

Sickle Selections

a quarterly newsletter from the University of Rochester Sickle Cell Program

July 1997

STRONG SICKLE PROGRAM . . . what we're about

COUNSELING

Dr. Peter T. Rowley, medical geneticist, receives notification from NY State Newborn Screening in Albany of all babies born in a 9 county region* who test positive for an abnormal hemoglobin. His staff contacts the pediatrician of newborns who have a *symptomatic hemoglobinopathy* to explain the need for repeat testing and our role in counseling the family. After the result of the repeat sample is received, our genetic counselor and the nurse from the Sickle Cell Clinic make a home visit to the family. During the home visit we explain the hereditary basis of the baby's disease and the risks of a hemoglobinopathy in future children. We explain the symptoms of the disease, the need

to contact the physician at the first sign of fever or anemia, and the importance of prophylactic penicillin. We recommend hemoglobinopathy testing of the parents to determine the risk of disease for their future children and to make an accurate diagnosis for the baby. Written materials about the baby's diagnosis are provided to the family.

We contact all mothers of *newborns with trait* by phone or letter to inform them of their baby's trait status. If the mother was screened through our Prenatal Hemoglobinopathy Screening Program we discuss her results with her. (On average we prenatally screen 40% of these mothers). We explain the inheritance of hemoglobin type and what trait status means for the baby and the implications for future children. We offer testing to parents not previously tested. On average, 68% of the mothers we contact are unaware of their newborn's trait status.

* Chemung, Monroe, Livingston, Ontario, Schuyler, Seneca, Stueben, Wayne, Yates counties.

LABORATORY TESTING

We screen for all detectable abnormal hemoglobins by cellulose acetate electrophoresis and for b-thalassemia trait by measuring hemoglobin A₂ by column chromatography. We confirm sickle hemoglobin by a solubility test and other hemoglobins by citrate agar electrophoresis. We perform other tests as necessary.

SICKLE CELL CLINIC

Dr. Norma B. Lerner, pediatric hematologist, is the new director of pediatric sickle cell disease clinical services. Patricia K. Lamarche, R.N., P.N.P. will be joining Susan Weidner, R.N., P.N.P. as of August 1997 to provide additional nursing coverage. Plans are currently being made for the restructuring of services and the provision of innovative approaches to care. These will be described soon in a separate mailing.

As in the past, outpatient services are offered for the comprehensive care of patients with sickle cell disease and related hemoglobinopathies. The intent is to share regularly scheduled visits with the general pediatrician so that the patient may benefit from the expertise of both primary provider and the physician (and nurse) specialist. Physical examination and age appropriate tests are performed at each appointment and counseling regarding preventive care is provided. A social worker and educational liaison are always available. Specialty evaluations in the fields of cardiology, pulmonary disease, orthopedics, etc. are arranged as needed with the Strong Memorial Hospital staff.

Those children who require regular transfusion therapy, most usually those who have experienced cerebrovascular accidents, receive blood in the treatment center of the ambulatory unit. Recent expansion of center hours to 8 pm on Mondays and Wednesdays have made transfusions available at more convenient times for some families.

Inpatient and outpatient acute care services are provided to children with sickle cell disease at Strong Memorial Hospital where a pediatric hematologist is always available for consultation.

Communication with local providers is encouraged and coordinated; excellent care is the goal of all personnel.

UNIVERSITY OF
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MEDICAL CENTER
Strong Sickle Program
Offers Help

Regarding treatment contact:

Dr. Norma Lerner or Pat Lamarche R.N., P.N.P. Department of Pediatrics 275-2981
Dr. Karen Kaplan, Department of Medicine 275-3761

Regarding laboratory diagnosis, newborn screening and genetic counseling, contact:

Dr. Peter Rowley, Sandra LaBella or Starlene Loader, Division of Genetics 275-4602

Visit our website: www.urmc.rochester.edu/genetics