

California Department of Health Services, Genetic Disease Branch
Newborn Screening Program
Metabolic Disorders (excluding PKU and Galactosemia)
Confirmatory Laboratory, Quest Diagnostics
Key Points

7/20/05 – Revised 03/21/06

Scope of Services

The Newborn Screening Confirmatory Laboratory for Metabolic Disorders, Quest Diagnostics, will provide the State of California with confirmatory testing on: 1) blood, urine, and tissue specimens from newborns born in California whose screening results are defined as indicative of a potentially significant metabolic disorder; and 2) blood, urine and tissue specimens from the parents of newborns, whose results are necessary to confirm screening test results on their infants.

Note: recall/confirmatory testing for newborns with initial positive results for phenylketonuria (PKU) and galactosemia will continue to be done at the established confirmatory laboratories (State Genetic Disease Laboratory for PKU and CHLA for Galactosemia and Biopterin testing) and will not be done through this contract. Confirmatory testing for congenital adrenal hyperplasia and congenital hypothyroidism are also not provided as part of this laboratory contract.

Ordering Physician (Metabolic Specialist)

Tests must be ordered through a metabolic specialist at a CCS-approved Metabolic Special Care Center (SCC). In some situations the newborn will not be seen at a SCC prior to testing and the metabolic specialist will work closely with the primary care provider or neonatologist in making arrangements for specimen collection.

Test Request Form

Quest Diagnostics has supplied the metabolic centers with copies of the test request form (TRF) to be used for ordering tests under this contract. The State NBS Program is requesting some modifications to this form but in the meantime Metabolic Centers should use the current version of the form until notified. See appendix 1 for a sample of the Quest Diagnostics TRF.

Other Supplies

Quest Diagnostics has supplied the Metabolic Centers with collection kits and an order form for additional supplies. If they have not been received or if additional supplies and/or order forms are needed, they can be obtained by calling 1-800-LABTEST.

Instructions for Collection and Handling of Specimens

Quest Diagnostics has sent Metabolic Centers Medical Directors the *Genetic Test Directory* which provides instruction for the tests done on-site at Quest Diagnostics. Additional copies are being sent to metabolic clinic coordinators. Instructions for collection and handling of specimens not listed in its directory or additional copies of the directory may be obtained by calling 1-800-LABTEST. Specimen requirements may also be viewed on Quest's website at www.questdiagnostics.com. From the home page, click on "healthcare professionals", choose "Interpretive Guide", "browse by test", click on "genetics", and select the specific test desired.

Specimen Collection Sites

In addition to the Metabolic Centers, hospitals and doctors' offices/clinics, specimens may be collected at any of the Quest Diagnostics Patient Service Centers (draw stations). For a complete list of locations and hours of operation visit our website for the [Patient Services List](#). Under the table of contents for Laboratory and Healthcare Services click on "Find a Quest-Diagnostic Location". There are over 400 pediatric Patient Service Centers in California.

Specimen Pick-up

In addition to the Quest Diagnostics Patient Service Centers, specimens will be picked up from the Metabolic Centers, local hospitals, or doctors' offices/clinics. To arrange for a pick-up, call 1-800-LABTEST (as listed on the test request form) In addition, Dr. Neidich at Quest Diagnostics should be contacted by the Metabolic Center to coordinate specimen pick up from any non-Quest draw station at (949) 728-4936 or (800) 642-4657 ext. 4936. Quest is not currently trained on SIS, so Dr. Neidich must also be contacted to alert her that a specimen is on the way. When Quest is "up and running" on SIS, a "heads up" call to Dr. Neidich will not be required.

The time required to pick up a specimen is dependent upon when the call is placed to 1-800-LABTEST, where the collection site is located, and if that facility has an existing scheduled (daily) pick up from Quest.

Quest has existing business relationships with ten of the fourteen Metabolic Centers and Quest's couriers have daily scheduled stops at those Centers. For those Metabolic Centers, pickup should occur within a few hours a center's staff member places the call. The remaining four Metabolic Centers, all of which are within Quest's courier route, may not receive a specimen pick up until the end of the Metabolic Center's business day. (The four Metabolic Centers that do not have existing daily Quest pick ups are: Lucille Packard Children's Hospital, Children's Hospital & Research Center/Oakland, Children's Hospital of Orange County, and UCI Medical Center.)

Quest's courier service operates 8:00 am to 6:00 pm. If a specimen will not be picked up the same day the specimen is collected, Dr. Neidich should be contacted regarding storage of the specimen for pick up the following day or instructions for overnight delivery, if applicable. Quest can provide the Metabolic Centers with pre-paid FEDEX envelopes.

Available Testing and Turn Around Times

See appendix 2 for a list of tests and turn around times. Tests will be run 24 hours a day, seven days a week. Turn around time is based upon calendar days (and weekend days are counted).

Questions About Results, etc.

Dr. Neidich is the "single point" contact for Quest. Phone: (949) 728-4936 or (800) 642-4657 ext.4936.

Mailing Address: Quest Diagnostics Inc.
33608 Ortega Highway
San Juan Capistrano, CA 92675

Reporting of Results

Fax: Results for all disorders will be faxed as well as mailed to the ordering physician.

SIS look-up: Quest is not yet entering results into SIS but will be doing so in the near future.

On-line at Quest Diagnostics' Website (Care 360): Metabolic specialists will be able to access test results via Quest Diagnostics' Website. A Quest Diagnostics representative will contact the Metabolic Centers to set up a formal training session to review Quest Diagnostics' system. Passwords will be supplied during the training session.

Additional Reporting for Urgent Conditions: Results will be phoned (as well as faxed and mailed) to the ordering physician (metabolic specialist) for the following disorders classified by the NBS program as urgent:

- Citrullinemia (CIT),
- Glutaric Acidemia Types I and II (GA-1, GA-2)
- Isovaleric Acidemia (IVA)
- Long-Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHADD)
- Malonic Acidemia (MAL)
- Multiple Carboxylase Deficiency (MCD)
- Maple Syrup Urine Disease (MSUD)
- Propionic Acidemia/Methylmalonic Acidemia (PA/MMA).

Results will be reported out on a 24hour/7day a week basis, so metabolic centers submitting specimens for urgent disorders should include a phone number where they or the on-call physician may be reached after hours/on weekends.

Billing

The state contract allows for the collection of fees for testing and specimen collection/handling from any insurance plan, including Medi-Cal, for which the newborn or newborn's family is enrolled. However, Quest Diagnostics is not allowed to bill families for any services, or collect fees from them for any co-payments for services not covered by their insurance carrier for services provided for in this contract. These services include specimen collection, handling, transport and testing on newborns and family members ordered by the metabolic specialist which are deemed necessary to arrive at a diagnosis as a result of a California Newborn Screening Program authorized referral. In addition, Quest Diagnostics may not bill the ordering physician or metabolic center for any of the work done as part of this contract scope of work.

If specimen collection is done at the metabolic center, other hospital, or a doctor's office/clinic, the institution collecting the specimen may charge the family or their insurance a reasonable fee for specimen collection.

The cost of transporting the specimen (via courier or other method) will be paid by Quest Diagnostics if arrangements for transport are made by calling 1-800-LABTEST.

Genetic Disease Branch Contact: any issues regarding services provided for as part of this contract that you are unable to address/resolve with Quest Diagnostics should be directed to John Sherwin, PhD, Chief Genetic Disease Laboratory at (510) 231-1790.