Written by CDPHE

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Cherry Creek— Beginning Saturday, July 1, the Colorado newborn screening program will expand to include testing for 23 additional metabolic diseases, bringing the total number of screened conditions to 30 metabolic diseases plus hearing.

Hospitals and health care providers throughout the state routinely submit blood samples from newborns to the Colorado Department of Public Health and Environment's Laboratory Services Division, where screening tests are conducted.

According to Jim Beebe, program manager for the laboratory, Colorado's current newborn screening panel includes seven metabolic disorders plus hearing. As of July 1, the panel will include 23 additional conditions recommended by the federal government, the American College of Medical Genetics and the March of Dimes. Beebe said the expanded screening will test for amino acid, organic acid and fatty acid oxidation disorders.

The health department's lab will test for the additional conditions, along with one of the original seven, with a relatively new screening technology known as tandem mass spectrometry, Beebe said. The lab will use traditional methods for the six remaining original tests.

The Inherited Metabolic Diseases (IMD) clinic at Children's Hospital will provide follow-up consultation to primary care physicians of babies who test positive for these diseases. Eleven of the conditions, considered "metabolic emergencies," warrant immediate medical intervention, which can save the lives of affected babies.

"We're pleased that this mandate will make the expanded testing a routine procedure," Beebe said, "because the tests will give health care providers crucial information they need to intervene in time to save the lives of infants."

The Colorado health department's Laboratory Services Division also conducts newborn screening tests for Wyoming, and that state is expanding its screening panel on July 1 to include the 30 conditions as well.

A list of the 30 conditions on the expanded newborn screening panel

accompanies this press release.

Colorado's expanded newborn screening panel will include tests for hearing plus the following 30 metabolic diseases:

Phenylketonuria
Congenital Hypothyroidism
Hemoglobinopathies
Galactosemia
Cystic Fibrosis
Biotinidase Deficiency
Congenital Adrenal Hyperplasia
Medium Chain Acyl-CoA dehydrogenase deficiency
Very Long Chain Acyl-CoA dehydrogenase deficiency
Long-chain Acyl CoA dehydrogenase deficiency
Trifunctional protein deficiency
Carnitine Acyl-carnitine translocase deficiency
Short Chain Acyl-CoA dehydrogenase deficiency
Carnitine palmitoyltransferase II deficiency
Glutaric acidemia Type 2
Arginosuccinic acidemia
Citrullinemia
Tyrosinemia
Hypermethionemia
Maple Syrup urine disease
Homocystinuria
Isovaleric acidemia
Glutaric acidemia Type 1
3-hydroxy-3 methyl glutaric aciduria
Multiple Carboxylase deficiency
3-methylcrotonyl-CoA carboxylase deficiency
3-methylglutaconic aciduria
Methylmalonic acidemias
Propionic acidemia
beta-Ketothiolase deficiency