Randall Children's Hospital

Co-Management and Referral Guidelines

Initial Management of Abnormal Newborn Hemoglobinopathy Screening

Randall Children's Cancer and Blood Disorders Program

Phone: **503-276-9300**
Fax: **503-276-9351**

Introduction

- The hemoglobinopathies are a group of disorders associated with mutations in the alpha-globin and beta-globin genes. Two alpha and two beta-globin chains produce the normal adult hemoglobin molecule.
- Currently, all states require testing of newborns for hemoglobinopathies. Most newborn screening programs will detect hemoglobins F, S, A, C, E, D and Bart's. Less often, other hemoglobin variants may be detected. The hemoglobins present are reported in order of abundance.
- Hemoglobinopathies include sickling and non-sickling disorders.
  - **The sickling disorders** are found primarily in the African-American population and include Sickle Cell Anemia (Hgb SS), Hemoglobin SC disease (Hgb SC) and Sickle-Beta-Thalassemia (Hgb SA).
  - **Non-sickling hemoglobinopathies**, including the thalassemias, are found primarily in individuals of Mediterranean, Asian and African-American ancestry.
    - → Alpha-thalassemia results from loss of alpha-globin genes
      - → A single deletion is a silent mutation.
      - → Deletion of two genes is "alpha-thal trait" and may be asymptomatic or cause mild anemia; Bart's hemoglobin may be reported.
      - → Deletion of three genes causes Hgb H disease and may be described as "Elevated Bart's hemoglobin."
      - → Deletion of all four genes usually causes severe hydrops and is often fatal in utero.
    - → Beta-thalassemia results from underproduction of beta-chains due to abnormal beta-globin genes.
      - → A single abnormal beta-thalassemia gene is "beta-thal trait" and may be asymptomatic or cause mild anemia.
      - → Two abnormal beta-thalassemia genes can result in marked decrease of beta-chains or complete absence of beta-chains. This results in severe anemia and may require life-long transfusion support.

Evaluation and Management

- With any hemoglobinopathy detected on newborn screening, the family should be contacted with the results and this communication documented in the medical record.
- For the sickling disorders, a CBC and hemoglobin electrophoresis should be obtained for confirmation (either immediately or at the first visit to the pediatric hematologist).
- For the non-sickling disorders, including the "traits" and "carriers," the timing of a CBC and confirmatory hemoglobin electrophoresis is dependent on the potential blood disorder. (See tables on next page.)
- Evaluation of parents and siblings should also be performed if their hemoglobin pattern is unknown.

When to refer

- Refer any baby found to have any of the sickling disorders (see tables on next page) to the pediatric hematologist within one month of life. The American Academy of Pediatrics (AAP) recommends starting prophylactic antibiotics by 2 months of age.
- In general, those hemoglobinopathies that are "traits" or "carriers" do not require a referral, but we would be more than willing to see these patients for additional counseling, if so desired.

(continued)

Referral process

Randall Children's Cancer and Blood Disorders Program

Phone: **503-276-9300** or toll-free **1-877-KIDS-ONC (877-543-7663)**
Fax: **503-276-9351**

For urgent referrals, call Legacy One Call Consult & Transfer: 1-800-500-9111 to speak with the on-call pediatric hematologist/oncologist.

With all referrals, please fax pertinent lab results, unless visible via Epic Care Everywhere.

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Additional Resources

National Heart, Lung and Blood Institute: Sickle Cell Disease Guidelines
http://www.nhlbi.nih.gov/files/docs/guidelines/sc_mngt.pdf

Sickling hemoglobin disorders requiring expeditious referral (within one month)

Newborn Screen Results	Description	Testing
FS	Sickle Cell Anemia or Sickle-Beta-0 Thalassemia or Sickle with Hereditary Persistence of Fetal Hemoglobin	CBC with manual differential and hemoglobin electrophoresis immediately or at the first hematology clinic visit
FSC	Hemoglobin SC disease	
FSA	Sickle-Beta+ thalassemia	
FSV	Sickle with Hemoglobin Variant	
FSE/O or FSD/G	Sickle with indeterminate hemoglobin pattern; both indeterminate patterns indicate increased risk for sickling disorder	

Non-sickling hemoglobin disorders that require less urgent referral (within 1–3 months)

Newborn Screen Results	Description	Testing
FC	Hemoglobin C disease or Hemoglobin C-Beta-0 Thalassemia	CBC with manual differential and hemoglobin electrophoresis at 1–3 months of life or at the first hematology clinic visit
FE/O	Multiple possibilities of E, O and Beta-0 Thalassemia	
F	Hereditary persistence of fetal hemoglobin or Beta-Thalassemia Major	
FA Elevated Bart's	Possible Hemoglobin H disease	

Non-sickling hemoglobin disorders not necessarily needing a referral

Newborn Screen Results	Description	Testing and Referral
FAS	Sickle cell trait (carrier)	CBC with manual differential and hemoglobin electrophoresis should be obtained between 6 and 12 months of age. Contact hematology if necessary after confirmatory results are available.
FAC	Hemoglobin C trait (carrier)	
FAV, FAO/E, FAD/G	Carrier of hemoglobin variant including E, O, D or G	
FA Bart's	Probable alpha-thal "silent carrier" or alpha-thal "trait"	

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Find this and other co-management/referral guidelines online at: ***www.legacyhealth.org/randallguidelines***

RANDALL CHILDREN'S HOSPITAL
LEGACY EMANUEL