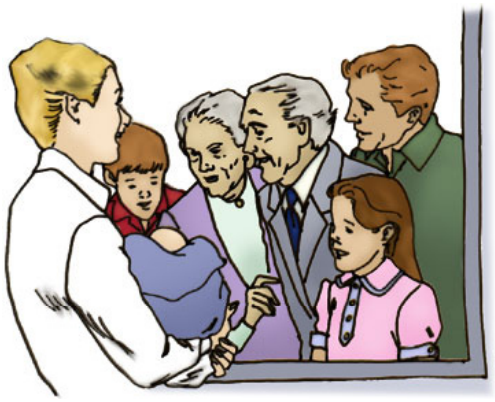
X-Plain™
Newborn Screening
Reference Summary

Introduction

Newborn screening is a program that usually begins with a blood test or other test and is used to identify serious or life-threatening conditions before symptoms begin. Treatment can be started before a baby's health is harmed by these conditions.



Even though each disease by itself is rare, all together these diseases affect 1 in 1500 babies. If untreated they can lead to health problems, such as poor growth or mental retardation, or even death.

Newborn screening commonly may include either a blood test or a variety of other tests. Multiple tests are run on the blood and if those results are not normal, the lab notifies the baby's doctor. Once diagnosed, treatment can save the baby from death or lifelong health problems.

This reference summary explains newborn screening. It discusses its reasons, the procedure and interpreting results.

Why Screen

Newborn Screening is important because a baby with one of these illnesses may seem healthy but by the time symptoms appear, permanent damage may have occurred. Damage can lead to mental retardation, or, in many cases, disability or death.

With the advances in medical technology, it is now possible to find extremely small amounts of body chemicals in the blood. This shows whether a newborn baby is likely to have one of these diseases. Additional testing, such as genetic testing, is commonly required if the newborn has some of these diseases.

Phenylketonuria, or PKU, was the first disease to be screened for in newborns. People with PKU do not have the enzyme that processes phenylalanine (Phe). Because of this, phenylalanine builds up in the blood and brain, which leads to mental retardation.

Treatment for PKUinvolves a special diet with very small amounts of Phe. This allows the child to grow normally without mental or physical retardation.

Some of these newborn diseases are very dangerous. One example is a rare disease called Medium-chain acyl-Coenzyme Dehydrogenase deficiency (MCAD). Twenty five percent of children with unrecognized MCAD die the first time they have an illness. The treatment for MCAD can be as simple as avoiding fasting and giving the child frequent meals.

Screening newborns for such dangerous and rare diseases gives them the best chance for a healthy life. Testing varies from state to state. Most states test for more than 30 disorders at present, but some states screen for less than 10. Ask your healthcare provider about how many disorders your state screens for.

Screened Disorders

There are several types of diseases that can be found through newborn screening, such as metabolic disorders, hormonal disorders, blood disorders and hearing disorders. Most of the required newborn screening tests are for metabolic disorders.

Metabolic disorders have to do with metabolism, which is the way the body uses nutrients to keep tissues healthy and produce energy. Hormonal disorders have to do with hormones, which are chemicals in the blood that regulate important bodily functions.

Screening a newborn baby's hearing is required in many states. If your baby's hearing is not checked at birth, make sure he or she gets tested. Measuring how a baby responds to sound and identifying hearing loss early makes a big difference, since early treatment improves the long term outcome. Unrecognized hearing loss can cause learning disabilities.

The following are examples of metabolic disorders.

- PKU[1]
- Galactosemia[2]
- Biotinidase deficiency[3]
- MSUD[4]
- Homocystinuria[5]

The following are examples of hormonal disorders.

- Congenital hypothyroidism[6]
- Congenital adrenal hyperplasia[7]
- Sickle cell anemia is an example of a blood disorder[8]

Procedure

Blood for newborn screening is taken by pricking your baby's heel. The paper is allowed to dry and then sent to a laboratory, where several different tests will be performed.

The sample is usually taken when the baby is between 24 to 48 hours old. This is because some diseases, such as PKU, do not always show up if the blood sample is taken too soon after birth. If the mother and newborn are discharged before the baby is 24 hours old, the test should still be done before they leave and repeated after 48 hours of age. Some of the conditions that are detectable can result in the baby's death within 5 days if left untreated.

Results

Some labs send the results to the doctor and others send them to the hospital where the baby was born. Ask your doctor how you will get the results and when you should expect them.

If a test result comes back "normal," it means there is no indication that the baby has one of the rare diseases. If a test result comes back "abnormal," it either means the baby may have one of the rare diseases tested for or that the test result is a "false positive." Either way, more testing is needed.

If further tests confirm that your child has a disease, your child's doctor may refer you to a specialist for treatment. If you have other children who were not screened for the disease, you may want to have them screened. They may be at risk for the disease and need treatment also.

Risks

Pricking a baby's heel for a blood sample rarely leads to complications.

A test that shows an abnormality is called a "positive" test. If the newborn screening result comes back positive but further tests are negative, it is called a "false positive." A risk involved with a false positive may be parental anxiety.

State Requirements

All states have mandatory screening. States vary in the number of conditions for which they screen, and the number of tests performed is increasing in all states.

A lot of states offer expanded screening with tandem mass spectrometry[9] on every baby. Some hospitals offer screening beyond what state law requires. Parents can ask for additional screening if they live in a state that screens for a limited number of conditions. Ask your healthcare provider if you want further information.

Additional Screening

The decision whether or not to get additional screening can be a difficult one. You should discuss this with your doctor. If your baby has a close relative who has an inherited disorder or who has died from one, you should consider additional screening.

If you have health insurance, keep in mind that it may not pay for additional screening. You may have to pay it out of your own pocket. This should NOT change your decision to get additional screening. If you will have trouble paying for additional screening, make sure to let your doctor know. She or he will refer you to a social service specialist who may be able to help you.

Conclusion

Newborn screening is a program that usually begins with a blood test or other test used to identify serious or life-threatening conditions before symptoms begin.

Such diseases are usually rare. However, they hinder an infant's normal development in a variety of ways, physically and mentally.

Newborn screening usually consists of a blood test that involves pricking the baby's heel to get few drops of blood. The lab results tell doctors and parents whether

newborns have certain conditions that could eventually cause problems. Once diagnosed, treatment can save the baby from lifelong health and developmental problems or even death.

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[1] Babies affected with PKU cannot process an amino acid, which results in mental retardation. This condition is treated with a special diet.

[2] Babies who have galactosemia lack the enzyme that breaks down milk sugar. Galactosemia can lead to liver failure, cataracts, severe mental retardation and even death. Following a special diet treats the condition.

[3] Babies who have biotinidase deficiency don't have enough of the enzyme biotinidase. This condition causes seizures, poor muscle control, immune system impairment, hearing loss, mental retardation, coma, and even death. Giving the baby biotin treats this condition.

[4] Babies affected with MSUD cannot process multiple amino acids. If not detected and treated early, MSUD can cause mental retardation, physical disability and even death. The condition can be controlled with a special diet.

[5] Babies with homocystinuria cannot break down homocystine. This condition can lead to dislocated lenses of the eyes, mental retardation, skeletal abnormalities and abnormal blood clotting. The disorder can be controlled through a special diet and medications.

[6] Babies that have congenital hypothyroidism don't have enough thyroid hormone, which causes physical and mental retardation. Giving the baby thyroid hormone treats this condition.

[7] Babies that have congenital adrenal hyperplasia lack certain hormones produced by the adrenal gland. This condition can affect the development of the genitals and may cause death due to loss of salt from the kidneys. It can be managed by giving the baby needed hormones.

[8] Babies with sickle cell anemia have red blood cells that form abnormal "sickle" shapes, which can cause episodes of pain, damage to vital organs, strokes, infections and even death. Complications of this condition can be managed with preventative antibiotics and close monitoring of blood counts.

[9] A new technology called tandem mass spectrometry or MS/MS screens for more than 20 disorders with a single test. Because of this technology, more states are beginning to offer expanded newborn screening tests.