

APPENDIX I

Hemoglobinopathy Screening and Follow-Up

Sickle Cell Testing: Hemoglobinopathy Screening and Follow-up

The hemoglobinopathies are genetic disorders that affect the production and function of hemoglobin molecules. These disorders include sickle cell disease and trait, hemoglobin C, E and other rare variants as well as the thalassemias. Fewer than 1000 Americans have beta thalassemia major, but sizable numbers of Italian Americans, Greek Americans, and immigrants from Southeast Asia carry the genetic trait.

I. Incidence of Sickle Cell Disease by Ethnicity

- In the United States, sickle cell disease affects approximately 72,000 people. The vast majority of people with sickle cell disease are of sub-Saharan African descent. In the United States, this accounts for about 90% of all sickle cell disease patients.
- In the United States, the incidence of sickle cell disease is approximately:
 - 1 in every 350 African-American births
 - 1 in 58,000 Caucasians
 - 1 in every 1000 to 1400 Hispanic American births.
 - 1 in 11,500 Asians (Sickle/E Disease)
 - 1 in 2,700 Native Americans
- Approximately 2 million Americans, or 1 in 12 African Americans, carry the sickle cell trait.

In the United States, sickle cell disease is also found in a small percentage of individuals of Caribbean, South and Central America and the Mediterranean ancestry.

SICKLE CELL TESTING IS NOT A ROUTINE SCREEN

- **In adult populations, screening is useful primarily for providing preconception and prenatal genetic counseling to carriers.**
- **Blood for Hemoglobin Electrophoresis should NOT be drawn unless requested by a client after education regarding the purpose for screening.**

II. Purpose for screening

- **Diagnosis of disease in adult.**

The purpose for screening for hemoglobinopathies is to identify any hemoglobin trait or disease that may affect the health of the client.

- **Couple “At-Risk” Status**

Screening tests may also be done on young adults who wish to determine whether or not they and/or their partner carry the sickle cell trait prior to beginning a family.

- **Prenatal Diagnosis/Genetic Counseling**

If a pregnant woman and her partner are identified as “At-Risk” for having a child with sickle cell disease, they can choose to have their unborn babies tested for the presence of sickle cell trait or disease.

- **Newborn Screening**

In Virginia, newborn testing for the identification of sickle cell disease began in July 1989. All babies, regardless of race are screened for sickle cell disease and other hemoglobinopathies. Currently 44 states, the District of Columbia and Puerto Rico screen for sickle cell disease.

Who should be provided education about the importance of screening for hemoglobin variants?

- All clients

Who should be provided screening?

- All clients in the high-risk populations who request the testing and meet the eligibility requirements. (See Tables)
- Partners of pregnant woman identified with a hemoglobin variant.

Who should not be screened through the health department?

- Anyone who indicates that they have been previously tested
- Minors and children enrolling in Head Start

Clients who indicate that they have been tested but do not have a copy of their test results should be encouraged to contact their provider to obtain a copy.

III. Criteria for Hemoglobinopathy Screening through the Division of Consolidated Laboratory Services

Solubility testing must not be used as a primary screening method for the detection of hemoglobin variants.

Hemoglobin Electrophoresis will be provided for individuals meeting the following criteria:

- a) Family planning and maternity patients who have had financial screening and are designated as Income A.
- b) Parents and siblings of infants identified with sickle cell disease or other hemoglobinopathy through Virginia’s Newborn Screening Program.
- c) Partners of women identified with a hemoglobinopathy through prenatal screening.
- d) Limited and planned community health education and screening programs targeted at high risk populations in the childbearing ages.

VASCAP will provide:

- a) Staff development and materials for nurses providing education and follow-up counseling to clients identified with a hemoglobinopathy.
- b) Hemoglobin identification cards and educational materials for all individuals identified with a hemoglobin variant through our screening contract with DCLS.

Contact Information for Sickle Cell Services

The Virginia Sickle Cell Awareness Program (VASCAP)

The Virginia Sickle Cell Awareness Program is designed to offer access to current and accurate information regarding sickle cell disorders and other hemoglobin variants through both community partners and collaboration with Family Planning and Maternity clinics operating through Virginia Department of Health.

Program Contact Person:

Jene Radcliffe-Shipman, Program Manager

Virginia Department of Health

109 Governor Street – 825E

Richmond, Virginia 23219

Phone: (804) 864-7769 **FAX:** (804) 864-7771

E-mail: jene.radcliffe-shipman@vdh.virginia.gov

Patient Educational Materials can be downloaded from our website:

<http://fhsweb/sicklecell/>

Table 1: Common Hemoglobin Types

Hemoglobin	Incidence	Race/Ethnicity
A/S Sickle Trait	8-10%	African American
	2%	Hispanic Italians, East Indians, Saudi Arabians
A/C C Trait	2-5%	African Americans
	20-25%	West Africans (Ghana)
A/E E Trait	30-40%	South East Asians
Beta Thalassemia	5%	Mediterranean
	5%	Asian
	4%	East Indians
	2%	African Americans

Table 2: Incidence of sickle hemoglobin by race/ethnicity

Race/Ethnicity	Sickle Trait	All Types of SCD
African American	1:10	1:350
Asians		1: 11,5000 (SE)
Central and South American	1:183	1:45,622 (SS)
Hispanic/Americans		1:1000 to 1400 (Eastern States) 1:32,000 (Western States)
Native Americans		1:2,700
Caucasians of European Ancestry	1:625	1:58,000

