



California
Department of
Health Services

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State of California-Health and Human Services Agency
Department of Health Services



ARNOLD SCHWARZENEGGER
Governor

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TO: ALL COUNTY CALIFORNIA CHILDREN'S SERVICES (CCS)
ADMINISTRATORS, MEDICAL CONSULTANTS, AND STATE
CHILDREN'S MEDICAL SERVICES (CMS) STAFF

SUBJECT: AUTHORIZATION OF DIAGNOSTIC AND TREATMENT SERVICES FOR
INFANTS REFERRED BY THE CALIFORNIA NEWBORN SCREENING
(NBS) PROGRAM INCLUDING FOR ADDITIONAL METABOLIC
DISORDERS AND CONGENITAL ADRENAL HYPERPLASIA

I. Background

The California NBS Program currently screens more than 500,000 newborns annually for phenylketonuria (PKU), galactosemia, primary congenital hypothyroidism, and hemoglobinopathies including sickle cell disease. The NBS Program will expand on or before August 1, 2005, to include over 40 additional metabolic conditions detectable via Tandem Mass Spectrometry (MS/MS), as well as classical congenital adrenal hyperplasia (CAH) including the salt-wasting and simple virilizing forms. All the conditions for which the NBS Program screens are CCS-eligible (see enclosed list of disorders). It is imperative that these disorders are diagnosed early to avoid serious disabilities and even death in some cases. A delay of even a few days can result in sudden death for some of these infants.

Annually, we estimate that with the newborn expansion, the following approximate number of referrals may be made for diagnostic evaluations throughout the state:

- 575-875 referrals to Metabolic Centers (PKU, galactosemia, and expanded inborn errors of metabolism),
- 175 to Sickle Cell Center (sickle cell disease and other hemoglobinopathies), and
- 1350-1700 to Endocrine Centers (primary congenital hypothyroidism and CAH).

In order to expedite the authorization of diagnostic services for the infants identified with a positive NBS report, the CCS program has developed an "Expedited Diagnostic Service Request" form (enclosed). Receipt of this form (completed by the NBS Coordinator) will ensure that the Special Care Center (SCC) Medical Director will be guaranteed reimbursement for the initial office visit and evaluation, and ensure that the SCC will make timely appointments for these services and see the patient prior to receipt of a CCS authorization.

The NBS Coordinator will facilitate the referral and evaluation process by

- Notifying the infant's primary care provider;
- Notifying the specialist at the CCS-approved SCC of the positive NBS result and requesting that the specialist contact the primary care provider to discuss the diagnostic evaluation;
- Contacting the family to verify notification from the doctor and providing information; and
- Completing the "Expedited Diagnostic Service Request" form and faxing it, together with a blank CCS application form and a copy of the positive NBS report, to the SCC.

The SCC will facilitate the referral and evaluation process by:

- Contacting the primary care provider to discuss the diagnostic evaluation;
- Scheduling the appointment at the SCC; and
- Reporting the diagnostic test results and the diagnosis to the NBS Coordinator and the CCS program.
- Completing the referral process by assisting the family to complete and sign the CCS application at the time of the visit and faxing the completed application to the local CCS office.

II. Policy

- A. CCS shall issue an authorization to the appropriate CCS-approved SCC to perform a diagnostic evaluation on **ALL** infants referred by the NBS

Program. The authorization shall be for three (3) months. The NBS Program staff will identify the SCC to which the infant will be referred. The identified SCC may be a Metabolic, Endocrine, or Sickle Cell Center depending on the screening result. (See enclosed list of SCCs that can be authorized).

- B. These authorizations shall be issued within five working days of receipt of all the following documentation:
1. An "Expedited Diagnostic Service Request" form (to be faxed by the NBS Coordinator);
 2. The positive NBS report (to be faxed by the NBS Coordinator);
 3. A "New Referral CCS/GHPP Client Service Authorization Request (SAR)" form (signed and faxed by the NBS Coordinator); and
 4. A signed CCS application for infants who do not have full scope no share of cost Medi-Cal or who are not Healthy Families subscribers. Authorizations shall be issued **without** a signed CCS application for infants who have full scope no share of cost Medi-Cal or who are Healthy Families subscribers.
- C. The \$20 CCS assessment fee is to be waived for these diagnostic services.

III. Policy Implementation

- A. Authorizations for diagnostic evaluations using SARs should be given to the SCC for medical Service Code Group (SCG) 02.
- B. Diagnostic confirmation for some of these disorders may require multiple SCC or specialist visits and more extensive laboratory testing. The initial diagnostic evaluation shall be issued for three (3) months and may be modified as needed if the diagnostic evaluation is not completed in three months.
- C. Authorizations shall include the following information in CMS Net web selected from special instructions or similar language for a legacy authorization:

Provider must bill other health insurance (OHC) first; submit Explanation of Benefits (EOB) with claim.

- D. For infants whose diagnostic evaluation confirms the presence of a metabolic, endocrine or hematologic disorder, the CCS program shall initiate the steps to determine eligibility for ongoing treatment services. All these disorders are medically eligible for CCS.
1. Authorizations for treatment services shall be issued to CCS approved Metabolic, Endocrine, or Sickle Cell SCCs for infants who have full scope, no share of cost Medi-Cal or who are Healthy Families (HF) subscribers.
 2. Families of other infants who are not eligible for full scope, no share of cost Medi-Cal and who are not HF subscribers must complete program eligibility requirements prior to the issuance of treatment authorizations.
- E. For some of the infants/children diagnosed with inborn errors of metabolism, treatment will include specific therapies, but for others treatment will be primarily once-a-year-follow-up with the SCC or specialist that consists of assessment of the infant/child and instruction/teaching for the family. As long as the infant/child is followed by the SCC or specialist, regardless of the frequency of treatment, the infant/child shall remain a CCS client. For infants/children without full scope no share of cost Medi-Cal or HF coverage, program eligibility will need to be confirmed annually.
- F. Authorizations for treatment using SARs should be provided to the SCC for medical SCG 02.
- G. Authorizations for primary care physicians may be issued for treatment and follow-up of the CCS eligible condition, in conjunction with the SCC or specialist, using a SAR with medical SCG 01.

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If you have any questions regarding the above policy or policy implementation, please contact your Regional Office Medical Consultant.

Original Signed by Marian Dalsey, M.D., M.P.H.

Marian Dalsey, M.D., M.P.H., Acting Chief
Children's Medical Services Branch

Enclosures:
List of Conditions
List of SCCs
Form
Letter to SCCs

California Department of Health Services
Genetic Disease Branch
Newborn Screening Program
Disorders Detectable by NBS Program as of mid-2005¹ *

I. Metabolic Disorders

A. Carbohydrate Disorders (1)

- **classical galactosemia**

B. Amino Acid Disorders (18)

- **classical phenylketonuria (PKU)**
- **variant PKU**
- **biopterin disorders** (4 types)
- argininemia/arginase deficiency
- argininosuccinic acid lyase deficiency (ASAL deficiency)
- citrullinemia, Type I/argininosuccinic acid synthetase deficiency (ASAS deficiency)
- citrullinemia, Type II (citrin deficiency)
- gyrate atrophy of the choroid and retina
- homocitrullinuria, hyperornithinemia, hyperammonemia –HHH
- homocystinuria/cystathionine beta-synthase deficiency (CBS deficiency)
- Methionine adenosyltransferase deficiency (MAT deficiency)
- maple syrup urine disease – (MSUD)
- non-ketotic hyperglycinemia
- prolinemia
- tyrosinemia

C. Organic Acid Disorders (18)

- 2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency
- 2-methylbutyryl-CoA dehydrogenase deficiency
- 3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMGCoA lyase deficiency)
- 3-methylcrotonyl-CoA carboxylase deficiency (3MCC deficiency)
- 3-methylglutaconic aciduria (MGA), Type I (3-methylglutaconyl-CoA hydratase deficiency)
- 5-oxoprolinuria
- beta-ketothiolase deficiency (BKT)
- ethylmalonic encephalopathy (EE)
- glutaric acidemia type-1 (GA-1)
- isobutyryl-CoA dehydrogenase deficiency
- isovaleric acidemia (IVA)
- malonic aciduria
- methylmalonic acidemia, mut –
- methylmalonic acidemia, mut 0
- methylmalonic acidemia (Cbl A, B)
- methylmalonic acidemia (Cbl C, D)
- multiple carboxylase deficiency (MCD)
- propionic acidemia (PA)

¹ Disorders in bold are currently detected by the newborn screening program

D. Fatty Acid Oxidation Disorders (10)

- carnitine transporter deficiency
- carnitine-acylcarnitine translocase deficiency (CAT deficiency)
- carnitine palmitoyl transferase deficiency-type 1 (CPT-1 deficiency)
- carnitine palmitoyl transferase deficiency-type 2 (CPT-2 deficiency)
- long chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD deficiency)
- medium chain acyl-CoA dehydrogenase deficiency (MCAD deficiency)
- multiple acyl-CoA dehydrogenase deficiency (MAD deficiency)/glutaric acidemia type-2 (GA-2)
- short chain acyl-CoA dehydrogenase deficiency (SCAD deficiency)
- trifunctional protein deficiency (TFP deficiency)
- very long chain acyl-CoA dehydrogenase deficiency (VLCAD deficiency)

II. Endocrine Disorders (4)

- **primary congenital hypothyroidism**
- **variant hypothyroidism**
- congenital adrenal hyperplasia-salt wasting (21-hydroxylase deficiency)
- congenital adrenal hyperplasia-simple virilizing (21-hydroxylase deficiency)

III. Hemoglobin Disorders (24)

- **sickle cell anemia (Hb S/S disease)**
- **sickle C disease (Hb S/C disease)**
- **sickle D disease (Hb S/D disease)**
- **sickle E disease (Hb S/E disease)**
- **Hb S/ hereditary persistence of fetal hemoglobin (Hb S/HPFH)**
- **sickle cell disease variant (other sickle cell disease, Hb S/V)**
- **Hb S/ Beta⁰ thalassemia**
- **Hb S/Beta⁺ thalassemia**
- **Hb C disease (Hb CC)**
- **Hb D disease (Hb DD)**
- **alpha thalassemia major**
- **Hb H disease**
- **Hb H/ Constant Spring disease**
- **beta thalassemia major**
- **Hb E/ Beta⁰ thalassemia**
- **Hb E/Beta⁺ thalassemia**
- **Hb E/ Delta Beta thalassemia**
- **Hb C/ Beta⁰ thalassemia**
- **Hb C/Beta⁺ thalassemia**
- **Hb D/ Beta⁰ thalassemia**
- **Hb D/Beta⁺ thalassemia**
- **Hb Variant/ Beta⁰ thalassemia**
- **Hb Variant/Beta⁺ thalassemia**
- **other hemoglobinopathies (Hb variants)**

* Due to biological variability of newborns and differences in detection rates for the various disorders in the newborn period, the Newborn Screening Program will not identify all newborns with these conditions. While a positive screening result identifies newborns at an increased risk to justify a diagnostic work-up, a negative screening result does not rule out the possibility of a disorder. Health care providers should remain watchful for any sign or symptoms of these disorders in their patients. **A newborn screening result should not be considered diagnostic, and cannot replace the individualized evaluation and diagnosis of an infant by a well-trained, knowledgeable health care provider.**

CCS Approved Metabolic SCCs

Cedars –Sinai Medical Center
Children’s Hospital and Health Center of San Diego
Children’s Hospital of Central California
Children’s Hospital of Los Angeles
Children’s Hospital of Oakland
Children’s Hospital of Orange County
Harbor-UCLA Medical Center
Kaiser Permanente Medical Center (Oakland)
Kaiser Permanente Southern California
LAC-USC Medical Center
Lucile Salter Packard Children’s Hospital
Sutter Medical Center
UC Davis Medical Center
UC Irvine Medical Center
UCLA Medical Center
UC San Francisco

CCS Approved Endocrine SCCs

Cedars –Sinai Medical Center
Children’s Hospital and Health Center of San Diego
Children’s Hospital of Central California
Children’s Hospital of Los Angeles
Children’s Hospital of Oakland
Children’s Hospital of Orange County
Harbor-UCLA Medical Center
Loma Linda University Medical Center
Lucile Salter Packard Children’s Hospital
Miller Children’s at Long Beach Memorial Medical Center
Sutter Medical Center
UC Davis Medical Center
UCLA Medical Center
UC San Francisco

CCS Approved Sickle Cell SCCs

Cedars –Sinai Medical Center
Children’s Hospital and Health Center of San Diego
Children’s Hospital of Central California
Children’s Hospital of Los Angeles
Children’s Hospital of Oakland
Children’s Hospital of Orange County

City of Hope Medical Center
Harbor-UCLA Medical Center
Kaiser Permanente Medical Center (Oakland)
Kaiser Permanente Southern California
LAC-USC Medical Center
Loma Linda University Medical Center
Lucile Salter Packard Children's Hospital
Miller Children's at Long Beach Memorial Medical Center
Sutter Medical Center
UC Davis Medical Center
UC Irvine Medical Center
UCLA Medical Center
UC San Francisco

Expedited Diagnostic Service Request
For Infants With A Positive Newborn Screening (NBS) Test
To: The Special Care Center (SCC)

- Metabolic
- Endocrine
- Sickle Cell Disease

The purpose of this request is to expedite the diagnostic evaluation for an infant with a positive newborn screening test. **The CCS program will authorize these evaluations, but due to the scheduled appointment occurring quickly, the SCC may not have the authorization at the time of the visit.** The authorization will be forthcoming.

The infant named below will be scheduled for a diagnostic evaluation with the following SCC :

SCC _____ Physician _____

SCC Contact _____ Fax _____

Date of Visit (If known) _____

Infant's Name _____ AKA _____
(as on the positive newborn screening report) (when applicable)

Infant's Date of Birth _____ NBS Number _____
(initial NBS screening test accession #)

Mother's Name _____ AKA _____
(as on the positive newborn screening report) (when applicable)

Mother's Date of Birth _____

Primary Care Provider _____ Phone _____

The attached application* must be completed by the parent/legal guardian at the time of the SCC visit and then faxed to the appropriate CCS program.

The CCS program will issue an authorization to cover the diagnostic services within five working days of receipt of all necessary documents. **The SCC shall not charge the infant's family for any services (including room fees) related to this diagnostic evaluation.**

If you have any questions, please contact the following:

NBS Coordinator _____ Phone _____

Area Service Center _____ Phone _____

Attachments: (1) CCS Application (2) Copy of the positive NBS report

*The application can also be downloaded from
<http://www.dhs.ca.gov/pcfh/cms/ccs/publications.htm>. Click on DHS 4480 (English or Spanish)

cc: _____ County CCS program _____ Fax _____



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Department of Health Services



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May 26, 2005

**TO: MEDICAL DIRECTORS OF CALIFORNIA CHILDREN'S SERVICES
(CCS) APPROVED METABOLIC, ENDOCRINE, AND SICKLE CELL
SPECIAL CARE CENTERS (SCCs)**

**SUBJECT: EXPEDITING DIAGNOSTIC SERVICES FOR INFANTS REFERRED BY
THE CALIFORNIA NEWBORN SCREENING (NBS) PROGRAM**

The California NBS Program currently screens more than 500,000 newborns annually for phenylketonuria (PKU), galactosemia, primary congenital hypothyroidism, and hemoglobinopathies including sickle cell disease. The NBS Program will expand on or before August 1, 2005, to include over 40 additional metabolic conditions detectable via Tandem Mass Spectrometry (MS/MS), as well as classical congenital adrenal hyperplasia (CAH) including the salt-wasting and simple virilizing forms. The NBS Program has already initiated pilot testing of the expanded program in the Los Angeles area prior to launching the statewide program.

All the conditions for which the California NBS Program screens are CCS eligible. Therefore, the Genetic Disease Branch (GDB) and the CCS program have jointly developed a plan to ensure that infants with positive NBS reports receive timely diagnostic evaluations at CCS-approved SCCs. Your assistance is needed to ensure expedited diagnostic evaluations for all infants with positive NBS tests.

CCS and the NBS programs have developed an "Expedited Diagnostic Service Request" form (enclosed with this letter). Receipt of this form (completed by the NBS Coordinator) ensures that the SCC Medical Director will be guaranteed reimbursement for the initial office visit and evaluation, and ensures that the SCC will make timely appointments for these services prior to receipt of a CCS authorization.

When the NBS Coordinator notifies the specialist at the SCC of the positive NBS result, the Coordinator will fax the SCC a completed "Expedited Diagnostic Service Request" form, along with a CCS application and the positive NBS report. The only variation to this procedure would be when the NBS Coordinator receives the positive screen on a

MEDICAL DIRECTORS OF CALIFORNIA CHILDREN'S SERVICES (CCS)
APPROVED METABOLIC, ENDOCRINE, AND SICKLE CELL SPECIAL CARE
CENTERS (SCCs)

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holiday or weekend. The Coordinator may instead give the information verbally to the SCC and fax the documents on the next business day. The NBS Coordinator will also fax a "New Referral CCS/GHPP Client Service Authorization Request" to the county CCS office or CMS Branch Regional Office

To facilitate receipt of the authorization from the CCS program, the SCC should assist the family in completing and signing the CCS application. The CCS application will be faxed by the NBS Coordinator. The application can also be downloaded from <http://www.dhs.ca.gov/pcfh/cms/ccs/publications.htm> by clicking on DHS 4480 (English or Spanish). Please fax the completed application to the local County CCS program. Your assistance with the completion of the CCS application will further enable timely evaluations of these infants with positive screening tests.

The County CCS program or CMS Regional Office will authorize a diagnostic evaluation for **ALL** infants referred by the NBS Program. The initial authorization will be for three months and may be extended as needed. These authorizations shall be issued within five working days of receipt of all the following documentation:

- "New Referral CCS/GHPP SAR" form (from NBS Coordinator),
- "Expedited Diagnostic Service Request" form (from NBS Coordinator),
- Positive NBS report (from NBS Coordinator), and
- The CCS application (preferably faxed from the SCC).

CCS authorizations will include the following information or similar language:

Provider must bill other health insurance (OHC) first; submit Explanation of Benefits (EOB) with claim.

Thank you for your continued commitment to infants and children with special health care needs and their families. I realize scheduling appointments for these infants with positive screens will require juggling of appointment calendars, but the critical need for these babies to be seen quickly necessitates that we all make adjustments in the way we normally provide services.

MEDICAL DIRECTORS OF CALIFORNIA CHILDREN'S SERVICES (CCS)
APPROVED METABOLIC, ENDOCRINE, AND SICKLE CELL SPECIAL CARE
CENTERS (SCCs)

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If you have questions about the described procedure, you may contact the appropriate
NBS Coordinator/Area Service Center.

Original Signed by Marian Dalsey, M.D., M.P.H.

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Enclosure

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