DEPARTMENT OF PUBLIC HEALTH AND ENVIRONMENT

Laboratory Services Division

NEWBORN SCREENING AND SECOND NEWBORN SCREENING

5 CCR 1005-4

Adopted by the Board of Health on _____; effective _____.

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SECTION 2: NEWBORN SCREENING REQUIREMENTS FOR NAMED SUBMITTERS

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2.4 List of Conditions for Newborn Screening

The Laboratory shall conduct screening tests for the following conditions:

2.4.1 Phenylketonuria

2.4.2 Congenital Hypothyroidism

2.4.3 Hemoglobinopathies

2.4.4 Galactosemia

2.4.5 Cystic Fibrosis

2.4.6 Biotinidase Deficiency

2.4.7 Congenital Adrenal Hyperplasia

2.4.8 Medium Chain Acyl-CoA Dehydrogenase Deficiency

2.4.9 Very Long Chain Acyl-CoA Dehydrogenase Deficiency

2.4.10 Long-Chain L-3-Hydroxy Acyl-CoA Dehydrogenase Deficiency

2.4.11 Trifunctional Protein Deficiency

2.4.12 Carnitine Acyl-Carnitine Translocase Deficiency

2.4.13 Short Chain Acyl-CoA Dehydrogenase Deficiency

2.4.14 Carnitine Palmitoyltransferase II Deficiency

2.4.15 Glutaric Acidemia Type 2

2.4.16 Arginosuccinic Acidemia

2.4.17 Citrullinemia

2.4.18 Tyrosinemia

2.5.19 Hypermethionemia

2.4.20 Maple Syrup Urine Disease

2.4.21 Homocystinuria

2.4.22 Isovaleric Acidemia

2.4.23 Glutaric Acidemia Type 1

2.5.24 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency

2.4.25 Multiple Carboxylase Deficiency

2.4.26 3-Methylcrotonyl-CoA Carboxylase Deficiency

2.4.27 3-Methylglutaconic Aciduria

2.4.28 Methylmalonic Acidemias

2.4.29 Propionic Acidemia

2.4.30 Beta-Ketothiolase Deficiency

2.4.31 Carnitine Uptake Defect

2.4.32 Arginase Deficiency

2.4.33 Malonic Acidemia

2.4.34 Carnitine Palmitoyltransferase Deficiency 1a

2.4.35 Severe Combined Immunodeficiency

2.4.36 Spinal Muscular Atrophy due to homozygous deletion of exon 7 in Survival Motor Neuron 1 gene

SECTION 3: SECOND NEWBORN SCREENING REQUIREMENTS FOR NAMED SUBMITTERS

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3.2.2.2 Section 25-4-1004.5(3)(b)(V), C.R.S. allows exceptions to testing of second newborn screening specimens. Second newborn screening specimen testing is not required for the conditions identified at 3.3.1, 3.3.4, 3.3.5 and 3.3.6 unless: an unsatisfactory specimen was submitted for an initial newborn screening specimen; a~~n abnormal~~ screen positive result was obtained on an initial newborn screening specimen from the same newborn; ~~or~~ there is no record of a satisfactory initial newborn screening specimen submission, or; for 3.3.1 only, the initial newborn screening specimen from the same newborn was collected before 24 hours of life.

3.3 List of Conditions for Second Newborn Screening

The Laboratory shall conduct screening tests for the following conditions:

3.3.1 Phenylketonuria

3.3.2 Congenital Hypothyroidism

3.3.3 Hemoglobinopathies

3.3.4 Galactosemia

3.3.5 Cystic Fibrosis

3.3.6 Biotinidase Deficiency

3.3.7 Congenital Adrenal Hyperplasia

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