*Incidence: less than 1 in 100,000(1)*

Most commonly, symptoms begin in the first few days of life, with build-up of argininosuccinic acid and ultimately ammonia resulting in brain swelling, coma and, sometimes, death. Survivors often suffer permanent neurological damage. Other affected children may develop symptoms later in infancy or childhood. Early diagnosis and treatment can be lifesaving; however, in spite of treatment, affected individuals remain susceptible to episodes of ammonia build-up, and most have some degree of brain damage. Treatment consists of a low-protein diet, avoiding fasting, medications to prevent ammonia build-up, nutritional supplements, and in some cases, liver transplant.

## Beta-Ketothiolase deficiency = BKT

*Incidence: less than 1 in 100,000(1)*

Periodic episodes of acid build-up, often triggered by some childhood illness, can progress to coma, brain damage and death. These serious consequences are most often seen in infants. With early diagnosis and prompt intravenous treatment to keep blood sugar levels up and blood acid levels down during an illness, children can develop normally. Parents must be alert to early signs of illness. Additional treatments may vary, but can include avoidance of protein-rich diets and long-term treatment with bicarbonate.

## Argininosuccinic acidemia = ASA

*Incidence: less than 1 in 100,000(1)*

This defect in processing the amino acid leucine can lead to brain damage, seizures, liver failure and death in infancy or no symptoms at all into adulthood. Symptoms often develop following a childhood illness. Treatment with a low-protein diet and, in some cases, nutritional supplements may be helpful. (An abnormal result by newborn screening could be related to abnormal metabolites in the mother and not the baby. This will be clarified by further diagnostic testing of the infant.)

## 3-Methylcrotonyl-CoA carboxylase deficiency = 3MCC

*Incidence: greater than 1 in 75,000(1)*

Biotinidase is the enzyme that recycles the vitamin biotin. An inherited deficiency of this enzyme may cause serious complications, including frequent infections, uncoordinated movement, hearing loss, seizures, and mental retardation. Undiagnosed and untreated, the deficiency can lead to coma and death. If the condition is detected soon after birth, these problems can be completely prevented with daily oral doses of biotin.

## Biotinidase deficiency = BIOT

*Incidence: less than 1 in 100,000(1)*

Affected babies are missing the liver enzyme needed to convert galactose, a major sugar from the breakdown of lactose in milk, into glucose, another simple sugar that the body can use. Galactose then accumulates in and damages vital organs, leading to blindness, severe mental retardation, infection, and death. Milk and other dairy products must be eliminated from the baby's diet for life. Though treatment dramatically improves the outlook for affected infants, there is still some risk of developmental delays.

## Carnitine uptake defect = CUD

*Incidence: greater than 1 in 50,000(1)*

Due to a missing transporter, cells cannot bring in carnitine from the blood. Carnitine is needed for the transfer of fatty acids across the membranes of the mitochondria (cellular organelles that produce energy for the cell). Symptoms include episodes of hypoglycemia (low blood sugar) and sudden unexpected death in infancy. Older children may present with progressive heart failure. Early diagnosis and treatment with carnitine permits normal development.

## Citrullinemia = CIT

*Incidence: less than 1 in 100,000(1)*

Build-up in the body of citrulline and ultimately ammonia can begin during the newborn period or later in infancy. Without treatment, seizures, coma, brain damage and death can result. With early diagnosis and treatment, normal development is possible. Treatment includes a low-protein diet, medications to rid the body of amino groups to prevent ammonia build-up, and nutritional supplements.

## Classical galactosemia = GALT

*Incidence: greater than 1 in 50,000(1)*

Cystic fibrosis is one of the most common inherited disorders in the U.S. Abnormalities in the cystic fibrosis protein result in lung and digestive problems, and death at an average age of 30-35 years. Studies suggest that early diagnosis and treatment improves the growth of babies and children with CF. Treatment varies depending on severity of symptoms, but may include a high-calorie diet supplemented with vitamins and medications to improve digestion, respiratory therapy to help clear mucus from the lungs, and medications to improve breathing and prevent lung infections.

## Cystic fibrosis = CF

*Incidence: greater than 1 in 5,000(1)*

This thyroid hormone deficiency severely retards both growth and brain development. If detected soon after birth, the condition can be treated simply with oral doses of thyroid hormone to permit normal development.

## Congenital hypothyroidism = CH

*Incidence: greater than 1 in 5,000(1)*

CAH refers to a set of inherited disorders resulting from defects in the synthesis of hormones produced by the adrenal gland. In female infants, CAH sometimes results in masculinization of the genitals. Certain severe forms of CAH cause life-threatening salt loss from the body if undetected and untreated. Treatment includes salt replacement and hormone replacement.

## Congenital adrenal hyperplasia = CAH

*Incidence: greater than 1 in 25,000(1)*

Periodic episodes of acid build-up, often triggered by some childhood illness, can progress to coma, brain damage and death. These serious consequences are most often seen in infants. With early diagnosis and prompt intravenous treatment to keep blood sugar levels up and blood acid levels down during an illness, children can develop normally. Parents must be alert to early signs of illness. Additional treatments may vary, but can include avoidance of protein-rich diets and long-term treatment with bicarbonate.


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# Alabama Newborn Screening Division


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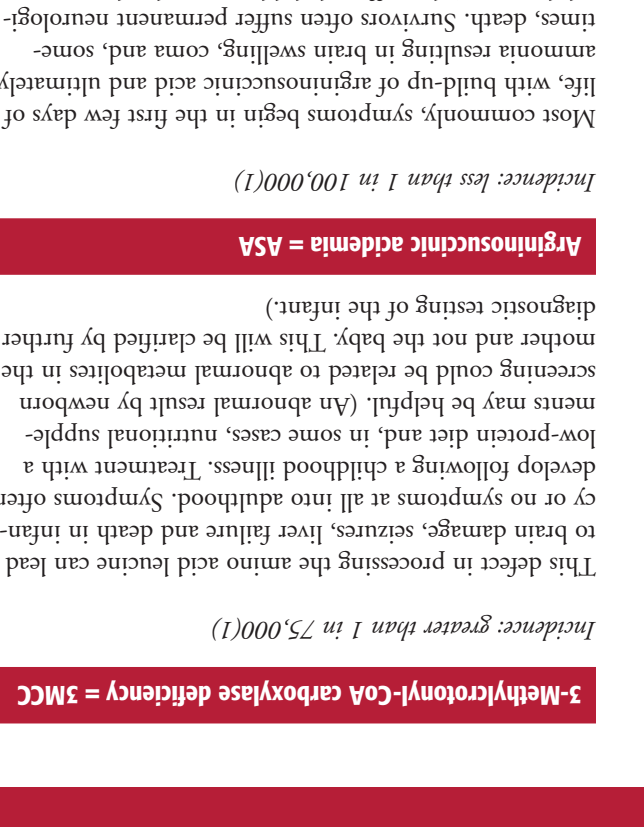
Alabama Department of Public Health
Bureau of Family Health Services

ADPH-FHS-533/Rev. 7-07-kw


# NEWBORN *screening*



*definitions for 29 core conditions*


Alabama Department of Public Health
Bureau of Family Health Services

## Glutaric acidemia type I = GA1

*Incidence: greater than 1 in 75,000(1)*

Babies may develop normally for up to 18 months until something affects a child's health, such as a mild viral illness, which may trigger the onset of symptoms. Without prompt treatment, this can lead to brain damage, seizures, low muscle tone, cerebral-palsy like symptoms and death within the first decade of life. Some affected babies also are born with an enlarged head (macrocephaly). Treatment can vary, but may include dietary protein restriction and supplementation with a nutrient called L-carnitine. With early diagnosis and prompt treatment of illness and fever, brain damage may be prevented.

## Hb S/Beta-Thalassemia = HBSA

*Incidence: greater than 1 in 50,000(1)*

In this form of sickle cell anemia, the child inherits one sickle cell gene and one gene for beta thalassemia, another inherited anemia. Symptoms are often milder than for Hb SS, though severity varies among affected children. Routine treatment with penicillin may not be recommended for all affected children.(3)

## Hb S/C disease = HbSC

*Incidence: greater than 1 in 25,000(1)*

Another form of sickle cell disease, in which the child inherits one sickle cell gene and one gene for another abnormal type of hemoglobin called HbC. As with Hb S/Th, this form is often milder the Hb SS and routine penicillin treatment may not be recommended.(3)

## Hearing loss = HEAR

*Incidence: greater than 1 in 5,000(1); up to 3-4 per 1,000 newborns(4)*

Without early testing, most babies with hearing loss are not diagnosed until 2 or 3 years of age. By this time, they often have delayed speech and language development. Early diagnosis allows use of hearing aids by 6 months of age, helping prevent serious speech and language problems.

## Homocystinuria = HCY

*Incidence: less than 1 in 100,000(1)*

Individuals with this disorder lack an enzyme responsible for converting the amino acid homocysteine into cystathionine, which is needed for normal brain development. If undetected and untreated, homocystinuria leads to mental retardation, eye problems, skeletal abnormalities, and stroke. Treatment consists of a special diet, one or more vitamins (B6 or B12), and other supplements (betaine).

## Hydroxymethylglutaric aciduria or HMG-CoA lyase deficiency or 3-OH 3-CH3 glutaric aciduria = HMG

*Incidence: less than 1 in 100,000(1)*

An inability to process the amino acid leucine leads to low blood sugar and accumulation of several organic acids, especially following illness or fasting. Without treatment, the disorder can lead to brain damage, mental retardation, coma and death. Avoiding fasting and following a diet low in protein and fat and high in carbohydrates can lead to normal development.

## Isovaleric acidemia = IVA

*Incidence: less than 1 in 100,000(1)*

This disorder is caused by an inability to process the amino acid leucine. The newborn form of the disorder often results in coma, permanent neurological damage, and death. In other cases, symptoms develop later in infancy and childhood, frequently following an infectious illness. With early diagnosis and treatment, most children have normal development. Treatment includes a low-protein diet and nutritional supplements.

## Long-chain 3-OH acyl-CoA dehydrogenase deficiency = LCHAD

*Incidence: greater than 1 in 75,000(1)*

Symptoms can begin soon after birth, resulting in heart, lung or liver failure and death. In other cases, symptoms such as low muscle tone, developmental delay, heart, lung or liver failure may develop later in infancy or childhood, most likely following an illness. Early diagnosis and treatment effectively prevent life-threatening events, though some children may still develop symptoms. Treatment includes a high-carbohydrate/low-fat diet, nutritional supplements, and avoidance of fasting. Women who are pregnant with fetuses with LCHAD are at increased risk of developing acute fatty liver of pregnancy and other pregnancy complications.

## Maple syrup urine disease = MSUD

*Incidence: less than 1 in 100,000(1)*

This inborn error of metabolism can be lethal if unrecognized and untreated. There is a wide spectrum of clinical presentations, from mild to severe. Affected babies appear normal at birth but soon begin to have neurological symptoms. The disorder gets its name from the fact that the urine smells like maple syrup. Without dietary treatment, severely affected babies do not survive the first month; even those who do receive treatment may have irreversible mental retardation. Rapid diagnosis and treatment are major factors in survival and outcome. Treatment consists of a special low-protein diet, which will vary depending on severity of symptoms, and sometimes, supplementation with a vitamin, thiamin. The diet must be continued indefinitely with frequent monitoring.

## Medium-chain acyl-CoA dehydrogenase deficiency = MCAD

*Incidence: greater than 1 in 25,000(1)*

Seemingly well infants and children can suddenly develop seizures (caused by low blood sugar), liver failure, coma, and death. Identifying affected children before they become ill is vital to preventing a crisis and averting these consequences. Treatment includes avoidance of fasting and nutritional supplements.

## Methylmalonic acidemia cblA and cblB forms = CBLAB

*Incidence: less than 1 in 100,000(1)*

This inherited defect of vitamin metabolism can lead to build-up of acids in the blood and result in brain damage, seizures, paralysis, coma and death. Symptoms can begin as early as the first week of life, though a minority of affected individuals remain symptom-free. Treatment with vitamin B12 injections and a low-protein diet often prevents serious problems.

## Methylmalonic acidemia due to mutase deficiency = MUT

*Incidence: greater than 1 in 75,000(1)*

A defect in the processing of four essential amino acids and other substances results in illness in the first week of life. Though severity of symptoms varies greatly, death during the first month of life and brain damage in survivors is common. Treatment includes a low-protein diet, vitamin B12 injections, and nutritional supplements. Some children die during the first year of life or develop brain damage despite nutritional intervention.

## Multiple carboxylase deficiency = MCD

*Incidence: less than 1 in 100,000(1)*

This disorder is caused by a defect of an enzyme required to activate several biotin-dependent enzymes. Without these enzymes, lactic acid and other organic acids build up in the body. Without treatment, brain damage, coma and death can result. Symptoms usually begin between birth and 15 months of age, and may include skin rashes and hair loss. Early diagnosis and treatment with biotin allows normal growth and development.

## Phenylketonuria = PKU

*Incidence: greater than 1 in 25,000(1)*

Affected individuals have an inability to properly process the essential amino acid phenylalanine, which then accumulates and damages the brain. PKU can result in severe mental retardation unless detected soon after birth and treated with a special formula. Affected individuals must be kept on a low-phenylalanine diet at least throughout childhood, adolescence, and for females during pregnancy.

## Propionic acidemia = PROP

*Incidence: greater than 1 in 75,000(1)*

This defect in the processing of four essential amino acids leads to illness during the newborn period. Without treatment, brain damage, coma and death can result. Even with treatment, including a low-protein diet and nutritional supplements, some affected children suffer from developmental delays, seizures, abnormal muscle tone, frequent infections and heart problems.

## Sickle cell anemia = HBSS

*Incidence: greater than 1 in 5,000(1); higher incidence among African-Americans (1 in 400)(2)*

A blood disease that can cause severe pain, damage to the vital organs, stroke, and sometimes death in childhood. Young children with sickle cell anemia are especially prone to dangerous bacterial infections such as pneumonia and meningitis. Vigilant medical care and treatment with penicillin, beginning in infancy, can dramatically reduce the risk of these adverse effects and the deaths that can result from them. Affected babies should receive all regular childhood vaccinations (including hemophilus influenza B and pneumococcal vaccines) to help prevent serious bacterial infections. Additional treatments may vary according to severity of symptoms, but may include intermittent pain medications and regular blood transfusions.

## Trifunctional protein deficiency = TFP

*Incidence: less than 1 in 100,000(1)*

A seemingly healthy infant can die suddenly of what appears to be sudden infant death syndrome. Other infants may develop low muscle tone, seizures, heart failure and coma, often following an illness. Treatment is based on strict avoidance of fasting, a low-fat diet and nutritional supplements.

## Tyrosinemia type I = TYR1

*Incidence: less than 1 in 100,000(1)*

Due to absence of an enzyme, byproducts of the amino acid tyrosine, particularly a very toxic compound called succinylacetone, build up in the liver. Without treatment, symptoms generally begin in the first few weeks or months of life and progress to liver or kidney failure, nerve damage and death. Drug treatment, sometimes along with a low-protein diet, is very effective in preventing liver and kidney damage.

## Very long-chain acyl-CoA dehydrogenase deficiency = VLCAD

*Incidence: greater than 1 in 75,000(1)*

Symptoms can first appear at any age from the newborn period through adulthood, but tend to be most severe in infants. Without treatment, affected infants often develop heart and liver failure and die during the first year of life. Treatment includes a high-carbohydrate/low-fat diet, nutritional supplements, avoidance of fasting and prolonged exercise.

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