

MICHIGAN NEWBORN SCREENING DISORDERS

Disorder/Year Screening Began	Michigan Incidence	Goal of Screening	Inheritance	Signs, Symptoms, and Long Term Effects if Not Treated	Treatment
Phenylketonuria (PKU) 1965	1 in 8,801	Prevention of mental retardation	Autosomal recessive disorder	Normal at birth. Neurologic disturbances such as epilepsy. Undiagnosed children may have an unpleasant musty smell. 90% have blond hair and blue eyes. May also have eczema. Brain damage and mental retardation.	Low phenylalanine diet for life through the use of special formula and low protein food products
Congenital Hypothyroidism (CH) 1977	1 in 1,942	Prevention of mental retardation and poor growth and development	Most cases are sporadic (85%), but some may be inherited (15%)	No symptoms until 3-4 months of age; irreversible brain damage; prolonged jaundice, growth failure, lethargy, poor appetite, and constipation. Mental retardation, growth failure, and neurological problems when treatment is delayed.	Oral thyroid hormone (replacement) administered daily
Galactosemia 1984	1 in 41,227	Prompt identification and treatment of affected infants to prevent neurological damage and death	Autosomal recessive disorder	Jaundice, diarrhea, vomiting, and failure to gain weight within first week of life. Liver disease, cataracts, mental retardation, and death if not detected immediately. Some children may have speech delays and females typically have ovarian failure even with treatment.	Immediate change to soy formula and lifelong exclusion of galactose from the diet
Maple Syrup Urine Disease (MSUD) 1987	1 in 234,992	Prompt identification and treatment of affected infants to prevent neurological damage and death	Autosomal recessive disorder	Poor feeding, lethargy, convulsions, and death. Sweet, burnt sugar, or maple syrup smell of urine. Amino acids accumulate in the blood causing a toxic effect that interferes with brain function.	Lifelong strict diet limiting the intake of branched chain amino acids through the use of special formula and low protein food products
Biotinidase Deficiency 1987	1 in 27,325	Prevention of neurological damage and death	Autosomal recessive disorder	Normal at birth. Hypotonia, ataxia, seizures, developmental delay, alopecia, seborrheic dermatitis, hearing loss and optic nerve atrophy. Metabolic acidosis can result in coma and death.	Lifelong daily oral biotin supplement

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Hemoglobinopathies Sickle Cell Diseases 1987	1 in 1,956 overall. 1 in 600 African American newborns. Also seen in individuals of Indian, Mediterranean and Middle Eastern heritage.	Prevention of death from pneumococcal sepsis by instituting penicillin prophylaxis	Autosomal recessive disorders of the adult β -hemoglobin chain	Newborns may develop septicemia and die within the first year of life. Extremely high mortality for children under the age of five years.	Penicillin prophylaxis should begin as soon as possible and continue until six years of age
Congenital Adrenal Hyperplasia (CAH) 1993	1 in 17,716	Prompt identification and treatment of affected infants to prevent death from adrenal crisis or shock, and correct sex assignment in female newborns	Autosomal recessive disorders of adrenal steroidogenesis	<u>Classic</u> : ambiguous genitalia in a newborn girl resulting in incorrect sex assignment; severe salt and hormonal imbalances in both girls and boys. Without treatment, heart failure and death from adrenal crisis or shock may occur within the first weeks of life. <u>Non-classic</u> : severe acne, excess facial and/or body hair, early development of pubic hair, receding scalp hairline, menstrual disturbances, and infertility in both males and females.	Administration of Glucocorticoid and salt retaining hormones. Surgical correction of ambiguous genitalia
Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD) 2003	~1 in 26,205	Prevention of hypoglycemia that could lead to coma, encephalopathy, liver failure or death	Autosomal recessive disorder	Some individuals: recurrent episodes of metabolic acidosis, hypoglycemia, lethargy, and coma. Mental retardation and death may occur. More than 50% of MCADD individuals die from their first crisis if it occurs after age two.	Lifelong low-fat diet and avoidance of fasting. Carnitine supplements are also used. Acute episodes are managed by the administration of intravenous glucose
Arginosuccinic Aciduria (ASA) October 2004	Not yet determined	Prevention of neurological damage and death	Autosomal recessive disorder	Hyperammonemia accompanied by lack of appetite, vomiting, listlessness, seizures, and coma. Brain damage and death.	High caloric, protein restrictive diet. Sodium Benzoate and sodium phenylacetate supplements may be used.

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Citrullinemia October 2004	Not yet determined	Prompt identification and treatment to prevent neurological damage and death	Autosomal recessive disorder	Hyperammonemia accompanied by lack of appetite, vomiting, listlessness, seizures, and coma. Brain damage and death.	High caloric, protein restrictive diet. Sodium Benzoate and sodium phenylacetate supplements may be used. Medication to remove waste products from the blood. Dialysis may be necessary.
Homocystinuria October 2004	Not yet determined	Prevention of mental retardation, seizures, optical lens dislocation, osteoporosis, scoliosis, and/or thrombi formation.	Autosomal recessive disorder	Mental retardation, seizures, psychiatric disturbances, developmental delay, displacement of the lens of the eye (ectopia lentis), abnormal thinning and weakness of the bones (osteoporosis and scoliosis), and/or the formation of blood clots (thrombi) in various veins and arteries. Approximately 50% die before age 25 if not treated. Optical lens dislocation may occur even if treatment is begun early.	Lifelong strict diet limiting the intake of methionine through the use of special infant formulas. Cystine and vitamin B6 supplements may be used.
Cystic Fibrosis (CF) October 2007	1 in 3,000	Prompt identification and treatment of affected infants to prevent development of pulmonary disease and poor growth and development	Autosomal recessive disorder	Normal at birth, intestinal obstruction (meconium ileus), pneumonia, failure to thrive. May also have jaundice, dehydration	High calorie diet. Medications to dilate air way and remove mucus from lung. Treatment of pneumonia, pulmonary exacerbation and pancreatic insufficiency



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