PRACTICAL PEARL: HEMOGLOBIN ABNORMALITIES ON NEWBORN SCREEN

Introduction	• Newborn screening (NBS) for hemoglobinopathies allows identification of children with hemoglobin abnormalities associated with disease and initiation of referral and treatment. • A detailed review of the NBS program for hemoglobinopathies is available on the New England Pediatric Sickle Cell Consortium website: **https://nepscc.org/health-providers/screening/**
Initial Evaluation and Management by Primary Care	• NBS results are reported for hemoglobins in order of relative abundance, e.g., "FA" indicates abundance of hemoglobin F is greater than A (normal finding). • Hemoglobinopathies are identified when hemoglobins other than F and A are present. In sickle cell anemia, the relative abundance is important in determining trait versus disease. Sickle cell trait would demonstrate an FAS pattern; sickle cell disease demonstrates an FS pattern. • If patterns consistent with sickle cell disease (FS, FSA, FSC, FSV) are reported, the child should immediately be treated with prophylactic penicillin (125mg po BID) and referred to Pediatric Hematology. • Bart's hemoglobin, reported as FAB, reflects some degree of alpha-thalassemia; follow up is determined by family history and the frequency of the condition in certain ethnic groups.
When to Refer	• Hemoglobin patterns consistent with a form of sickle cell disease (FS, FSA, FSC, FSV).
How to Refer	• (413) 794-KIDS • Questions for the hematologist: (413) 794-9338
What to Expect From Baystate Children's Hospital Visit	• Confirmatory testing, consultation, treatment and follow-up care based on the diagnosis.

Authors: **Matthew Richardson, MD**
Pediatric Hematology/Oncology
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Contact: Baystatechildrenshospital@baystatehealth.org