

# Disorders Detected by Newborn Blood Spot Screening

The Washington State Newborn Screening Program tests all infants born in the state for a set of rare but serious health disorders that can be treated if caught early in life. Washington State screens for 35 disorders.

## Galactosemia

1 in 11,000 births

Babies with galactosemia cannot digest galactose, a sugar present in milk. When babies drink milk (including breast milk), galactose builds up in the body and can cause blindness, mental disability, or death. A lifelong diet without milk products can prevent these complications.

## Congenital Hypothyroidism

1 in 950 births

Babies with congenital hypothyroidism do not produce enough thyroid hormone to grow and develop normally. Early treatment with thyroid medication can prevent developmental disability and ensure normal growth and development.

## Cystic Fibrosis

1 in 5,500 births

Babies with cystic fibrosis develop poor lung function and struggle with malnutrition. This leads to serious health problems and a shortened lifespan. Early treatment can improve growth and development, and decrease the risk of infections and other complications.

## Biotinidase Deficiency

1 in 86,000 births

Babies with biotinidase deficiency cannot efficiently use a vitamin called biotin. If untreated, this can cause rashes, hearing loss, seizures and developmental delay. Lifelong treatment with biotin supplements can prevent these problems.

## Congenital Adrenal Hyperplasia

1 in 14,000 births

Babies with congenital adrenal hyperplasia have adrenal glands that cannot make enough of the hormones needed for healthy body function. These infants can have life-threatening episodes of dehydration and coma. Baby girls may have abnormal genitalia. Early treatment to replace the needed hormones can restore healthy body function.

## Severe Combined Immunodeficiency

1 in 88,000 births

Babies with severe combined immunodeficiency are born without a working immune system. They cannot fight germs that cause disease and even the most common infections can be life-threatening. A bone-marrow transplant early in life cures the baby by giving them a working immune system to prevent and fight infections.

## Sickle Cell & Hemoglobinopathies

1 in 4,700 births

Babies with sickle cell disease or other hemoglobinopathies have abnormal red blood cells that are unable to carry oxygen efficiently throughout the body. These disorders can cause frequent infection, severe pain, anemia and other complications. Early treatment and proper lifelong management can prevent serious health problems. Note: Some babies have a hemoglobin trait; this is not a disease and will not affect their health.

## Fatty Acid Oxidation Disorders (5)

1 in 11,000 births

Babies with fatty acid oxidation disorders cannot use fats in the body for energy. If these babies do not eat often, severe damage to the heart, liver and other organs can occur. If untreated, this will result in serious health problems and sometimes death. A special lifelong diet, eating frequently, and medications can help prevent these problems.

## Organic Acid Disorders (9)

1 in 29,000 births

Babies with organic acid disorders cannot digest certain parts of proteins found in food. If untreated, harmful substances build up in their blood and urine, which can have serious effects on their health, growth, and learning and can result in death. This can be prevented by early treatment with a special lifelong diet, close monitoring, and medications.

## Amino Acid Disorders (9)

1 in 10,000 births

Babies with amino acid disorders cannot process foods containing protein. If untreated, amino acids (the building blocks of protein) and other toxic substances build up in the body and have serious effects on health, growth and learning and can result in death. A special lifelong diet and supplements can help prevent these problems. An example of an amino acid disorder is phenylketonuria (PKU).



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## X-Linked Adrenoleukodystrophy

PREVALENCE 1 in 17,000 births

Babies with X-linked adrenoleukodystrophy cannot break down very long chain fatty acids. The build-up of these fats may cause fatty covering of the nerves, brain, and spinal cord (myelin) to break down. Boys can have this conditions but girls can be carriers. Possible treatments can include steroids, stem cell transplant, gene therapy, and supportive therapies and management like physical therapy.

## Lysosomal Storage Disorders (2)

PREVALENCE 1 in 31,850 births

Babies with lysosomal storage disorders have lysosomes that are unable to break down sugars and fats causing a toxic build-up. These infants have health problems of the brain, bone, muscle, or heart. Possible treatments can include enzyme replacement therapy, stem cell transplant, and supportive therapies and management like physical therapy, .

## Spinal Muscular Atrophy

PREVALENCE 1 in 15,000 births

Babies with spinal muscular atrophy lose motor nerve cells in their spinal cord. When these cells cannot send messages to the muscles, the no longer work. SMA can be mild or severe. Without treatment, SMA can cause missed motor milestones, difficulty breathing or eating, or even death.

## History of Newborn Screening in Washington State

