

Nebraska Newborn Screening Program Panel Core Conditions¹

Every baby born in Nebraska is required to have testing for the following treatable disorders (Core Conditions). Secondary Conditions may be detected by screening for core conditions)

X: Condition is in this category --: Condition is not in this category

| Core Condition | Metabolic Disorder - Organic Acid Condition | Metabolic Disorder -Fatty Acid Oxidation Disorder | Metabolic Disorder - Amino Acid Disorder | Endocrine Disorder | Hemoglobin Disorder | Other Disorder |
|---|---|---|--|--------------------|---------------------|----------------|
| Propionic Acidemia | X | -- | -- | -- | -- | -- |
| Methylmalonic Acidemia (Methylmalonyl-Coa Mutase) | X | -- | -- | -- | -- | -- |
| Methylmalonic Acidemia (Cobalamin Disorders) | X | -- | -- | -- | -- | -- |
| Isovaleric Acidemia | X | -- | -- | -- | -- | -- |
| 3-Methylcrotonyl-Coa Carboxylase Deficiency | X | -- | -- | -- | -- | -- |
| 3-Hydroxy-3-Methylglutaric Aciduria | X | -- | -- | -- | -- | -- |
| Holocarboxylase Synthase Deficiency | X | -- | -- | -- | -- | -- |
| β-Ketothiolase Deficiency | X | -- | -- | -- | -- | -- |
| Glutaric Acidemia Type I | X | -- | -- | -- | -- | -- |

| Core Condition | Metabolic Disorder - Organic Acid Condition | Metabolic Disorder -Fatty Acid Oxidation Disorder | Metabolic Disorder - Amino Acid Disorder | Endocrine Disorder | Hemoglobin Disorder | Other Disorder |
|--|--|--|---|---------------------------|----------------------------|-----------------------|
| Carnitine Uptake Defect/Carnitine Transport Defect | -- | X | -- | -- | -- | -- |
| Medium-Chain Acyl-Coa Dehydrogenase Deficiency | -- | X | -- | -- | -- | -- |
| Very Long-Chain Acyl-Coa Dehydrogenase Deficiency | -- | X | -- | -- | -- | -- |
| Long-Chain L-3 Hydroxyacyl-Coa Dehydrogenase Deficiency | -- | X | -- | -- | -- | -- |
| Trifunctional Protein Deficiency | -- | X | -- | -- | -- | -- |
| Argininosuccinic Aciduria | -- | -- | X | -- | -- | -- |
| Citrullinemia, Type I | -- | -- | X | -- | -- | -- |
| Maple Syrup Urine Disease | -- | -- | X | -- | -- | -- |
| Homocystinuria | -- | -- | X | -- | -- | -- |
| Classic Phenylketonuria | -- | -- | X | -- | -- | -- |
| Tyrosinemia, Type I | -- | -- | X | -- | -- | -- |
| Primary Congenital Hypothyroidism | -- | -- | -- | X | -- | -- |

| Core Condition | Metabolic Disorder - Organic Acid Condition | Metabolic Disorder -Fatty Acid Oxidation Disorder | Metabolic Disorder - Amino Acid Disorder | Endocrine Disorder | Hemoglobin Disorder | Other Disorder |
|---|--|--|---|---------------------------|----------------------------|-----------------------|
| Congenital Adrenal Hyperplasia | -- | -- | -- | X | -- | -- |
| S,S Disease (Sickle Cell Anemia) | -- | -- | -- | -- | X | -- |
| S, Beta-Thalassemia | -- | -- | -- | -- | X | -- |
| S,C Disease | -- | -- | -- | -- | X | -- |
| Biotinidase Deficiency | -- | -- | -- | -- | -- | X |
| Critical Congenital Heart Disease | -- | -- | -- | -- | -- | X |
| Cystic Fibrosis | -- | -- | -- | -- | -- | X |
| Classic Galactosemia | -- | -- | -- | -- | -- | X |
| Glycogen Storage Disease Type II (Pompe) | -- | -- | -- | -- | -- | X |
| Hearing Loss | -- | -- | -- | -- | -- | X |
| Severe Combined Immunodeficiencies | -- | -- | -- | -- | -- | X |
| Mucopolysaccharidosis Type I | -- | -- | -- | -- | -- | X |
| X-Linked Adrenoleukodystrophy | -- | -- | -- | -- | -- | X |

| Core Condition | Metabolic Disorder - Organic Acid Condition | Metabolic Disorder -Fatty Acid Oxidation Disorder | Metabolic Disorder - Amino Acid Disorder | Endocrine Disorder | Hemoglobin Disorder | Other Disorder |
|--|---|---|--|--------------------|---------------------|----------------|
| Spinal Muscular Atrophy Due To Homozygous Deletion Of Exon 7 In SMN1 | -- | -- | -- | -- | -- | X |

Nebraska Newborn Screening Secondary² Conditions ¹

| Secondary Condition | Metabolic Disorder - Organic Acid Condition | Metabolic Disorder -Fatty Acid Oxidation Disorder | Metabolic Disorder - Amino Acid Disorder | Endocrine Disorder | Hemoglobin Disorder | Other Disorder |
|---|---|---|--|--------------------|---------------------|----------------|
| Methylmalonic Acidemia With Homocystinuria | X | -- | -- | -- | -- | -- |
| Malonic Acidemia | X | -- | -- | -- | -- | -- |
| Isobutyrylglycinuria | X | -- | -- | -- | -- | -- |
| 2-Methylbutyrylglycinuria | X | -- | -- | -- | -- | -- |
| 3-Methylglutaconic Aciduria | X | -- | -- | -- | -- | -- |
| 2-Methyl-3-Hydroxybutyric Aciduria | X | -- | -- | -- | -- | -- |
| Short-Chain Acyl-Coa Dehydrogenase Deficiency | -- | X | -- | -- | -- | -- |
| Medium/Short-Chain L-3-Hydroxyacyl-Coa Dehydrogenase Deficiency | -- | X | -- | -- | -- | -- |
| Glutaric Acidemia Type II | -- | X | -- | -- | -- | -- |
| Medium-Chain Ketoacyl-Coa Thiolase Deficiency | -- | X | -- | -- | -- | -- |
| 2,4 Dienoyl-Coa Reductase Deficiency | -- | X | -- | -- | -- | -- |
| Carnitine Palmitoyltransferase Type I Deficiency | -- | X | -- | -- | -- | -- |
| Carnitine Palmitoyltransferase Type II Deficiency | -- | X | -- | -- | -- | -- |

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|--|---|---|--|--------------------|---------------------|----------------|
| Carnitine Acylcarnitine Translocase Deficiency | -- | X | -- | -- | -- | -- |
| Argininemia | -- | -- | X | -- | -- | -- |
| Citrullinemia, Type II | -- | -- | X | -- | -- | -- |
| Hypermethioninemia | -- | -- | X | -- | -- | -- |
| Benign Hyperphenylalaninemia | -- | -- | X | -- | -- | -- |
| Biopterin Defect in Cofactor Biosynthesis | -- | -- | X | -- | -- | -- |
| Tyrosinemia, Type II | -- | -- | X | -- | -- | -- |
| Tyrosinemia, Type III | -- | -- | X | -- | -- | -- |
| Various Other Hemoglobinopathies | -- | -- | -- | -- | X | -- |
| Galactosepimerase Deficiency | -- | -- | -- | -- | -- | X |
| Galactokinase Deficiency | -- | -- | -- | -- | -- | X |
| T-Cell Related Lymphocyte Deficiencies | -- | -- | -- | -- | -- | X |

1. Nomenclature for Conditions based upon "Naming and Counting Disorders (Conditions) Included in Newborn Screening Panels." *Pediatrics*. 2006; 117 (5) Suppl: S308-S314.

2. Disorders that can be detected in the differential diagnosis of a core disorder.