



Screening Test for Your Baby



What is Maryland's Newborn Screening Program?

Maryland's Newborn Screening Program has two parts: Newborn Blood Spot (Metabolic) Screening and Newborn Hearing Screening. This booklet will tell you about these programs. We hope that you will talk about newborn screening with your spouse, another family member, a friend, doctor, or nurse

Metabolic Newborn Screening

The Maryland Department of Health and Mental Hygiene offers metabolic newborn screening as a service to families with new babies. This program finds newborn babies with certain rare, serious disorders of body chemistry. These disorders can be treated if detected early. Before your baby can be tested, you, as the parent, must agree.

Why is my baby tested?

The testing is done to make sure that your baby will be as healthy as possible. The Newborn Screening Program finds babies who **may** have one of several rare disorders and alerts the doctors to this possibility. Serious illness can usually be prevented with early diagnosis and medical treatment.

How is my baby tested?

Once you give permission, your baby's heel will be pricked to get a few drops of blood. This blood will then be placed on special paper, dried, and mailed to the State Laboratory.

The laboratory needs just a tiny bit of blood to do all the tests described in this booklet. The heel prick feels no worse than being stuck by a pin. Problems from the test, such as infection of the heel, are very rare.

What disorders can metabolic newborn screening identify?

Maryland recently expanded the number of disorders in the program. The Maryland program now tests babies for 35 conditions. They are:

- **Biotinidase Deficiency** (BI-oh-TIN-a-dase) - The body cannot recycle the B vitamin called Biotin (BI-oh-TIN). The body does not have the enzyme (a chemical made by the body) needed to do this. Babies with this deficiency need more biotin

than other babies. Treatment with extra biotin prevents the mental retardation and deafness that would otherwise result.

- **Branched Chain Ketoaciduria** (KEE-toe-acid-u-ree-ah) (**BCK**) - The body cannot use some parts (the branched chain amino acids) of the protein in food. The body does not have the enzyme needed to use these amino acids. This defect is also called Maple Syrup Urine Disease (MSUD) because the urine smells like maple syrup! Treatment with a special diet can prevent life-threatening complications. MSUD belongs to the group of disorders called organic acidurias. Maryland has screened for BCK since 1973 and screened for biotinidase deficiency since 1984. The other organic acidurias were added to the program in 2003.
- **Congenital Adrenal Hyperplasia** (con-GEN-i-tal ad-RE-nal hi-PER-play-see-ah) (**CAH**) – The body lacks an enzyme needed by the adrenal glands to make the hormones, cortisol and aldosterone. Cortisol is needed for making energy and aldosterone is needed for proper salt and water balance. Without aldosterone, the baby will lose salt and water in the urine and get dehydrated. This can happen very fast. The adrenal glands also make androgens, the male sex hormones. When the adrenal gland cannot make cortisol and aldosterone, it makes more androgens. Too much male hormone makes the genitals of baby girls look unusual and causes early sexual development and short stature in childhood in both sexes. CAH is treated by giving the missing hormones.
- **Cystic fibrosis (CF)**- A problem with moving salt in and out of cells causes thick, sticky mucus in the lungs. This leads to lung infections and problems with breathing. Some babies with CF also have problems digesting (breaking down) the food they eat, causing slow growth. CF is diagnosed with a test that measures the amount of salt in the sweat. CF is treated with breathing treatments and antibiotics for the lungs, and with medicine and special diet for babies with digestion problems.
- **Fatty Acid Oxidation Disorders** – The body usually gets energy from sugars and fats. The sugar is used first but when the sugar is all used up, the body must use fats. In this group of disorders, the body cannot use fats because of the lack of one of several enzymes. The disorders in this group do not have common names. They are usually described by the length of the fatty acid that cannot be used. The most common of these disorders is Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency. Because they cannot use stored fat for energy, babies with these disorders may develop seizures, coma and life-threatening complications. Treatment includes making sure the baby eats regularly and avoiding fasting. A special diet and medications may also be used. Screening for these disorders is very new. The 11 fatty acid oxidation disorders included in the Maryland Newborn Screening Program are:
 - Carnitine Uptake Disorder
 - Carnitine /Acylcarnitine Translocase Deficiency
 - Carnitine Palmitoyl Transferase I Deficiency (CPT I Deficiency)
 - Carnitine Palmitoyl Transferase II Deficiency (CPT II Deficiency)
 - Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
 - Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
 - Multiple Acyl-CoA Dehydrogenase Deficiency (MADD) or Glutaric Acidemia Type II
 - Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)

- Short Chain 3- Hydroxyacyl-CoA Dehydrogenase Deficiency (SCHAD)
- Trifunctional Protein Deficiency (TFP Deficiency)
- Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)

- **Galactosemia** (ga-LAK-toe-see-me-as) – The body cannot use a sugar in milk (galactose) because of the lack of an enzyme. A diet low in galactose can prevent life-threatening complications. Babies with the most severe form of this disorder can get very sick very fast.

- **Homocystinuria** (HO-mo-SIS-tin-u-ree-ah) – The body lacks an enzyme in the liver that is needed to handle sulfur. This can lead to mental retardation and other changes in the body. A special diet, and sometimes vitamin B6, can prevent mental retardation and other complications.

- **Hypothyroidism** (HI-po-THI-royd-ism) - The body is cannot make enough thyroid hormone (thyroxin). This leads to mental retardation and slow growth. These problems can be prevented by giving thyroxin pills.

- **Organic Acidurias** - The body cannot use the branched chain amino acids from the protein in food because of the lack of one of several enzymes. The breakdown products of these amino acids are organic acids. The organic acids build up to dangerous levels in the blood damaging the nervous system. Babies with some of these disorders can become very sick very fast. BCK/ MSUD is one of the disorders in this group. Symptoms can sometimes be reduced with special diets (low in protein) and medications. Most of these disorders do not have common names and are described by the name of the organic acid found in the urine. Screening for most of these disorders is very new. The 13 organic acidurias included in the Maryland Newborn Screening Program are:
 - Branched Chain Ketoaciduria (BCK)/ Maple Syrup Urine Disease (MSUD)
 - Biotinidase Deficiency
 - Glutaric Acidemia Type I (GA I)
 - 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG CoA Lyase Deficiency)
 - Isobutyryl-CoA Dehydrogenase Deficiency
 - Isovaleric Acidemia
 - 2-Methylbutyryl-CoA Dehydrogenase Deficiency
 - 3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC Deficiency)
 - 3-Methylglutaconyl-CoA Hydratase Deficiency
 - Methylmalonic Acidemia (MMA)
 - Mitochondrial Acetoacetyl-CoA Thiolase Deficiency (3-Ketothiolase Deficiency)
 - Multiple Carboxylase Deficiency
 - Propionic Acidemia (PA or PPA)

- **Phenylketonuria** (Fen-nil-KEE-tone-u-ree-ah) (**PKU**) - The body cannot use a part of the protein in food (the amino acid phenylalanine) because of the lack of an enzyme. A special diet low in phenylalanine can prevent the brain damage that would otherwise result.

- **Sickle Cell Anemia** - This is a serious blood disorder. Under certain conditions, the abnormal blood cells can clog up the small blood vessels causing painful “crises.” Babies with sickle cell anemia also have less resistance to infections, which may be

life threatening. A regular treatment program using penicillin and special immunizations helps avoid serious complications.

- **Tyrosinemia** (TY-ro-SIN-e-me-ah) - Tyrosine, another component of the protein in food, cannot be used properly. Some forms of this problem can result in liver and brain damage, and may be life-threatening. Other forms can damage the eyes and skin. Babies with this disorder are treated with a special diet, medication and in some cases, a liver transplant.
- **Urea Cycle Disorders** - The body cannot dispose of nitrogen properly. Nitrogen is found in proteins. The body usually disposes of nitrogen by changing it into a substance called urea, which leaves the body in the urine. Because of the lack of one of several enzymes, the body cannot make urea effectively and the nitrogen builds up as ammonia in the blood. This makes the baby very sick very fast. These disorders cause seizures, poor muscle tone, respiratory distress and coma. Death will result if the baby is not treated. Symptoms can be reduced with special diet and medications that help the baby to excrete ammonia in other ways. Screening for this group of disorders is very new. The three urea cycle disorders included in the Maryland Newborn Screening Program are:
 - Argininosuccinic aciduria
 - Argininemia
 - Citrullinemia

Newborn screening is a rapidly growing service. The number of disorders included in the program has recently expanded from nine disorders to 35 disorders. The number of disorders included is not a good way to judge the quality of a newborn screening program. Different programs count in different ways. However you count, Maryland screens for all the disorders recommended by the March of Dimes and the American College of Medical Genetics.

Work is still going on to develop tests for still other serious disorders. Your baby may be tested for these as well. You will be told if any of these new tests suggests that your baby might have a problem.

From time to time, the State Laboratory does studies on other important health problems using these samples. These are only done after all the other tests are finished. These studies show how many babies are affected by the problem being studied. They do not tell which babies have the problem, because the samples are not identified by name.

Is my permission required?

Most parents want to know what is important to their baby's health so they can be sure that their baby gets good care. Testing newborn babies for these disorders is an important part of good baby care.

The State of Maryland requires that you be asked to give permission before metabolic newborn screening tests are done. The permission form is in the back of this booklet. You will need to sign this form after your baby is born. This will give you the chance to say "yes" or "no." Please say, "YES!"

My baby seems healthy. Are the tests still needed?

Yes, the tests are needed. Most infants with conditions found by the Newborn Screening Program have no obvious signs of illness right after birth. However, each of these disorders can cause serious problems, *if not found early and treated*. Many of these disorders can cause brain damage and mental retardation. Others, like sickle cell disease and CAH, can cause serious physical illness.

There are many causes of mental retardation. This booklet only deals with the disorders for which there are good screening tests.

Effective treatment is available for most of these disorders. A few disorders, recently added to the program, are not yet well understood and treatment may not be effective. But every baby with a disorder deserves the best treatment that is available.

These disorders do not always show symptoms in the same way. Some babies may become dangerously ill within the first days or few weeks of life. Others may appear healthy for months or even the first few years of life. In either case, serious harm is occurring that could be prevented, in most cases, with early treatment. Once the damage has occurred, it can never be repaired.

How much will these tests cost?

The State Laboratory charges the hospital a small fee (\$42.00) for doing the screening tests on your baby's blood sample. In addition, the doctor, clinic, or hospital may charge a small fee to do the heel prick. No baby will be refused testing because a family cannot pay.

How does a baby get one of these disorders?

Except for hypothyroidism, a baby inherits the disorder from both parents. Parents and relatives usually show no sign of the disorder. In most cases of hypothyroidism, the thyroid gland does not develop or is too small or in the wrong place in the body. These problems are usually not inherited. In a few cases, there is an inherited defect in the gland's ability to make thyroxin.

These disorders are quite rare, and the chances are excellent that your baby will not have one of them.

The few babies who are born with these problems are generally from healthy families. Testing EVERY baby after birth will make sure that each infant who has a disorder will be identified and started on early treatment.

When is the best time to test my baby?

Each baby should be tested twice. The first test is usually done shortly before the newborn baby leaves the hospital. For the most reliable results, it should be done after the baby has received at least 24 hours of breast or formula feedings.

In some cases, babies leave the hospital before they are 24 hours old or have had milk for 24 hours. They *should* be tested before they leave the hospital, but some of the test results will not be accurate. The test should be repeated when the baby is 2 to 3 days old.

If your baby was not born in a hospital, the first test should be done when your baby is 2 to 3 days old.

Most doctors will routinely obtain a second blood sample when the baby is a little older (about 2 weeks), even if the first test was ideal and normal. This second test gives another chance to find the babies with problems. It is especially important for finding babies with CF and hypothyroidism.

How soon will I know the results?

In most cases, the test results will be normal and you will not be personally notified. Your doctor or clinic will get the report. Results are usually available in about ten days. Generally, parents are notified only if there is a problem. You should ask about the results when you bring your baby to the doctor or clinic for a regular checkup.

Although “no news is good news,” it is important to remember that these tests only find the disorders listed in this booklet. There may be other medical problems that cannot be picked up by these tests. Therefore, it is very important for your baby to have regular checkups and good general medical care.

What if I am told that a “retest” is necessary?

A retest may be needed for a number of reasons. It does **not** mean there is anything wrong with the baby. It usually means that another sample is needed so all tests can be completed.

Rarely, the first test indicates a possible problem. In this case, a new blood sample is also requested, and the tests are repeated or the baby is sent to a specialist.

If you are asked to have your baby retested, please act quickly!

What if my baby has one of these disorders? Is there a cure? Because most of these disorders are inborn problems of the body chemistry, they cannot be “cured” (just as eye color or height cannot be permanently changed). However, the serious effects of the disorder can usually be controlled and sometimes completely prevented by early treatment. Effective treatment is available for most of these disorders. The treatment is usually a special diet or medication. A few disorders are not yet well understood and treatment may not be effective. However, every baby with a disorder deserves the best treatment that is available. The Health Department or your doctor will make arrangements for referral and treatment.

If this child has a disorder, will my future children also have it?

The answer to this question will depend on your child’s diagnosis. It can best be answered by a genetic counselor. With your help, the counselor can study your family’s health history and explain the chance of this happening again. This process is called genetic

counseling. If you would like more information about genetic counseling, your doctor or Health Department will help you get it.



Newborn Hearing Screening

The second part of Newborn Screening in Maryland is screening of your baby's hearing. This program is designed to help find babies who may have hearing loss and need early intervention.

Why is my baby tested?

There are two reasons that the State screens all babies for hearing loss. If a baby with a hearing loss is helped before six months of age, the baby will have a very good chance of learning normal communication skills. Also, twice as many babies with hearing loss are found when all babies are tested rather than just those who are "at risk" for hearing problems.



How is my baby tested?

Since healthcare workers at the hospital cannot ask your baby if he or she hears, they have to use computers to help them! Two methods are used. Your hospital may use one or both of them. One method measures the response of your baby's ear to sound. This is the "otoacoustic emissions" method, or the *OAE* test. The other method measures the response of your baby's brain to sound. This is the "auditory brain stem response" method, or the *ABR* test. Both tests are completely painless and can be done while your baby is asleep!



What types of hearing loss can Newborn Hearing Screening identify?

The Maryland program identifies the two basic types of hearing loss: sensorineural and conductive. *Sensorineural* hearing loss is caused by problems in the baby's inner ear. Although this type of hearing loss cannot be cured, it can be treated. *Conductive* hearing loss is caused by problems in the baby's middle ear. This type of hearing loss sometimes goes away by itself. If not, it can be successfully treated medically or surgically. The screening tests indicate whether the problem is in one ear or both ears.

Is my permission required?

The State does not require parental consent for newborn hearing screening. But we want you, as a parent, to understand why hearing is tested.

How much will these tests cost?

There are no fees for hearing screening in the hospital.

When is the best time to test my baby?

Your newborn baby's hearing will be tested before leaving the hospital. The best time to screen is when your baby is quiet and comfortable. If your baby did not pass the first screening, you will be asked to return with your baby for another screening before the baby is one month old.

How soon will I know the results?

In most cases, you will know the results of the hearing screening before you leave the hospital. Also, you and your baby's pediatrician will receive a letter from the Health Department that explains the results.

What if I am told that my baby needs a "retest"?

A retest will be needed if your baby did not pass the first screening test. This does not mean that your baby has a hearing loss. If the baby does not pass the retest, the baby will need further evaluation.

If you are asked to have your baby retested, please act quickly!

It is important that we give your baby the best possible chance to develop speech and language. Studies show that if hearing loss is not diagnosed and treated before 6 months of age, language development is delayed by almost 50%. This means that a 4 year old may only have the language skills of a 2 year old.

How can I make it easier for doctors to help my baby?

Please give your doctor or clinic a telephone number where you can be reached. If your address changes after you leave the hospital, it is important that your doctor or clinic know how to reach you. Please give them your new address and telephone number.

If your doctor asks you to bring the baby in for retesting, do so as soon as you can. If your child does have a disorder or a hearing loss, your prompt action in following the doctor's instructions can be very important.

As a parent, you can help to assure the health of your child and the new generation by your cooperating with Maryland's Newborn Screening Program.

PERMISSION FORM - NEWBORN SCREENING

Newborn babies can be tested for some conditions that cause mental retardation or other serious health problems. Treatment, if it is started early, can usually prevent these problems. Testing all babies is important because babies with these conditions usually look normal.

Maryland tests babies for all the conditions recommended by the March of Dimes and the American College of Medical Genetics. A list of the conditions in the Maryland program is attached. However, some conditions are better understood than others. Effective treatment is available for many of these conditions. Treatments are being developed for others. Every baby found to have a disorder will have access to the best treatment available.

The tests are done on a small amount of the baby's blood. The blood is collected by pricking the baby's heel. This has already been done on over two million babies born in Maryland. Complications such as infection of the heel are very rare.

You will be told if any of your baby's screening tests are not normal. An abnormal test does not always mean that your baby has one of these conditions. It means another test should be done to find out if the baby needs treatment.

Screening tests done at a few days of age will find almost all babies with these conditions. However, a few babies may be missed. Therefore, your doctor or clinic should test your baby again between one and four weeks of age. This gives the program another chance to find babies whose problem did not show up on the first test. Even if the baby seems healthy, the test should be repeated.

If you have any questions, please ask your doctor or nurse.

CHECK THE ONE STATEMENT THAT APPLIES

_____ I have read the above. My questions have been answered.
YES, I agree to the collection of blood sample from my baby for the Newborn Screening Tests.

_____ I have read the above. My questions have been answered.
NO, I do not agree to the collection of a blood sample from my baby for the Newborn Screening Tests.

Witness Date Parent or Guardian Date

Disorders in the Maryland Newborn Screening Panel

Amino Acid Disorders

- Phenylketonuria (PKU)
- Homocystinuria
- Tyrosinemia

Urea Cycle Disorders

- Argininosuccinic aciduria
- Argininemia
- Citrullinemia

Organic Acid Disorders

- BCK (branched chain ketoaciduria) also called MSUD (maple syrup urine disease)
- Biotinidase deficiency
- Glutaric Acidemia Type I (GA I)
- 3-Hydroxy-3-Methyl-CoA Lyase deficiency (HMG CoA Lyase Deficiency)
- Isobutyryl-Co-A Dehydrogenase Deficiency
- Isovaleric Acidemia
- 2-Methylbutyryl-CoA Dehydrogenase Deficiency
- 3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC Deficiency)
- 3-Methylglutaconyl-CoA Hydratase Deficiency
- Methylmalonic Acidemia (MMA)
- Mitochondrial Acetoacetyl-CoA Thiolase Deficiency (3-Ketothiolase Deficiency)
- Multiple Carboxylase Deficiency
- Propionic Acidemia (PA or PPA)

Fatty Acid Oxidation Disorders

- Carnitine Uptake Disorder
- Carnitine/Acylcarnitine Translocase Deficiency (Translocase Deficiency)
- Carnitine Palmitoyl Transferase I Deficiency (CPT I Deficiency)
- Carnitine Palmitoyl Transferase II Deficiency (CPT II Deficiency)
- Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD Deficiency)
- Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD Deficiency)
- Multiple Acyl-CoA Dehydrogenase Deficiency (MADD or Glutaric Acidemia Type II or GA II)
- Short Chain Acyl-Co-A Dehydrogenase Deficiency (SCAD Deficiency)
- Short Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (SCHAD Deficiency)
- Trifunctional Protein Deficiency (TFP Deficiency)
- Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD Deficiency)

Endocrine Disorders

- Hypothyroidism
- CAH (Congenital Adrenal Hyperplasia)

Carbohydrate Metabolism Disorders

- Galactosemia

Hemoglobin Disorders

- Sickle Cell Disease (SCD)

Cystic Fibrosis

Hearing

All babies are screened before hospital discharge

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