PENNSYLVANIA Screening Services FOR NEWBORN BABIES

Protecting Babies

Preventing Problems

Screening Right After Birth

Starting Treatment Early

Newborn Screening...
A Special Time...A Special Test!

UPDATED

This brochure describes special tests given to your new baby. These tests are called newborn screening. If the result of one of your baby's tests is "positive," this could mean a possible danger to your baby's health. Because of this, further testing may be needed. This brochure gives information about where you can go for help.

A baby may appear to be healthy but may be born with a serious medical condition. If your baby seems to be having a problem after leaving the hospital, immediately contact your baby's primary care provider or go to the hospital.

Today, we know a lot about helping babies with serious medical or hearing problems.

Many serious medical conditions can be identified with simple tests soon after a baby is born.

We know that the best way to help is to start treatment soon after babies are born.

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CHECKING Babies for Special MEDICAL CONDITIONS

What conditions are babies tested for in Pennsylvania?

By Pennsylvania law, all babies are required to be tested for six medical conditions, but most health care providers already test for many more conditions. The law now requires the results of the conditions listed in this brochure to be reported to the Department of Health. These conditions are only found in a few babies. By testing at birth, babies with these conditions can be found quickly to begin treatment. Early treatment can prevent very serious medical problems or death. When necessary, your primary care provider will work with a specialist to provide appropriate treatment.

The conditions required by law include:

1. Congenital Adrenal Hyperplasia (CAH)
(Kon-JEN-i-tal Ah-DRE-nal Hi-per-PLA-zhah)
Babies born with this condition have a defect in an important substance (enzymes) the body needs. CAH can cause dehydration, shock and even death within a few days of birth. Medical problems can be prevented when treatment is started soon after birth.

2. Congenital Hypothyroidism (CH)

(Kon-JEN-i-tal Hi-po-THI-roid-ism) Babies born with this problem do not have a thyroid hormone. They may look healthy. If not detected, the condition can cause poor growth and mental retardation. Giving the baby special medicine every day can prevent these results.

3. Galactosemia

(Gah-LAK-toe-SEE-mee-ah) Babies with this disorder cannot digest galactose. Galactose is a simple sugar found in breast milk, many formulas and milk products. This condition can harm the baby's eyes. It can cause serious liver and brain damage. Giving the baby a special milk-free diet as soon as the condition is found can prevent problems.

4. Maple Syrup Urine Disease (MSUD)

Babies born with MSUD cannot digest part of a food protein. Without treatment MSUD can cause severe mental retardation or even death shortly after birth. To prevent these results, babies are given a special formula and diet.

5. Phenylketonuria (PKU)

(FEN-nil-KEE-tone-u-ree-ah) Babies who are born with PKU cannot digest a different part of a food protein. Untreated, PKU causes nerve and brain cell damage, which can result in mental retardation. This damage can be prevented when a baby gets a special formula and diet.

6. Sickle Cell Disease and other Hemoglobin Diseases

Sickle cell disease and other hemoglobin diseases are a group of genetic conditions that cause abnormalities with the blood. This leads to problems with blood circulation and anemia. Infants and children with sickle cell disease can die from lung and brain infections. Early treatment to prevent infections greatly reduces the chance of sickness or death. Sickle cell trait may also be identified through the screening.

Most health care providers test for the following additional conditions. Ask your health care provider to test for these additional conditions:

7. Organic Acid Disorders (OAs) *
Babies with these inherited conditions are unable to break down the proteins they ingest. If untreated, OAs may cause breathing problems, seizures, brain swelling, stroke and coma, sometimes leading to death. To prevent these problems, babies are given a special formula, diet and sometimes medication.

8. Fatty Acid Oxidation Disorders (FAODs) *
Babies with these inherited conditions are unable to break down fat. If untreated, FAODs disorders may lead to serious complications affecting the liver, heart, eyes and general muscle development, and possibly death. To prevent these problems, treatment may vary and could include avoiding long periods without food, medications and diet.

9. Amino Acid Disorders (AAs) *
Babies with these inherited conditions are unable to process certain amino acids. If untreated, AAs can result in muscle weakness, breathing problems, seizures, swelling of the brain, coma and sometimes death. To prevent these problems, babies are given a special formula, diet and sometimes medication.

10. Cystic Fibrosis (CF) *
Babies with this inherited condition have thick, sticky mucus and fluid build up in certain organs, especially the lungs and pancreas. Some symptoms include repeated lung infections, poor weight gain and growth. If untreated, CF may cause serious health problems that could lead to early death. Many symptoms of CF can be controlled with medication and treatments.

11.Biotinidase Deficiency **
(Bio-tin-a daze de-fish-en-see)
Babies with this inherited condition have a lack of an enzyme called biotinidase. Without treatment, this disorder may lead to seizures, developmental delays, eczema and hearing loss. With early diagnosis and treatment, all symptoms can be prevented.

**** Reference taken from Screening, Technology, and Research in Genetics- www.newbornscreening.info/parents/facts.html***
***** Reference taken from Save Babies Through Screening Foundation- www.savebabies.org***

How is my baby tested?

The hospital or birth facility gets a drop of blood by pricking the baby's heel. The blood is placed on a special filter paper and mailed to a laboratory.

By Pennsylvania law, your baby's blood filter paper test cannot be used for scientific research by any laboratory without your signed permission.

When is the testing done?

Usually the drop of blood is taken when the baby is 24 to 48 hours of age. This often happens just before the baby is discharged. The newborn screening test must be repeated if the baby is tested before he or she is 24 hours of age.

My baby seems very healthy. Is the test still needed?

Yes. Most infants with these conditions show no signs of illness immediately after birth.

If my baby has one of these conditions, can it be cured?

No. Your baby cannot be cured. When treatment is started very early in life, the medical problems related to the condition are often prevented.

Will I receive a report of the test results?

Yes. Test results are known within seven to 10 days after the blood was taken. Test results are sent to the birth facility or the health care provider who took the test and are placed in the baby's medical record. Ask about the test results during your baby's regular checkup. Your baby's primary care provider can call the Pennsylvania Department of Health Newborn Screening and Follow-Up Program at 717-783-8143 or 1-877-724-3258 to get the results.

If the test results show that there may be a problem, your baby's primary care provider will contact you. For this reason, ***it is very important that you give your birth facility the best address and phone number to find you as well as the address and phone number of the primary care provider who will be caring for your baby after discharge from the hospital.*** *If possible, also provide an emergency contact number. This will prevent delays in contacting you if further testing is needed.*

If my baby needs another test, does this mean that my baby has one of these conditions?

No, not necessarily. Another test may be needed because:

- The first blood sample was not large enough to complete the tests.
- Your baby left the birthing facility before 24 hours of age.
- The first test showed a possible problem. A new blood sample or further testing is needed. Generally, if the result of any additional testing is not normal, your baby's primary care provider will discuss with you what should be done next.

IF YOU ARE ASKED TO HAVE YOUR BABY RETESTED, PLEASE HAVE IT DONE RIGHT AWAY! BRING YOUR BABY BACK TO THE BIRTH FACILITY OR YOUR BABY'S PRIMARY CARE PROVIDER AS INSTRUCTED.

How does the Department of Health's Newborn Screening and Follow-Up Program help you?

The Newborn Screening and Follow-Up Program's role is to help notify your baby's primary care provider if your baby has a "positive" result for any of the conditions listed in this brochure so appropriate follow-up services can take place.

May I say NO to the blood tests?

Yes, you may ask not to have the test done for religious reasons. If you say "no," this will be recorded in your baby's record with your signature. However, if your child is born with one of these conditions, your child will not have the benefits of early treatment.

CHECKING Babies for Hearing Loss

Hearing Screening

Babies start learning how to use sound as soon as they are born. Listening in the first months of life prepares a baby to speak.

Babies learn to talk by listening to their families talk around them. Imagine that a baby has a hearing loss but no one knows about it. This can lead to slow development of speech and language.

Your newborn baby cannot tell you if he or she is unable to hear. This is why your baby should be screened immediately after birth to check for hearing problems.

Why screen my baby?

Hearing loss is found in three out of every 1,000 newborn babies. More than half of babies born with hearing problems have no other health problems and no family history of hearing loss.

If a newborn's hearing loss is not found and treated, the child may:

- Be slow in learning to speak or use sign language
- Have problems learning at school
- Suffer from social and emotional problems

What is the screening test?

There are two ways to screen your baby's hearing - both are safe and are usually done when your baby is asleep.

The first test measures a tiny sound the ear makes when the baby is hearing properly. This test is called an Otoacoustic Emissions (OAE) test.

The second test measures the baby's nerve response to sound. This is called an Auditory Brainstem Response (ABR) test.

If your baby passes the hearing screening

If your baby passes the newborn hearing screening, you do not need follow-up testing at this time.

However, a small number of babies who pass the screening at birth can lose their ability to hear – suddenly or gradually - before they are one year of age or older.

If you have a family history of permanent childhood hearing loss, your baby should be tested every year.

Some babies may develop hearing loss later. Repeated ear infections, meningitis, head injury or other medical conditions are some of the causes of hearing loss in children. Children with a history of these conditions should receive hearing tests frequently.

If your baby does NOT pass the hearing screening

There may be several reasons why babies do not pass the hearing screening:

- The nursery is too noisy to get good screening results
- They are too active or do not sleep during the screening;
- They have a hearing loss - either temporary or permanent.

IF YOUR BABY DOES NOT PASS, YOU SHOULD COME BACK FOR ANOTHER HEARING SCREENING.

Where CAN I GET HELP?

If you have any questions, ask your baby's primary care provider. If your baby is diagnosed as deaf or hard of hearing, Early Intervention services, located in every county in Pennsylvania, can:

- Help find answers to your questions about hearing loss
- Teach you how to communicate and care for your baby
- Help your baby acquire hearing aids and other listening devices

Early Intervention is a support service made possible by government funds.

Call the CONNECT Line for Early Intervention services:

(1-800-692-7288)

Other sources of information:

- Your baby's primary care provider
- Pennsylvania Department of Health's Web site: www.health.state.pa.us
- E-mail the Hearing Screening Program at nbhs@state.pa.us

An excellent source of information for all pregnant women and parents of newborns is the Healthy Baby help line. One of the programs you can contact through this line is the Love'em with a Checkup Program if you don't have a primary care provider or can not afford to pay for appointments. Call this program to connect you with a primary care provider in your area or to discuss health care coverage options.

1-800-986-BABY
voice & TTY
(1-800-986-2229)

Phone numbers to call for more information

1-877-724-3258 for Pennsylvania Department of Health Newborn Screening and Follow-up Program.

1-800-986-BABY (2229) for information on finding a primary care provider, getting health care coverage, immunizations and tests for baby.

1-800-986-KIDS (5437) for information on finding a primary care provider, getting health care coverage, immunizations or tests for your children.

1-800-WIC-WINS (942-9467) to obtain supplemental foods, nutrition education and breastfeeding information.

1-800-986-4550 for information on services available for children with special needs.

1-800-4-A-CHILD (22-4453) 24-hour crisis hotline to offer support, information and referrals on coping with a crying baby and preventing child abuse.

Additional Resources and Information

www.health.state.pa.us/newbornscreening for information on newborn screening and other Department of Health programs.

www.ins.state.pa.us for information on Pennsylvania Insurance Department's Children's Health Insurance Program (CHIP) and adultBasic health care coverage programs.

www.dpw.state.pa.us for information on the Pennsylvania Department of Public Welfare's Medicaid health care coverage program.

www.compass.state.pa.us to apply for state social service programs online.

www.4woman.gov for information on breastfeeding.

www.acog.org for information on the American College of Obstetrics and Gynecologists.

www.aap.org for information on the American Academy of Pediatrics.

www.savebabies.org for information on screening tests and support groups.

www.marchofdimes.com for information and videos on newborn screening.

www.newbornscreening.info for genetic and metabolic condition fact sheets for Parents.

www.ghr.nlm.nih.gov for general information on genetic and metabolic conditions.

www.familydoctor.org for information on the American Academy of Family Physicians.

www.medlineplus.gov for health information from the National Institutes of Medicine and Health.

www.healthfinder.gov for health information from Health and Human Services, Office of Disease Prevention/Health Promotion.

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www.health.state.pa.us/newbornscreening

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