

Newborn Screen

The newborn screening program is a state sponsored preventive public health program designed to detect the newborns who are born with certain metabolic (digestive) disorders. Each baby in Texas is required to have 2 newborn screens, the first screen in the first 72 hours of life and the second one at 1-2 weeks of age. The cost of each screening is \$20 and results are available within 7 days.

The screen is done by collecting blood from the heel of the baby and sending it in to the Texas Department of Health laboratory. The newborn screen tests for the following disorders.

Congenital Hypothyroidism

The thyroid is necessary for normal growth, normal brain growth and development. If left untreated this disorder can result in mental retardation and stunted growth.

Symptoms of Congenital Hypothyroidism

Infants with untreated congenital hypothyroidism may appear clinically normal up to three months of age, by which time some brain damage will likely have occurred. When symptoms or clinical signs are present, they may include prolonged neonatal jaundice, constipation, lethargy, poor muscle tone, feeding problems, a large tongue, mottled and dry skin, distended abdomen and umbilical hernia.

Congenital Adrenal Hyperplasia

In this disorder, the adrenal glands do not produce enough cortisol. If left untreated, the baby could not maintain energy and blood sugar levels necessary for normal life which could lead to lethargy and coma and eventually death.

Symptoms of Congenital Adrenal Hyperplasia

Male infants with CAH usually appear normal at birth but may develop symptoms within the first 2 weeks of life. Female infants may appear normal or may show the effects of virilizing hormones: an enlarged clitoris and fusion of the labia majora over the vaginal opening and may also exhibit symptoms within the first two weeks. Occasionally the female infant will appear to have a normal male penile structure with hypospadias. These females never have a palpable gonad in the labial/scrotal sac.

Phenylketonuria (PKU)

Defective metabolism of phenylalanine results in toxic accumulation of this amino acid and deficiencies in its metabolites.

Symptoms of PKU

Infants with untreated PKU may appear normal in the first few months of life. While in utero, phenylalanine is maintained in the normal range by the mother's system. About 24 hours after the first protein feeding the level of phenylalanine begins to rise to toxic levels. Vomiting (occasionally projectile) may be one of the first signs. By one year of age, mental and motor retardation, microcephaly (small head), poor growth rate and seizures or tremors will be evident. Inadequate production of tyrosine (a precursor to pigment formation) results in lighter hair and skin than other family members. The skin may be oily and eczematous. If treatment is not initiated early, most individuals with PKU will achieve an IQ of less than 50.

Galactosemia

The major sugar in milk is lactose. This sugar is digested to galactose and glucose in the intestine. Galactosemia results from a deficiency in one of the enzymes necessary for the metabolism of galactose.

Symptoms of Galactosemia

The affected infant may appear normal at birth. Within a few days to 2 weeks after initiating milk feedings, the infant develops vomiting, diarrhea, lethargy, jaundice and liver damage. Untreated, this disorder may result in death, frequently associated with E. coli septicemia. Infants surviving the above symptoms evidence developmental retardation, hepatomegaly, Fanconi's syndrome, growth failure and cataracts.

Hemoglobinopathies

Hemoglobinopathies (sickle cell anemia and thalassemia) are recessively inherited abnormalities in the structure of hemoglobin. Sickle cell diseases affect about one in 2200 Texas infants and about 1:400 infants of African American descent. The malformed red blood cells of this disease are easily destroyed and tend to clump in and occlude small blood vessels resulting in pulmonary infarction (heart attack), stroke, spleen problems and painful damage to internal organs.

Symptoms of Hemoglobinopathies

The affected infant appears normal at birth. Anemia develops in the first few months of life, it is usually mild and requires no treatment. Enlargement of the spleen can result from the trapped cells and severe anemia develops rapidly and transfusions are necessary. Death can result. Infants and children with sickle cell anemia are particularly susceptible to infection. Growth is also affected and gallstones are common.

Newborn Screen Test

I/ we _____ the parent/ managing conservator/ guardian of _____, born on this date, _____

have had explained to me the benefits of the Newborn Screening Blood Test and the legal requirements for this test for all newborns in the State of Texas.

_____ I/ We have decided to have both tests done on the above named child.

_____ I/ We have decided to have only one test done on or after two weeks of age.

_____ I/ We have decided to decline all Newborn Screen testing on our child at this time.

Signature: _____
(Parent/ Managing Conservator/Guardian)

Date: _____

Signature: _____
(Witness)

Date: _____

A signed copy must be included in child's medical records if Parent/ Managing Conservator/Guardian refuses to permit the blood test.