Newborn Screening in Nebraska
REQUIRED TESTS

Every baby born in Nebraska is required to have testing for the following treatable disorders.

Condition	Testing for	Effects if Not Treated	Treatment
Biotinidase Deficiency (BIO)	p-aminobenzoate	Developmental disabilities, seizures, deafness, blindness, skin rash	Daily oral Rx Biotine
Congenital Adrenal Hyperplasia (CAH)	Steroid 17-alpha hydroxyprogesterone levels/reflex to extracted 17-OHP on subset of specimens	Variable: ambiguous genitalia, adrenal "salt-wasting" crisis with possible mortality	Steroid replacement and monitoring by Pediatric Endocrinologist and appropriate emergency intervention
Congenital Primary Hypothyroidism (CPH)	Thyroxin (T_4) reflex to Thyroid Stimulating Hormone (TSH) on T_4's in lowest 10%	Severe developmental disabilities and growth	Thyroid Hormone treatment
Cystic Fibrosis (CF)	Immunoreactive trypsinogen (IRT) levels/reflex to DNA on subset of specimens	Variable: pancreatic insufficiency, failure to thrive, decreased pulmonary function, respiratory infection risk, possible mortality	Management by Accredited CF Center Team
Galactosemia (GAL)	Total galactose & uridyl-transferase	Septicemia, cataracts, developmental disabilities, cirrhosis, ovarian failure, death if untreated	Lactose-free diet
Severe Combined Immune Deficiency (SCID)	T-cell receptor excision circles as a marker of T-cell production.	May suffer from repeated infections, death if untreated	Early intervention to prevent infection. Bone marrow stem cell transplant.
Hemoglobinopathies Including 1) Sickle Cell Disease,Sickle, 2) Hemoglobin C Disease, and 3) Sickle Beta Thalassemia	Hemoglobins F A S & C, D, E O-Arab Reflex to DNA	Anemia, septicemia, painful crisis, acute chest syndrome, splenomegaly, stroke, high mortality rate	Penicillin prophylaxis, folic acid, parent education and counseling.
Fatty Acid Conditions including: - **Carnitine Update Defect (CUD)** - **Medium Chain Acyl Co-A Dehydrogenase Deficiency (MCAD)** - **Long-chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)** - **Trifunctional Protein Deficiency (TFP)**	Acylcarnitine Profile	Hypoglycemia, vomiting, coma, possible seizures & possible death. Possible developmental disability if survive metabolic crisis.	Prevent fasting, follow low-fat diet and carnitine supplements. If illness presents, hospitalization to prevent metabolic crisis.

- **Very Long-chain Acycl-CoA Dehydrogenase Deficiency (VLCAD)**			
AMINO ACID CONDITIONS including: - **Argininosuccinic Acidemia (ASA)** - **Citrullinemia (CIT)** - **Homocystinuria (HCY)** - **Isovaleric Acidemia (IVA)** - **Maple Syrup Urine Disease (MSUD)** - **Methylmalonic Acidemia (MUT) or (Cbl A and B)** - **Phenylketonuria (PKU)** - **Propionic Acidemia (PA)** - **Tyrosinemia (TYR)** - **3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)**	Amino Acid Profile	Varies depending on condition. Failure to thrive, metabolic acidosis, vision problems, skeletal problems, severe developmental disabilities, seizures, and possibly death.	Special metabolic formula and diet.
ORGANIC ACID CONDITIONS including: - **Beta-ketothiolase Deficiency (BKT)** - **Glutaric Acidemia type 1 (GA1)** - **3-Hydroxy 3-Methyl Glutaric Aciduria (HMG)**	Amino Acid and Acylcarnitine profies	Metabolic crisis which includes: very low blood sugar, vomiting, possible seizures, coma and possible death Developmental problems may occur if the child has and survives the above. May also include heart problems.	Close monitoring with a metabolic specialist, special formulas, and diet.
VITAMIN METABOLISM CONDITIONS including: - **Multiple Carboxylase Deficiency (MCD)** - ***also listed above (Cbl A and B) amino acid and vitamin disorder** - ***also listed above (Biotinidase Deficiency is also in this category but not screened by tandem mass spectrometry)**	Amino Acid and Acyclcarnitine profiles Beutler and Baluda Enzyme Reation Units	Varies by condition but can include, mental retardation, seizures, nerve and brain cell damage and possibly death.	Vitamin supplementation (pharmaceutical doses) and monitoring by metabolic specialist.

LYSOSOMAL STORAGE DISEASES including: **- Pompe Diseases (PD)** **- Mucopolysaccharidosis Type 1 (MPS-1)**	Acid Alpha Glycosidase (GAA) enzyme for Pompe disease, and reflex to sequencing. Alpha-L-Iduronidase (IDUA) enzyme for MPS-1 and reflex to sequencing on repeat inconclusive results.	Pompe: Variable forms but can result in damage to cardiac & skeletal muscle. Without treatment leads to death. MPS-1: Neurocognitive impairment, skeletal abnormalities, heart & lung disease hepatosplenomegaly, hearing loss, corneal clouding.	Enzyme replacement therapy, and/or bone marrow stem cell transplant
PEROXISOMAL STORAGE DISEASE including: **- X-Linked Adrenoleukodystrophy (X-ALD)**	Very long chain fatty acid lysophosphatidylcholine (C26:0) and reflex to sequencing on repeat inconclusive results.	Variable forms but most severe childhood cerebral form suffer adrenal cortex damage, demyelination and progressive neurological symptoms.	Adrenal hormone replacement and for the most severe Childhood Cerebral form, hematopoietic stem cell transplant.