximately 8% of Virginia blacks have been found to have sickle cell trait, and 2% carry hemoglobin C trait. About 0.3% have the gene for hereditary persistence of fetal hemoglobin (HPFH), which is a gene mutation *not* associated with any disease. In addition, a large number of less common mutant hemoglobin genes have been recognized.

Though the alkaline electrophoresis test is the primary method used for the detection of hemoglobin genes, there are other methods which may be required in order to provide more conclusive results. The laboratory of the VaSCAP clinic provides both alkaline and acid or citrate agar electrophoresis. A_2 mini-columns for hemoglobin A_2 quantitations are very useful in detecting the Beta thalassemias; fetal hemoglobin staining aids in determining the presence

of HPFH. Family studies are necessary in certain cases and the clinic has screened as many as four generations in some families.

All of the services have been provided without charge to the public or physicians for whom diagnostic testing has been performed. The emphasis has been on testing persons in or approaching the childbearing age.

Education programs have been carried out in most high schools and colleges in central Virginia and the Tidewater area. Church groups and community organizations regularly request programs, many of which are provided in evenings or on weekends. An important part of the educational effort has been to provide workshops to train public health nurses in genetic counseling relating to hemoglobinopathies. State legislstion has provided funds for nurses to be designated as genetic counselors in all of the Health Department clinics in the State. VaSCAP has also worked closely with local health departments in providing consultations concerning diagnosis and counseling of individuals with unusual hemoglobin variants.

The number of individuals served by the VaSCAP clinic is a tangible measure of its performance, but it does not reflect all of the benefits to the public and health professionals. The tragic lack of public awareness of the importance of sickle cell anemia that characterized the beginnings of this program has disappeared. For health professionals, especially at MCV, the subject of hemoglobinopathies has been emphasized repeatedly, and the level of interest and understanding of the disease and its variants have risen considerably. Of particular significance are the improved diagnostic facilities provided by VaSCAP clinics which have made it possible to stress the more rigorous modern standard of diagnosis for hemoglobin disorders.[6] Perhaps most important to the field of genetics generally has been the extensive practical experience gained by the first mass screening efforts for a heritable disease. The ease with which recessive hemoglobin genes are detected made it the first such program to be implemented; as additional gene tests are developed, other genetic diseases will be the targets of similar programs.

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