

UCHSC Expanded Newborn Screening Program

Online at www.uchsc.edu/newbornscreening

Expanded Newborn Screening using Tandem Mass Spectrometry In Colorado and the United States

A technology known as tandem mass spectrometry (tandem MS or MS/MS) has now made it possible to easily and simultaneously screen a small sample of a newborn's blood, dried on filter paper, for more than twenty metabolic disorders. Screening using MS/MS has become part of the state-mandated programs in most states, including Colorado. Other states have mandated, but not yet implemented, MS/MS screening programs. Some states are evaluating the technology or offer such screening only upon parental request. In a few states, however, no such screening program is either established or planned.

To check the newborn screening program in your state,
view the latest National Newborn Screening Status Report online:

<http://genes-r-us.uthscsa.edu/nbsdisorders.pdf>

In Colorado, screening is done by collecting a small amount of blood from the baby onto filter paper during the first 1-2 days of life, and sending the filter paper to a lab at the State Health Department where the tests are done. This “mandatory” screening is done at no cost to the parents of the infants, but hospitals pay the Health Department a fee to sustain the program.

Parents of newborns in states where no such program exists can avail themselves of Expanded Newborn Screening only by sending blood samples on filter paper to one of a number of commercial laboratories. In addition to the Biochemical Genetics Laboratory at the University of Colorado Health Sciences Center in Denver, these include Pediatrix Screening in Pittsburgh and The Institute of Metabolic Disease laboratory at Baylor University Medical Center in Dallas. These laboratories all screen for a comparable panel of disorders. Most insurance companies do not cover this cost.

The disorders detected by tandem mass spectrometry include amino acid disorders, organic acid disorders, and fatty acid disorders. Infants with most of these conditions appear healthy at birth and symptoms appear later, in the first weeks or months of life. Diagnosis at that time can be very difficult because symptoms are quite nonspecific, and without rapid diagnosis and treatment many of these infants will die or sustain severe and irreversible brain damage. If diagnosis is made before symptoms develop, many of these infants can be treated and the adverse outcomes can be prevented.

Infants with amino acid disorders have high levels of amino acids in the blood, and those with organic acid and fatty acid disorders have high levels of compounds known as acylcarnitine esters. The elevations of particular amino acids or acylcarnitine esters are quite apparent in MS/MS analysis of newborn blood spots, and measures to make an accurate diagnosis and begin treatment can be undertaken.

For questions, information or screening kits, contact:

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Send specimen cards to:

Domestic Mail (via U.S. Postal Service):

UCDHSC Biochemical Genetics Laboratory
Mail Stop 8313
P.O. Box 6511
Aurora, CO 80045

International (ship via DHL, UPS, FedEx, etc.):

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