

DEPARTMENT OF HEALTH SERVICES

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January 18, 2002

N.L.: 01-0102
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TO: ALL COUNTY CALIFORNIA CHILDREN'S SERVICES (CCS)
ADMINISTRATORS AND MEDICAL CONSULTANTS, CHILD HEALTH
AND DISABILITY PREVENTION (CHDP) PROGRAM DIRECTORS AND
DEPUTY DIRECTORS, AND STATE CHILDREN'S MEDICAL SERVICES
(CMS) STAFF

SUBJECT: AUTHORIZATION OF DIAGNOSTIC SERVICES FOR INFANTS
REFERRED BY THE NEWBORN SCREENING PROGRAM (GENETIC
DISEASE BRANCH (GDB)) FOR UNUSUAL TEST RESULTS FROM THE
SUPPLEMENTAL SCREENING FOR MULTIPLE METABOLIC
DISORDERS TANDEM MASS SPECTROMETRY (MS/MS) RESEARCH
PROJECT

I. Background

The California Newborn Screening Program currently screens more than 500,000 newborns annually for phenylketonuria (PKU), galactosemia, primary congenital hypothyroidism, and sickle cell disease and other hemoglobinopathies. Through this mandatory screening program, about 350 newborns with these disorders are identified and treated every year. Recent advances in tandem mass spectrometry (MS/MS) have made it possible to identify more than 30 metabolic disorders through newborn screening. A list of 26 disorders (which are in addition to the conditions for which screening is already done) is enclosed. When these disorders are diagnosed late or left undiagnosed, serious disabilities and even death can result.

Concerned about early detection and treatment of these metabolic disorders, on September 28, 2000, Governor Gray Davis signed into law Assembly Bill 2427 (Kuehl) which provides for updating and expanding the newborn screening program in California. The law took effect on January 1, 2001, and requires the Department of Health Services (DHS) to investigate the feasibility of establishing a new and broader testing program, including development and evaluation of expanded genetic disease testing utilizing MS/MS. The GDB has initiated a research project that will determine which of the approximately 26 additional

disorders identifiable via MS/MS meet the criteria for inclusion in California's mandatory newborn screening program, i.e., which of the unusual results have clinical significance and warrant reporting. DHS is planning to launch this pilot research project on January 7, 2002.

The project begins with the education of prenatal care providers and hospital staff. The prenatal care providers will distribute a revised copy of the informational booklet, *Important Information for Parents about the Newborn Screening Test*, to women during their pregnancy. This revised booklet describes the research project and the voluntary screening, in addition to the mandatory newborn screening. During the maternity stay, hospital staff will give new mothers another copy of the same booklet, and the mothers will be asked to complete and sign a consent form indicating whether they want their newborn to participate in the research study. No additional blood collection will be necessary for the study, since the additional testing will be performed on the same blood spots collected for the mandatory screening. There will be no additional charge to the family for participation in the study.

The physicians of newborns with unusual MS/MS screening results will be contacted by telephone by the MS/MS follow-up coordinator and will then receive a results report by mail. The coordinator will also directly contact the parents of the newborn by telephone to discuss the need for a diagnostic evaluation. After contacting the parents, the coordinator will fax a referral form to the appropriate CCS office requesting an authorization for a diagnostic evaluation at a CCS approved metabolic center. The coordinator will schedule the appointment at the center and will mail a letter (copy enclosed), including information about CCS and the CCS application form, with instructions for submission to expedite the referral. The letter will contain the date of the appointment at the metabolic center.

The CCS approved metabolic centers in the state have been involved in the planning of this project and are prepared to evaluate these infants for the presence of one of the specified disorders. Once authorized, the metabolic center will make the arrangements for confirmatory testing and develop the diagnostic, and, if needed, treatment plan, which will then be forwarded to the primary care provider, GDB, and the CCS program that has authorized the services.

The estimated duration of the study will be 12-18 months. The GDB projects that about 400,000 newborns may be tested in that time period. There is a 0.3

percent positive rate, so approximately 1200 infants may have unusual results. Based on the national experience to date using MS/MS screening, this supplemental testing may identify 40-60 newborns with clinically significant metabolic disorders.

II. Policy

- A. CCS shall issue authorizations to CCS approved metabolic centers to perform a diagnostic evaluation on ALL infants referred through the GDB Newborn Screening Program. Please refer to the metabolic centers in III.A., as some of the CCS approved metabolic centers are not participating and Children's Hospital Oakland is participating with restrictions.
- B. These authorizations shall be issued within five working days of receipt of the referral.
- C. Issuance of this authorization for diagnostic services requires only the receipt of a Request for Service form and a signed CCS application. For infants with Medi-Cal full scope, no share of cost, or Healthy Families coverage, the authorization can be issued with only a Request for Service form.
- D. The CCS \$20 assessment fee is waived for these services.

III. Policy Guidelines

- A. An authorization for a diagnostic metabolic evaluation shall be issued to a CCS approved metabolic center for a period not to exceed three months. The CCS approved metabolic centers that are providing diagnostic and treatment services in conjunction with GDB are:
 - The Permanente Medical Group of Oakland
 - University of California San Francisco (UCSF)
 - Sutter Medical Center (Sacramento)
 - Valley Children's Hospital
 - Kaiser Permanente Southern California Region
 - Cedars-Sinai Medical Center
 - Children's Hospital Los Angeles
 - Harbor/University of California Los Angeles (UCLA)

- Los Angeles County (LAC)-University of Southern California (USC) Medical Center
- UCLA
- Children's Hospital and Health Center of San Diego

Please refer to Chapter 7 for information on authorization of services to these centers.

Children's Hospital Oakland will provide diagnostic services, but will only treat medium chain acyl-CoA dehydrogenase deficiency (MCAD). The center will refer infants diagnosed with other metabolic disorders to UCSF for ongoing care.

- B. Authorizations shall include the following information:
1. Claims for Medi-Cal eligible children should be forwarded directly to the authorizing CCS program for authorization of payment by the Medi-Cal program.
 2. Claims for services provided to children with other third-party coverage must be submitted to the insurance carrier or health maintenance organization prior to billing the CCS program for the services. A denial of payment from the third-party payer shall accompany the claim.
- C. For infants whose diagnostic evaluation confirms the presence of a metabolic disorder, the CCS program shall initiate the steps to determine eligibility for ongoing treatment services.
1. Authorizations for treatment services shall be issued to CCS approved metabolic centers for infants who are Medi-Cal full scope, no share of cost.
 2. Families of other infants must complete program eligibility requirements prior to the issuance of treatment authorizations.

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If you have any questions regarding this policy, please contact your Regional Office Medical Consultant.

A handwritten signature in black ink that reads "Maridee Gregory MD". The signature is written in a cursive style with a small flourish at the end.

Maridee A. Gregory, M.D., Chief
Children's Medical Services Branch

Enclosures

Disorders Detectable by Tandem Mass Spectrometry (MS/MS) Using Newborn Screening Dried Blood Spots*

Amino Acid Disorders

- maple syrup urine disease (MSUD)
- phenylketonuria (PKU)
- citrullinemia/argininosuccinic acid synthetase deficiency (ASAS)
- homocystinuria/cystathionine beta-synthase deficiency (CBS)
- argininosuccinyl-CoA lyase deficiency (ASAL)
- argininemia/arginase deficiency
- tyrosinemia

Organic Acid Disorders

- 3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMGCoA)
- glutaric acidemia type-1 (GA-1)
- isobutyryl-CoA dehydrogenase deficiency
- isovaleric acidemia (IVA)
- 2-methylbutyryl-CoA dehydrogenase deficiency
- 3-methylcrotonyl-CoA carboxylase deficiency (3MCC)
- propionic acidemia (PA)
- methylmalonic acidemia (MMA)
- beta-ketothiolase deficiency (BKD)

Fatty Acid Oxidation Disorders

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

****Some of these disorders may not be included in the California Department of Health Services, Tandem Mass Spectrometry (MS/MS) Research Project but should be reported by any health care provider making a diagnosis on or after 1/7/02.***

DEPARTMENT OF HEALTH SERVICES
GENETIC DISEASE BRANCH
2151 BERKELEY WAY, ANNEX4
BERKELEY, CA 94704



DRAFT: MS/MS Unusual Result-Parent Letter

Date:

Dear Ms. _____

Re: Name (baby): _____ aka: _____

DOB: _____

Hospital of Birth: _____

Accession #: _____

This letter reviews the information that we discussed by telephone on (DATE) regarding the results of your baby's tandem mass spectrometry (MS/MS) screen. The blood specimen was tested for additional metabolic disorders based on your consent in the hospital. The test indicated that your baby needs further testing to find out if (baby's name) has a metabolic disorder. (Your baby's doctor was notified about the test results.) It is important to know if your baby has one of these disorders because he/she may need special care such as a special diet or medicine in order to avoid serious health risks.

Your baby should be seen **immediately** for medical evaluation and testing by a metabolic specialist. I have made the referral to the _____ California Children Services (CCS) - approved Metabolic Center and have informed your doctor of this referral. **(Address and Phone number)** Your baby's appointment has been scheduled for **(date and time)**. Medi-Cal and the CCS program may also cover the cost of diagnosis and treatment for these disorders. (See enclosed brochure.) I have enclosed the CCS application, which must be completed and sent to _____ as soon as possible but no later than _____ if you are interested in applying for CCS coverage. The Newborn Screening Program will cover the cost of diagnostic testing if your insurance does not and you are not eligible for Medi-Cal or CCS coverage.

If you have any questions or concerns, please contact me at our toll-free number, 866-954-BABY (866-954-2229) or call your doctor at (telephone number).

Sincerely,

Cathy Berman, R.N., N.P., M.S.
Clinical Follow-up Coordinator
MS/MS Research Project
Newborn Screening Section

Enclosures: CCS brochure & CCS application

Cc: Newborn's Primary Care Provider