

Hemoglobinopathies

Use

Qualitative detection of normal and variant hemoglobins in blood specimens dried on filter paper used as an aid in the detection of sickle cell anemia, sickle C disease, S/beta-thalassemia, homozygous C disease, and variant hemoglobinopathies in newborns, children and adults.

Clinical Significance

Detecting hemoglobin variants is critically important for early diagnosis, clinical management, and genetic counseling of inherited hemoglobin disorders. Identification of variants, such as hemoglobin S, C, E (Hb S, Hb C, and Hb E) and others, allows for timely intervention to prevent or reduce disease-related complications, particularly in conditions like sickle cell disease where early treatment significantly improves outcomes. Detection also helps identify carriers and compound heterozygous states (e.g., Hb SC, Hb SE), supporting informed reproductive counseling and family risk assessment. In public health and newborn screening programs, early recognition of hemoglobin variants enables prompt referral, monitoring, and education, ultimately reducing morbidity, mortality, and long-term healthcare burden.

Methodology

Isoelectric focusing agarose gel electrophoresis

Specimen Type

- Newborns: Dried blood spots on ODH #450 Newborn Screening Form
- Adult/Child: Dried blood spots on ODH #485 Child/Adult Sickle Cell Screening Form

NBS and Child Adult Sickle Cell Screening forms can be ordered by contacting contact the OSDH Public Health Laboratory Client Services by calling 405-444-6494 or placing an order using the OSDH [Laboratory Supply Ordering System](#).

Minimum Volume/Size

- 5 filled circles (Newborn)
- 3 filled circles (Adult/Child)

Collection Instructions

- For newborns, see [Guidance for Collection of NBS Dried Blood Spots](#) (Specimen-specific Guidance).
- For adults/children, see [Guidance for Collection of Adult/Child Blood for Hemoglobin Analysis](#) (Specimen-specific Guidance). Acceptable persons for collection of adult/child blood analysis include females < 46 years of age, maternity or family planning individuals of any age, and males of any age.

Common Causes for Rejection

See [General Information on NBS Tests](#). Specimens improperly collected, processed or transported.

Shipping

See [General Information on NBS Tests](#).

Turn-around Time

- Within 5 working days of receipt (Newborn)
- Within 10 working days of receipt (Child/Adult)

Reference Range

- Hb FA (Newborn)
- Hb AA (Child Adult)

Reportable Results

- Within Normal Limits
- Outside Normal Limits
- Inconclusive

Interpretation

- Within Normal Limits
 - No Abnormal Bands Detected
- Outside Normal Limits
 - F Only: Submit new filter paper specimen at 2 months of age.
 - Consistent with transfusion: Submit new specimen at 4 months of age.
 - FA, Bart's: Recommend CBC with indices at 1 year of age; or at 3 months if of Asian descent.
 - FAS: Consistent with S trait. Submit new specimen at 4 months of age.
 - FAC: Consistent with C trait. Submit new specimen at 4 months of age.
 - FA, Other: Consistent with a trait. Submit new specimen at 4 months of age.
 - FS: Consistent with SS disease. Refer to pediatric hematologist for confirmatory testing.
 - FC: Consistent with CC disease. Refer to pediatric hematologist for confirmatory testing.
 - FSC: Consistent with SC disease. Refer to pediatric hematologist for confirmatory testing.
 - FSA: Consistent with S-thalassemia. Refer to pediatric hematologist for confirmatory testing.
 - FEO: Consistent with disease. Refer to pediatric hematologist for confirmatory testing.
 - FDG: Consistent with disease. Refer to pediatric hematologist for confirmatory testing.
 - F, Other: Consistent with disease. Refer to pediatric hematologist for confirmatory testing.
 - FAS, Barts: Consistent with S trait. Submit new specimen at 4 months of age. Recommend CBC with indices at 3 or 12 months of age as indicated.
 - FAC, Barts: Consistent with C trait. Submit new specimen at 4 months of age. Recommend CBC with indices at 3 or 12 months of age as indicated.
 - FA, Other, Barts: Consistent with a trait. Submit new specimen at 4 months of age. Recommend CBC with indices at 3 or 12 months of age as indicated.
 - A, Other: Family counseling recommended.
 - AS: Consistent with S-trait. Family counseling recommended.
 - AC: Consistent with C-trait. Family counseling recommended.
 - SAF: HGB - Consistent with S-beta thalassemia. Refer to pediatric hematologist for confirmatory testing.
- Inconclusive
 - Inconclusive. Submit new specimen as soon as possible.

Note: Any variant reported as "F, Other" or "FA, Other" will include an additional comment to convey location of the unidentified variant band, e.g., "Other Band in D/G Region" or "Other Band in E/O Region", etc.

Limitations/Interferences

- This is a screening testing only. A diagnostic procedure should be used to confirm the identity of variant hemoglobins.
- Older samples may yield "aging bands" due to decomposition and/or oxidation of hemoglobin.
- Isoelectric focusing can separate hemoglobin variants with isoelectric points (pI) that differ by 0.01 pH units or greater. Isoforms with the same pI migrate together; similar migrating variants may be missed.

- Specimens improperly collected, processed or transported may result in erroneous results.

CPT Code

83020-52

Notes

This test is approved for *in vitro* diagnostic use by the U.S. Food and Drug Administration.