**Section 661.390 Sickle Cell Disease/Trait and Other Hemoglobinopathies**

a) Interpretation of Results. Although the majority of infants affected by sickle cell disease/trait and other hemoglobinopathies will be identified by this screening, due to genetic variabilities and variations in health status, sample quality, and timing of sample collection, not all infants affected by the disorder may be identified. As with any laboratory test, false positive and false negative results are possible. Newborn screening test results are insufficient information on which to base diagnosis or treatment. Qualitative testing will determine the presence of various hemoglobins.

1) When hemoglobin F and hemoglobin S, but no hemoglobin A, are detected on the same sample, the Department will recommend referral to a designated medical specialist for follow-up and genetic counseling.

2) When hemoglobin F, hemoglobin S and hemoglobin C, but no hemoglobin A, are detected on the same sample, the Department will recommend referral to a designated medical specialist for follow-up and genetic counseling.

3) When hemoglobin F, hemoglobin A and hemoglobin C or hemoglobin F, hemoglobin A and hemoglobin S are detected on the same sample, the Department will recommend parental testing and genetic counseling by the attending physician or another qualified counselor.

4) When hemoglobin F and other hemoglobins, such as hemoglobin D, hemoglobin E or hemoglobin H (Bart's) are detected, the Department will recommend referral to a designated medical specialist for follow-up and genetic testing.

5) When hemoglobin A is detected as the predominant hemoglobin, and the blood spot sample was collected at less than two months of age, a written report will be sent to the submitter. The medical provider shall collect a repeat newborn screening blood spot sample at 120 days post-transfusion if the initial sample was collected post-transfusion.

b) Designation of Medical Specialist. In addition to the minimum qualifications set out in Section 661.230, medical specialists designated by the Department to follow-up on a screen positive for sickle cell disease/trait and other hemoglobinopathies shall have training in pediatric hematology and certification of special competence in pediatric hematology-oncology by the American Board of Pediatrics.

c) Diagnosis and Treatment. Medical management by a designated pediatric hematologist-oncologist is highly recommended. Antibiotic prophylaxis and immunization to prevent pneumococcal infections and treatment with hydroxyurea are currently the standard treatment after a designated medical specialist has made a definitive diagnosis of a sickling disease. Long-term follow-up of children with sickle cell disease/trait is necessary to assess growth and development.