

## Hemoglobinopathy

Included in newborn screenings, all filter paper specimens are routinely tested for sickle cell disease and other hemoglobinopathies. **Sickle Cell disease** is characterized by the presence of abnormal red blood cells that do not transport oxygen efficiently and may become sickle shaped, causing anemia and pain due to occlusion of the blood vessels. Most commonly found in persons of African ancestry, Sickle cell disease also affects people of Mediterranean, Caribbean, South and Central American, Asian and Middle Eastern descent.

Other defects in globin-chain synthesis yield other hemoglobin variants. Some cause serious health problems such as hemolytic anemia, splenomegaly, sepsis, progressive retinopathy, pulmonary vasoocclusion, hydrops fetalis, stillbirth, and others only a mild anemia. The most common structural variants other than HbS are HbC, HbD and HbE (the most common quantitative variants), are the thalassemias. Geographic origins to be considered include equatorial West and North Africa, India, Pakistan, Afghanistan, Turkey, Iran, Egypt, Southeast Asia, India, Sri Lanka, the Philippines, Greece, Sicily.

As Maine's population grows more diverse and many of these geographic areas are represented, it is important to be aware of children who need screening for various hemoglobinopathies. With any hemoglobinopathy, genetic counseling is recommended primarily for parental education and to assess risk for any significant hemoglobin disease in future children.

### **Sickle Cell Disease:**

**Prevalence:** 3:1000

**Analytes Measured:** Hemoglobin Fractions  
FA, AF, A

**Reporting Ranges:** Absence of Hemoglobin A

**Feeding Effect:** None

**Timing Effect:** None unless transfused

Note: If infant has been transfused, repeat specimen should be obtained at 2 months post transfusion due to potential for transfused blood to mask hemoglobinopathies.

**Confirmation:** By Isoelectric Focusing and citrate agar electrophoresis.

**Treatment:** Referral made to the Maine Hemophilia Treatment Center and a Pediatric Hematologist. Prophylactic use of penicillin and treatment of symptoms. Treatment may vary for different hemoglobin defects.

**Comments:** Affected babies are more likely to have pain, strokes and life-threatening infections. Treatment should begin after diagnosis and by no later than 2 months of age. With screening, penicillin prophylaxis, more aggressive and careful management, early death and complications from stroke and infections have been greatly reduced.