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: <u>https://nepscc.org/health-providers/screening/</u>
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.

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