

APPENDIX 1. EXPANDED NEWBORN SCREENING

Category	Disorders, Description, Prevalence	Common Symptoms	Treatment
Amino Acid Disorders	Argininosuccinic acidemia (ASA), Phenylketonuria (PKU), Tyrosinemia (TYR) Type 1, Homocystinuria (HCY), Citrullinemia (CIT) Type 1, Maple syrup urine disease (MSUD) Body cannot either metabolize or produce certain amino acids, resulting in toxic accumulation of substances Estimated prevalence: 1/5,100	Example—PKU: Hyperphenylalaninemia (neurotoxic), microcephaly, epilepsy, mental retardation, behaviour problems	Example—PKU: Diet: reduce phenylalanine, low protein, supplement cofactors or essential amino acids Avoidance of fasting
Endocrine Disorders	Congenital Hypothyroidism (CH): Caused by anatomic defect in gland or inborn error of metabolism. Iodine deficiency results in inadequate thyroid hormone production Estimated prevalence: 1/1,300–4,000 Congenital Adrenal Hyperplasia (CAH): Impaired synthesis of cortisol by adrenal cortex leads to increased androgen biosynthesis, causing inability to maintain energy & blood glucose levels to meet stress of injury and illness Estimated prevalence: 1/15,000	CH: mental retardation, reduced bone/growth maturation, neurologic problems: spasticity, abnormal gait, dysarthria, autistic behaviour CAH: virilization, precocious puberty, infertility, short stature, renal salt wasting leads to failure to thrive, vomiting, dehydration, hypotension, hyponatremia, hyperkalemia	CH: Thyroid hormone replacement CAH: Glucocorticoid replacement therapy
Fatty Acid Oxidation Disorders	Medium chain acyl-CoA dehydrogenase (MCAD) deficiency, Very long chain 3-Hydroxyacyl-CoA dehydrogenase (VCLAD) deficiency, Long chain 3-Hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency, Trifunctional protein (TFP) deficiency, Carnitine uptake defect (CUD) Body is unable to break down fatty acids (an essential part of body's ability to produce energy) Estimated prevalence: 1/10,000–15,000	Decompensation with any catabolic stress—fever, fasting, intercurrent illness, hypoketotic hypoglycemia, Liver, muscle, heart disease, lethargy, seizures, coma, death, SIDS	Avoidance of fasting Frequent feeding: increase carbohydrates, decrease fats Supplements: carnitine, cornstarch
Hemoglobinopathies	Sickle cell disease (Hb SS), SC disease (Hb SC), β -sickle thalassemia (HbS/ β th) Body is unable to produce normal hemoglobin Estimated prevalence: Depends on ethnic background	Example—Sickle cell disease: Painful vaso-occlusive crises, hemolytic anemia, frequent infections, tissue ischemia, chronic organ dysfunction	Example—Sickle cell disease: Prophylactic penicillin Vaccinations: pneumococcal, influenza
Organic Acid Disorders	Isovaleric acidemia (IVA), Glutaric acidemia type 1 (GA1), HMG-CoA lyase deficiency (HMG), Multiple carboxylase deficiency (MCD), Methylmalonic acidemia (MUT, Cbl A,B), 3 methylcrotonyl-CoA carboxylase (MCC) deficiency, Propionic acidemia (PROP), β -ketothiolase (BKT) deficiency Body cannot metabolize certain amino acids & fats resulting in accumulation of organic acids in blood & urine Estimated prevalence: 1/15,000–55,000	Acute encephalopathy, vomiting, metabolic acidosis, ketosis, hyperammonemia, hypoglycemia, coma, dehydration, failure to thrive, hypotonia, global developmental delay, sepsis, death	Low protein diet/amino acid restriction Supplements: carnitine, biotin, riboflavin, glycine Avoidance of fasting
Other	Biotinidase deficiency (BIOT): Body is unable to recycle biotin (a cofactor for 4 dependent carboxylases) Estimated prevalence: 1/120,000 Galactosemia (GALT): Body is unable to break down galactose (a metabolite of lactose) Estimated prevalence: 1/40,000 Cystic fibrosis (CF): Body has problem with sodium chloride channels resulting in a multisystem disorder Estimated prevalence: 1/1,300	BIOT: Metabolic ketoacidosis, organic aciduria, mild hyperammonemia, seizures, hypotonia, ataxia, developmental delay, vision problems, hearing loss, cutaneous abnormalities GALT: Feeding problems, failure to thrive, bleeding, infection, liver failure, cataracts, MR CF: Chronic sinopulmonary disease, GI/nutritional abnormalities, azoospermia (males), salt loss syndrome	BIOT: 5–10 mg oral biotin/day GALT: Lactose-galactose-restricted diet (must be started in first 10 days of life to prevent symptoms) CF: antibiotics, bronchodilators, anti-inflammatories, mucolytics, physiotherapy, nutritional therapy

Sources:

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