

7.2.3 California Newborn Screening (NBS) Program Confirmatory Testing for Initial Positive NBS Results & Referral to CCS

| <i>Disorder</i> | <i>Confirmatory Testing Included in NBS Program Fee</i> | <i>Where Confirmatory Testing Done</i> | <i>When Cases Referred to CCS Center</i> |
|---|---|--|---|
| Phenylketonuria (PKU) | Yes | State Newborn and Prenatal Screening Laboratories | After positive confirmatory test. NOTE: NBS Coordinator facilitates Biopterin studies for all confirmed positives through the Genetic Disease Laboratory, |
| Biopterin | Yes | Genetic Disease Laboratory | After positive confirmatory PKU test |
| Galactosemia | Yes | Associated Regional and University Pathologists, Inc] (ARUP) | After confirmatory testing (Screen Positive for Classical (GG) and Duarte/ Galactosemia (DG) |
| Primary Congenital Hypothyroidism | No | Private California licensed laboratory (including hospital laboratories) | After initial positive TSH test result (Tests are ordered by SCC, Endocrine Specialist or primary care provider in consultation with specialist) |
| Sickle Cell Disease and other potentially clinically significant hemoglobinopathies (except FE) | Yes | Children's Hospital and Research Center at Oakland | After collection of confirmatory test, prior to test results |

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| <i>Disorder</i> | <i>Confirmatory Testing Included in NBS Program Fee?</i> | <i>Where Confirmatory Testing Done</i> | <i>When Cases Referred to CCS Center</i> |
|---|--|---|---|
| Hemoglobin (Hb) FE | Yes | Children's Hospital and Research Center at Oakland | After confirmatory testing completed (After confirmation of E/Beta Thalassemia (Hb EE not referred)) |
| Disorders Identified via MS/MS (other than PKU) | No, but parents will not be charged any of out of pocket expenses (except possibly small fee for specimen collection by hospital if not collected at Quest Patient Service Center) | Quest Diagnostics (started July 1, 2005) | After initial positive results (Tests are ordered by Metabolic Center) |
| Congenital Adrenal Hyperplasia (CAH) | No | Private Laboratory | After initial positive screening (1 st or 2 nd tier test. Tests ordered by SCC, Endocrine Specialist or Primary Care Physician or Neonatologist. |
| Cystic Fibrosis | No | CCS-certified and Cystic Fibrosis Foundation (CFF)-approved CF Center using a CFF-approved lab. | After initial positive results. |
| Biotinidase Deficiency | Yes | Stanford Biochemical Genetics Laboratory | After initial positive results of <6 ERU, 2 nd screen results of <10 ERU |