

Table I Summary of Conditions on the Screening Panel

Table I summarizes the conditions on the Oregon screening panel, including the incidence, symptoms, and treatment.

Disorders	Analyte Tested For	Incidence in NW Region	Symptoms if Not Treated	Treatment
Cystic fibrosis	Immunotrypsinogen (IRT)		Lung disease; growth failure	Pulmonary enzyme replacement therapy
Congenital adrenal hyperplasia (CAH)*	17-OH-Progestrone	1:12,000 1:300 in Yupik Eskimos	Addisonian crisis/salt wasting in 2/3 infants; dehydration; shock; hyperkalemia; virilization of females	Glucocorticoid and/or mineralocorticoid (Florinef)
Congenital hypothyroidism	Thyroid hormones (T4 with TSH confirmation)	1:3,000	Mental retardation, other brain damage; growth delay	Thyroid hormone (L-Thyroxine)
Hemoglobinopathies including sickle cell anemia	Hemoglobin patterns	1:15,000 (1:400 in African Americans)	In sickle cell disease: death by sepsis or splenic sequestration anemia; sickling crisis	Penicillin & comprehensive care
Biotinidase deficiency	Biotinidase	1:60,000	Mental retardation; seizures; skin rash; alopecia; hearing loss; death	Biotin
Galactosemia	Galactosemia enzyme (GALT)	1:60,000	Severe brain damage; liver disease; cataracts; death	Galactose-restricted diet
Argininase deficiency	Arginine	1:350,000	Irritability; developmental delay; spastic tetraplegia	Low protein diet, medication
Arginosuccinate lyase deficiency (ASA)*	ASA/Citrulline	1:88,000	Hyperammonemia; mental retardation; seizure; death	Low protein diet, medication
Arginosuccinate synthetase deficiency (Citrullinemia)*	Citrulline	1:71,000	Hyperammonemia; mental retardation; seizure; death	Low protein diet, medication
Homocystinuria	Methionine	1:118,000	Mental retardation; dislocation of lenses; marfanoid body habitus	Pyridoxine; methionine restricted, cysteine supplemented diet
Hyperphenylalaninemia, including phenylketonuria (PKU)	Phenylalanine	1:13,600	Profound mental retardation; seizures	Low phenylalanine diet
Tyrosinemia	Tyrosine	1:350,000	Vomiting, lethargy; liver disease; coagulopathy; renal tubular acidosis	Medication; low phenylalanine and/or low tyrosine diet

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Table I (continued)

Disorders	Analyte Tested For	Incidence in NW Region	Symptoms if Not Treated	Treatment
Beta-ketothiolase deficiency (BKD)	C5:1, C5OH	>1:300,000	Severe bouts of acidosis possibly resulting in mental retardation, death	IV support during episodes, bicarbonate supplement
Glutaric acidemia, Type I (GA I)	C5DC	1:71,000	Metabolic crisis; damages basal ganglia	IV support for brain illness
Isobutyryl CoA dehydrogenase deficiency (IBD)	C4	>1:300,000	None to cardiomyopathy	Carnitine therapy, protein restriction, avoid fasting
Isovaleric acidemia (IVA)	C5	1:120,000	Vomiting; lethargy; acidosis possibly resulting in coma, death	Protein restriction
Malonic aciduria	C3DC	>1:300,000	Mental retardation	Carnitine therapy, protein restriction
Maple syrup urine disease (MSUD)*	Leucine	1:350,000	Vomiting; lethargy; acidosis possibly resulting in death	Protein restriction
Methylmalonic acidemias (MMA/8 types)*	C3, C3/C2	1:50,000	Vomiting; lethargy; acidosis possibly resulting in death	Protein restriction; carnitine therapy, B-12
Multiple Carboxylase Deficiency (MCD)	C3, C5OH	>1:300,000	Hypotonia, seizures, skin rash, alopecia, lactic acidosis, brain damage. Average age at presentation: birth-18 months	Biotin
Propionic acidemia (PA)*	C3, C3/C2	>1:300,000	Vomiting; lethargy; acidosis possibly resulting in death	Protein restriction; carnitine therapy
3-hydroxy-3-methylglutaryl CoA lyase deficiency (HMG)	C5OH	>1:300,000	Hypoglycemia; acidosis possibly resulting in death; may be asymptomatic	Protein restriction
2-methyl-3-hydroxybutyryl CoA dehydrogenase deficiency (MHBD)	C5:1, C5OH	1:350,000	Asymptomatic 9–14 mos. Then severe mental and motor skill loss	Protein restriction
2-methylbutyryl CoA dehydrogenase deficiency (2MBC)	C5	>1:300,000	Hypoglycemia; mental retardation; may be asymptomatic	Avoid fasting, protein restriction
3-methylcrotonyl CoA carboxylase deficiency (3MCC)	C5OH	1:51,000	Most have been asymptomatic	None or protein restriction

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Disorders	Analyte Tested For	Incidence in NW Region	Symptoms if Not Treated	Treatment
3-methylglutaconyl CoA hydratase deficiency (3MGH)	C5OH	>1:300,000	Hypoglycemia; acidosis; may be asymptomatic	Protein restriction, avoid fasting
Short chain acyl-CoA dehydrogenase deficiency (SCAD)	C4	1:350,000	Most asymptomatic; hypotonia, mental retardation	Avoid fasting
Medium chain acyl-CoA dehydrogenase (MCAD)*	C6, C8, C10, C8/C10	1:11,000	Hypoglycemia possibly resulting in coma, death; may be asymptomatic	Avoid fasting, carnitine supplement
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)	C14:1, C16, C16OH, C18, C18OH,	1:177,000	Hepatic dysfunction; hypoglycemia; failure to thrive	Long chain fatty acid restriction, Medium Chain Triglycerides (MCT) oil, carnitine, avoid fasting
Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)	C14, C14:1, C16, C16:1, C18, C18:1,	1:71,000	Hypoglycemia with or without cardiomyopathy; muscle fatigue	Avoid fasting, low fat diet with MCT oil
Carnitine uptake/transport defects	C0, C16, C18	1:118,000	Hypoglycemia; cardiomyopathy	Avoid fasting, low fat diet, carnitine supplement
Carnitine palmitoyl transferase I deficiency (CPT I)	C0/C16+C18	Increased in Hutterite and Alaska native populations	Hypoketotic hypoglycemia, brought on by fasting or intercurrent illness. Average age at presentation: birth-18 months	Avoid fasting and long chain fatty acids; MCT oil supplement
Carnitine palmitoyl transferase II deficiency (CPT II)	C0, C4, C5, C6, C14, C16, C16:1, C18, C18:1	>1:300,00	Muscle weakness, pain and myoglobinuria leading to renal failure in 25%. Average age at presentation: 15-30 yrs; also a severe neonatal form usually lethal with multiple congenital anomalies	Avoid fasting and severe exercise; MCT oil supplement
Multiple acyl-CoA dehydrogenase deficiency (MADD)	C4, C5, C6, C8, C10, C14, C16, C18:1	>1:300,000	Multiple congenital abnormalities; acidosis, hypoglycemia	Low fat diet, avoid fasting, not effective in severe neonatal form

* Infants may have severe neonatal presentation.