Newborn Screening

Screened disorder	Incidence	Cause	Possible outcome without treatment
Phenylketonuria (PKU)	1:25,000	Inability to process phenylalanine.	Severe mental retardation
Maple Syrup Urine Disease (MSUD)	<1:100,000	Ineffective protein metabolism.	Mental retardation, death
Homocystinuria (HCY)	<1:100,000	Lacking enzyme homocysteine.	Mental retardation, eye problems, skeletal abnormalities, stroke
Citrullinemia (CIT)	<1:100,000	Cirulline, ammonia build-up in the body.	Seizures, coma, brain damage, death.
Argininosuccinic acidemia (ASA)	<1:100,000	Argininosuccinc acid and ammonia build-up in body.	Neurologic damage, death.
Tyrosinemia type I (TYR I)	<1:100,000	Lack of enzyme leads to toxic build-up of amino acid byproduct in liver.	Liver and kidney failure, nerve damage, death.
Isovaleric acidemia (IVA)	<1:100,000	Inability to process amino acid leucine.	Neurological damage, death.
Glutaric acidemia type I (GA I)	>1:75,000	Enzyme deficiency.	Seizures, cerebral-palsy like symptoms, death.
Hydroxymethylglutaric aciduria or HMG-CoA lyase, or 3-OH 3-CH3 glutaric aciduria (HMG)	<1:100,000	Inability to process amino acid leucine.	Brain damage, coma, death
Multiple carboxylase deficiency (MCD)	<1:100,000	Enzyme deficiency, leads to lactic acid build-up.	Brain damage, coma, death
Methylmalonic acidemia due to mutase deficiency (MUT)	>1:75,000	Inability to process four amino acids.	Brain damage, death.
Methylmalonic acidemia cblA and cblB forms (Cbl A, B)	<1:100,000	Defective vitamin metabolism.	Brain damage, seizures, paralysis, coma, death
3-methylcrotonyl-CoA carboxylase deficiency (3MCC)	>1:75,000	Ineffective leucine processing.	Brain damage, seizures, liver failure, death
Propionic acidemia (PROP)	>1:75,000	Ineffective processing of four amino acids.	Brain damage, coma, death
Beta-Ketothiolase deficiency (BKT)	<1:100,000	Acid build-up	Brain damage, coma, death
Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)	>1:25,000	Lack of enzyme affects conversion of fat to energy.	Seizures, liver failure, coma, death
Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)	>1:75,000	Lack of enzyme affects conversion of fat to energy.	Heart and liver failure, death
Long-chain 3-OH acyl-CoA dehydrogenase deficiency (LCHAD)	>1:75,000	Lack of enzyme affects conversion of fat to energy.	Heart, lung and liver failure, death
Trifunctional protein deficiency (TFP)	<1:100,000	Lack of enzyme affects conversion of fat to energy.	Low muscle tone, seizures, heart failure, coma, death

Carnitine uptake defect (CUD)	<1:100,000	Inability to transport carnitine.	Hypoglycemia, death
Sickle cell anemia (Hb SS)	>1:5,000, 1:400 in African Americans	Abnormally shaped hemoglobin.	Pain, vital organ damage, susceptibility to bacterial infection, stroke, death
Hb S/Beta-Thalassemia (Hb S/Th)	>1:50,000	Child inherits one sickle cell gene and one beta-thalassemia gene (another inherited anemia).	Milder versions of sickle cell outcomes
Hb S/C disease (Hb S/C)	>1:25,000	Child inherits one sickle cell gene and one gene for another abnormal hemoglobin, HbC.	Milder versions of sickle cell outcomes
Congenital hypothyroidism (CH)	>1:5,000	Thyroid hormone deficiency.	Slowed growth and brain development
Biotinidase deficiency (BIOT)	>1:75,000	Enzyme deficiency.	Frequent infections, uncoordinated movement, hearing loss, seizures, mental retardation, coma, death
Congenital adrenal hyperplasia (CAH)	>1:25,000	Defective synthesis of adrenal hormones.	Masculinization of female genitals, life-threatening salt loss
Classical galactosemia (GALT)	>1:50,000	Missing enzyme to convert galactose to glucose.	Blindness, severe mental retardation, infection, death
Cystic fibrosis (CF) (As of 7/1/07)	>1:5,000	Genetic disease that affects mucous production.	Lung, digestive problems

See these websites for more information:
http://www.newbornscreening.info/Parents/facts.html
http://www.marchofdimes.com/professionals/14332_15455.asp