Medical Conditions on the Oregon Newborn Bloodspot Screening Panel

Updated June 2022

Medical Condition	Incidence in NW region
Organic Acid Disorders	
Propionic acidemia (PA)*	1 per 271,000
Methylmalonic acid(MMA)*	1 per 95,000
Isovaleric acidemia (IVA)	1 per 148,000
3-methylcrotonyl CoA carboxylase deficiency (3MCC)	1 per 51,000
3-hydroxy-3-methylglutaryl CoA lyase deficiency (HMG)	Rare, less than 1 per 300,000
Multiple carboxylase deficiency (MCD)	Rare, less than 1 per 300,000
Beta-ketothiolase deficiency (BKT)	Rare, less than 1 per 1 million
2-methyl-3-hydroxybutyryl CoA dehydrogenase deficiency (2M3HBA)	Rare, less than 1 per 1 million
Glutaric acidemia, type 1 (GA-1)	1 per 85,000
Malonic acidemia (MAL)	Rare, less than 1 per 300,000
Isobutyrl-CoA dehydrogenase deficiency (IBD)	Rare, less than 1 per 300,000
2-methylbutyryl CoA dehydrogenase deficiency (2MBC)	1 per 181,000 (Hmong have higher incidence)
3-methylglutaconyl CoA hydratase deficiency (3MGH)	Rare, less than 1 per 1.3 million
Fatty Acid Oxidation Disorders	
Carnitine uptake deficiency (CUD)	1 per 116,000
Medium chain acyl-CoA dehydrogenase deficiency (MCAD)*	1 per 19,000
Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)*	1 per 62,500
Long chain 3 hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)*	1 per 541,000
Trifunctional protein deficiency (TFP)	Very rare. Incidence unknown

Medical Condition	Incidence in NW region
Short chain acyl-CoA dehydrogenase deficiency (SCAD)	1 per 81,000
Glutaric acidemia type II, also known as Multiple acyl-CoA dehydrogenase deficiency (MADD)	1 per 541,000
Carnitine palmitoyl transferase deficiency, type I (CPT-I)	1 per 812,000
Carnitine palmitoyl transferase deficiency, type II (CPT-II)*	1 per 400,000
Carnitine acylcarnitine translocase deficiency (CACT)	Very rare. Incidence unknown.
Amino Acid Disorders	
Arginoinosuccinate lyase deficiency (Arginosuccinicaciduria; ASA)*	1 per 125,000
Citrullinemia, type I (CIT)*	1 per 325,000
Maple syrup urine disorder(MSUD)*	1 per 271,000
Homocystinuria (HCY)	1 per 203,000
Phenylketonuria (PKU)	1 per 28,500
Tyrosinemia, type I	1 per 812,000
Tyrosinemia, type II andtype III	1 per 652,000
Arginase deficiency (ARG)	1 per 1.6 million
Endocrine Disorders	
Primary congenital hypothyroidism	1 per 2,300
Congenital adrenal hyperplasia (CAH)*	1 per 12,700
Pulmonary Disorders	
Cystic fibrosis (CF)	1 per 6,500
Other Metabolic Disorders	
Biotinidase deficiency	1 per 1.05 million
Classic galactosemia(GALT)*	1 per 95,000

Medical Condition	Incidence in NW region
Hemoglobin Disorders	
Sickle cell disease	1 per 10,000 (1 per 365 in Black orAfrican Americans)
Immunology Disorders	
Severe combined immunodeficiency (SCID)	1 per 50,000 to 1 per 100,000
Lysosomal Storage Disorders	
Pompe* (glycogen storagedisease Type II)	1 per 28,000
MucopolysaccharidosisType I (MPS I)*	Between 1 per 87,000 and 1 per 185,000
Fabry	Between 1 per 1,500 and 1 per 13,000
Gaucher*	1 per 57,000
Other Conditions	
Spinal muscular atrophy(SMA)†	1 per 11,000
X-linked adrenoleukodystrophy (X-ALD)‡	1 per 4,845

* Infants may have severe neonatal presentation.

† Screening for this condition is anticipated to begin June 1, 2022.

‡ Screening for this condition is anticipated to begin on or before January 1, 2023.

Newborn bloodspot screening may identify other related medical conditions that are not listed above. Information regarding these related conditions can be found in the relevant condition sections below. It is within the discretion of an infant's health care provider and parents or legal guardians to determine what, if any, medical follow-up is needed in these circumstances.