

5.2 (old 3.13.2) Follow-up of Significant Non-Sickling Hemoglobinopathies FD, FDa, FC, FCa, FV, FVa

GENERAL INFORMATION: The following hemoglobinopathies are included in this protocol:

- FC** Homozygous Hb C (CC) or Hb C/beta⁰ thalassemia.
Hb CC is usually a benign condition, but may be associated with hemolytic anemia and aplastic episodes.
Hb C/beta⁰ thalassemia can be associated with mild to moderate hemolytic anemia and moderate to marked splenomegaly.
- FCa** Hb C/beta⁺ thalassemia. Microcytosis and mild anemia are common in Hb C/beta⁺ thal.
- FD** Homozygous Hb D (DD) or Hb D/beta⁰ thalassemia.
Hb DD is a clinically benign condition which is not associated with anemia or hemolysis.
Hb D/beta⁰ thalassemia can be associated with microcytosis, mild anemia, splenomegaly and gallstones.
- FDa** Hb D/beta⁺ thalassemia. The possibility of transfusion cannot be ruled out. Hb D/beta⁺ thalassemia can be associated with microcytosis and mild anemia.
- FV** An immediate referral to a CCS Sickle Cell Disease Center is needed since this infant is producing no normal adult hemoglobin. Further laboratory analysis is needed to identify the variants and, if reported in the literature, possible clinical consequences. **NOTE:** This result may appear on the Case Summary screen in SIS as a pattern containing a number (e.g., F2) instead of V (unknown variant hemoglobin); the hospital and doctor NBS results mailers will show a pattern of FV.

POLICY: Written notification to parents and physicians will be made utilizing state-approved Parent and Doctor Letters (See 11.1 and 11.2).

PROTOCOL:

Resp. Person	Action
ASC NBS	<ul style="list-style-type: none"> Daily Reviews the interesting case report for the above significant non-

5.2 Follow-up of Significant Non-sickling Hemoglobinopathies FC, FCa, FD, FDa, FV, FVa

Coord.	<p>sickling hemoglobinopathy cases.</p> <ul style="list-style-type: none"> • Within 48 Hours notifies the physician of record by phone of the screening result and requests blood specimens to be drawn from the infant and both parents (if possible) for confirmatory testing to rule out a significant hemoglobin disorder. • Assists in arranging for specimen collection at birth hospital lab or other collection site and shipment to the Hemoglobin Reference Lab at Children's Hospital & Research Center at Oakland (CHRCO). • Assists primary care provider (PCP) with referral to a CCS Sickle Cell Disease Center (SCDC) (see <i>Referral to CCS Special Care Centers</i>). Infants with confirmatory lab results indicating homozygous hemoglobin C (CC) or homozygous hemoglobin D (DD) are referred for at least an initial evaluation and parent teaching. • Sends appropriate initial Doctor Letter (See Section 11.1) depending on diagnosis to PCP, summarizing the phone call. (For FC encloses pamphlet entitled "<i>Hemoglobin C</i>".)
ASC NBS Coord.	<ul style="list-style-type: none"> • Sends <i>Instructions for Collection, Handling, And Mailing Of Blood Specimens For Confirmatory Testing</i> (5.8), as well as the shipping materials (cylinder, GSO mailing label) to lab obtaining confirmatory specimen(s). • Within 2-3 days, follows up with the PCP to find out if family has been contacted. • After confirming that family has been notified, sends appropriate Parent Letter (Section 11.2) depending on diagnosis. • If asked by the PCP to notify the family directly, after the contact is made, sends appropriate Parent Letter depending on the diagnosis confirming discussion with the parent and providing information about immediate follow-up care for the infant. • Makes referral to CCS (See <i>Referral to CCS Special Care Centers</i>). • After one week of trying to contact the family, if the newborn's PCP and/or Coordinator is unable to reach the family, sends Parent Letter #2 informing the family to call the newborn's PCP or Coordinator regarding baby's test results. Parent Letter #2 shall be sent by regular 1st class mail and a second copy should be sent by Certified Receipt mail requested to maximize receipt by parent.

5.2 Follow-up of Significant Non-sickling Hemoglobinopathies FC, FCa, FD, FDa, FV, FVa

	<ul style="list-style-type: none"> • If contact with family is still not made after one week of sending Parent Letter #2, makes arrangements for home visit by local health department public health nurse.
Hb Reference Lab	<p>Conducts confirmatory testing on liquid blood specimen(s).</p> <ul style="list-style-type: none"> • Within 11 working days enters results in SIS and informs the ASC NBS Coordinator of the confirmatory test results by fax or phone, followed by a hard copy sent to the ASC NBS Coordinator and NBSB Hemoglobin Coordinator. Includes the following results: <ol style="list-style-type: none"> A) Separation of hemoglobins F, A, S, C, D, and E with relative concentrations for each hemoglobin on all specimens by cellulose acetate-citrate agar electrophoresis, isoelectric focusing, high pressure liquid chromatography, and/or DNA analysis as outlined in the Hb Reference Lab NBS vendor agreement scope of work or as approved by the NBSB. B) Hemogram on each suitable specimen, to include hemoglobin, hematocrit, and mean corpuscular volume (MCV) and mean corpuscular hemoglobin (MCH). C) Free Erythrocyte Protoporphyrin (FEP) on specimens with microcytic hypochromic anemia. D) Quantitative Hb A2 when necessary to resolve phenotype. E) Quantitative Hb F when necessary to resolve phenotype. a. Within 31 calendar days of receipt of specimen, enters results in SIS, faxes and mails reports on: b. Beta globin DNA analysis <ul style="list-style-type: none"> • To rule out heterozygosity for beta thalassemia in the absence of one parent • If heterozygous for beta thalassemia, to determine the beta thalassemia mutation • To examine inconsistencies between thin layer isoelectric focusing profiles of newborn and parents. c. Presumptive findings for unusual HB variants such as Hb Korle Bu, Hb T Cambodian, Hb Matsue-Okie, Hb O Arab, Hb C Harlem, Hb G Philadelphia. • Within 91 calendar days of receipt of specimen, reports and enters in SIS: <ol style="list-style-type: none"> a. Final determinations for unusual Hb Variants such as Hb Korle Bu, Hb T Cambodian, Hb Matsue-Okie, Hb O Arab, Hb C Harlem, Hb G Philadelphia. b. Analysis of rare variants carried in compound heterozygosity with clinically significant hemoglobins (including but not limited to Hb S, C, or E, beta thalassemia, or three alpha chain deletions) or as only the adult hemoglobin (VV).

5.2 Follow-up of Significant Non-sickling Hemoglobinopathies FC, FCa, FD, FDa, FV, FVa

ASC NBS Coord.	<ul style="list-style-type: none">• Phones the NBS Hb Coordinator with any confirmatory results that are inconsistent with the NBS results prior to contacting the PCP.• Phones the PCP with the confirmatory test results.• Sends appropriate follow-up Doctor letter (Section 11.1) depending on diagnosis with the lab report to the PCP and includes a copy of <i>Diagnosis And Treatment</i> form to be used for reporting the disease.• Sends a copy of Doctor letter and lab result to the SCDC.• Resolves case in SIS when <i>Diagnosis & Treatment</i> form has been received, or the SCDC has completed the Hemoglobin Service Report in SIS indicating that infant has been seen and treatment is either initiated or not required. See Case Resolution Protocol 7.30.• Reports any missed cases, lost to follow-up cases or other unusual occurrences of potential significance to NBSB Nurse Consultant/ASC Contract Liaison.• Refers case to Child Protective Services as appropriate, and with approval of NBS Nurse Consultant/Contract Liaison {See 7.1 <i>Referral of Cases to Child Protective Services (CPS)</i>}.
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