

5.1 (old 3.13.1) Follow-up for Sickle Cell Disease - FS, FSa, FCS, FSC, FSD, FDS, FSE, FES, FSV, and FVS

POLICY:

Because current California newborn screening hemoglobin testing methodology (high pressure liquid chromatography, HPLC) has a high level of specificity for identifying infants with sickle cell diseases, infants with positive screening results should be referred to a CCS Sickle Cell Disease Special Care Center (SCDC) for follow-up care and initiation of penicillin prophylaxis without waiting for the results of confirmatory testing.

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

Note: Initial screening results of FSV or FVS may appear on the SIS Case Summary screen as a pattern containing a number (e.g., FS2) instead of V (unknown variant hemoglobin). The interpretation on the SIS screen is "Hb Positive" indicating this is a disease pattern. The printed results sent to the hospital and physician show a hemoglobin result of FSV or FVS and an interpretation of Sickle Cell Disease.

Attachment:

5.1.1 (old 3.13.1 A) California NBS Program Recommendations for Care of Infants Identified on Newborn Screening with Sickle Cell Disease

PROTOCOL:

Resp. Person	Action
ASC NBS Coord.	<ul style="list-style-type: none">• Daily reviews the Headline Case Report for hemoglobinopathy cases.• Within 48 Hours of obtaining positive result for Sickle Cell Disease, notifies the physician of record by phone of the positive screening result and informs him/her of the following:<ul style="list-style-type: none">a. Need for immediate referral to a CCS Sickle Cell Disease Special Care Center (SCDC) for follow-up care and initiation of penicillin prophylaxis without waiting for the results of confirmatory testing (see Policy above).b. The <i>California Newborn Screening Program Recommendation for Care of Infants Identified on Newborn Screening with Sickle Cell Disease</i> includes: 1) referral to an SCDC, 2) recommendation for initiation of penicillin prophylaxis before 2 months of age, and 3) follow-up guidelines and care schedules.

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- c. Request blood specimens to be drawn from the infant and both parents (if possible) for confirmatory testing to determine the type of sickle cell disease. Assists in arranging for specimen collection at birth hospital lab or other collection site and sent to the Hemoglobin Reference Lab at Children's Hospital & Research Center at Oakland (CHRCO).

Note: A copy of prenatal diagnostic test results showing the infant's hemoglobin type is acceptable in lieu of confirmatory testing if results agree with the newborn screening results; fax a copy of the results to the NBS Hemoglobin Coordinator. Check whether birth hospital performed confirmatory testing of prenatal diagnosis at birth via cord blood or venous blood specimen; obtain copy of results if done.

- Documents all attempts at notification, interactions with physicians and parents using tracking events or case notes in SIS.
- Sends **Doctor Letter #1** (Section 11.1) along with a copy of *California Newborn Screening Program Recommendation for Care of Infants Identified on Newborn Screening with Sickle Cell Disease* (See Section 5.1.1) and a copy of *Instructions for Collection, Handling, and Mailing of Confirmatory Blood Specimens*. (Section 5.8).
- Provides the collection site where parents are referred with *Instructions for Collection, Handling, and Mailing of Confirmatory Blood Specimens*, Lab Intake form (Section 5.8), and shipping materials (including cylinder and GSO label).
- **Within 2 to 3 days of first phone call to the primary care provider (PCP)**, calls PCP again to confirm that he/she contacted family, and if not done, offers to contact family directly and arrange for medical follow-up.
- Contacts parents by phone to either reinforce information given by PCP or give initial information regarding the results and need for follow-up care.
- After contact, sends **Parent Letter #1** along with *Sickle Cell Disease in Babies* brochure.
- Makes referral to CCS (See *Referrals to CCS Special Care Centers* protocol).
- Continues to attempt parent contact about baby's result if initially unsuccessful.
- **If the PCP and ASC Coordinator are unable to contact the parents within one week after the first call to the PCP**, sends **Parent Letter #2** (Section 11.2), notifying parents to call the PCP about baby's test results. Parent Letter #2 shall be sent by regular 1st class mail and a second copy sent by Certified Receipt mail requested to maximize receipt by parent.

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	<ul style="list-style-type: none"> • If contact still not made in one week after sending Parent Letter #2, makes an arrangement for home visit by local Health Department Public Health Nurse. • Continues to follow up to obtain confirmatory specimen and have it sent to Hb Reference Lab, CHRCO.
<p>Hb Reference Lab (CHRCO)</p>	<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 (initial analysis) by fax , 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 contract scope of work or as approved by the State. b. Hemogram on each suitable specimen which includes hemoglobin, hematocrit, mean corpuscular volume (MCV) and mean corpuscular hemoglobin (MCH). c. Free Erythrocyte Protoporphyrin (FEP) on specimens with microcytic hypochromic anemia. d. Quantitative A2 when necessary to resolve phenotype. e. Quantitative F when necessary to resolve phenotype. • Within 31 calendar days of initial results, enters results in SIS and reports out the following: <ol style="list-style-type: none"> a. Beta globin DNA analysis <ul style="list-style-type: none"> • to differentiate between SS and S/Beta thalassemia in the absence of one parent • in infants heterozygous for beta thalassemia, to determine the thalassemia mutation • to examine inconsistencies between thin layer isoelectric focusing profiles of newborn and parents. b. Presumptive findings for unusual Hb variants such as: Hb Korle Bu, Hb T Cambodian, Hb Matsue-Oki, Hb O Arab, Hb C Harlem, Hb G Philadelphia. • Within 91 calendar days of initial results, enters results in SIS, faxes and mails reports on: <ol style="list-style-type: none"> a. Final determinations for unusual Hb variants such as Hb Korle Bu, Hb T Cambodian, Hb Matsue-Oki, Hb O Arab, Hb C Harlem, Hb G Philadelphia. b. Analysis of rare variants carried in compound heterozygosity with clinically significant hemoglobinopathies (including but not limited to Hb S, C, E, mutations, beta thalassemia major, three alpha chain deletions) or as having only the adult hemoglobin (A).

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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 confirmatory test results.• Sends Doctor Letter # 2 along with copy of the lab report to PCP.• Calls the SCDC the infant was referred to with the results and sends a copy of Doctor Letter #2 along with a copy of the <i>Diagnosis and Treatment</i> form (to be used for reporting the disease).• 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 to the NBS Hb Coordinator and lost to follow-up cases, delays in contacting family, delays in analysis or reporting of confirmatory results, or other unusual occurrences of potential significance to the NBSB Nurse Consultant /ASC Contract Liaison.• Refers case to Child Protective Services, as appropriate, and with approval of NBS Nurse Consultant/Contract Liaison.
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