



Parent Brochure for the Kentucky Newborn Screening Program.

DO YOU NEED MORE INFORMATION?

Talk with your child's doctor, your midwife, your local health department or contact our office at (502) 564-3756 ext. 3761 or

go to our website at

<http://chfs.ky.gov/dph/mch/ecd/newbornscreening.htm>

or

just type in

KY Newborn Screening

in any internet search engine.



WHAT IS NEWBORN SCREENING?

The state of Kentucky requires by law that all newborns have a blood test performed within 1-2 days after birth by the staff at the birth hospital or by the midwife who delivered your child. This test can identify any of 47 "hidden" disorders that can cause serious problems for your child if not treated soon after birth. Even if your baby looks and acts healthy, he or she may have one of these disorders.

WHAT DOES "HIDDEN" DISORDERS MEAN?

Hidden disorders are problems with the body's ability to make and use hormones, proteins, sugars, or blood cells. They are usually referred to as "inherited disorders." These disorders are difficult or impossible for you or your baby's doctor to find just by looking at your baby. These disorders can be found in the blood when a newborn screening test is performed and treatment can start before the disorder can cause serious health problems to your child. These disorders can cause mental retardation or death if not found and treated soon after birth. Babies who have these disorders may seem normal, but if not found and treated will slowly get very sick. It is thought that as many as 5% of all SIDS (sudden infant death syndrome) cases are caused by these types of "hidden" disorders.



HOW IS YOUR CHILD SCREENED?

When your child is between 1 and 2 days old, the hospital nursery staff or the midwife who delivered your child will prick your baby's heel to get a few drops of blood. It will feel like being stuck with a pin.

The blood drops are put on special paper, dried, and mailed to the state lab for testing.



Hatched area () indicates safe areas for puncture site.



HOW WILL YOU GET THE RESULTS OF THE SCREEN?

Your child's doctor will have a copy of the test results. You can ask about these results when you take your baby to the doctor or clinic for a regular check-up.



I'VE GOTTEN A LETTER SAYING MY CHILD NEEDS A "REPEAT SCREEN." WHAT DOES THIS MEAN?

We are required by law to notify you directly if a second screening test has been requested from your child's doctor and has not been received in our lab in a timely fashion. Please know that the need for a repeat newborn screen does not mean that your child has one of these "hidden" disorders. There are several reasons why your child may need to have another blood test performed to repeat the test. Some of these may include:



IMPROPER COLLECTION: This may mean that the specimen was mailed before it was properly dried or that it was not mailed soon enough for the specimen to be used.

UNSATISFACTORY SPECIMEN: This may mean that not enough blood was obtained for the test or that too much blood was put on the paper to the point where the results could not be determined.

"TOO EARLY" SPECIMEN: The specimen was collected before the infant was 24 hours old. A second specimen will need to be collected as soon as possible.

TRANSFUSION: A specimen that is collected from your baby after your child has had a blood transfusion will need to be repeated at 72 hours after the transfusion and again at 90 days after the transfusion.



ABNORMAL, INCONCLUSIVE, OR INDETERMINATE: Any of these results may mean that a "hidden" disorder is present.

WHAT SHOULD I DO NOW?

IF YOU ARE ASKED TO HAVE YOUR BABY'S TEST REPEATED, CONTACT YOUR CHILD'S DOCTOR TODAY FOR AN APPOINTMENT! IT IS IMPORTANT THAT REPEAT SCREENING BE DONE RIGHT AWAY. YOUR BABY'S HEALTH DEPENDS ON YOU.



IF MY BABY HAS ONE OF THESE DISORDERS, CAN IT BE CURED?

Each defect is different. But if treated early, many infants grow up to lead a normal healthy life. In a few cases, the defects may not be completely treatable. The early diagnosis and treatment of the defect will allow your baby the best chance of normal growth and development.

WHAT DISORDERS ARE INCLUDED IN THE NEWBORN SCREEN?



ENDOCRINE DISORDERS

In **Congenital Adrenal Hyperplasia (CAH)**, lack of an enzyme may cause shock or death in infants because the kidneys lose too much salt.

In **Congenital Hypothyroidism (CH)**, the lack of enough thyroxine (a hormone) can lead to mental and growth retardation.

HEMOGLOBINOPATHIES

Sickle Cell Disease is a disorder of the oxygen-carrying part of the red blood cells and can lead to a high risk of painful infections and anemia.

Hemoglobin S beta Thalassemia Disease and Hemoglobin S/C Disease are similar disorders of the red blood cells with different degrees of symptoms ranging from no symptoms to severe complications.

Sickle Cell Trait does not have the problems associated with the disease but there is the risk of the parents having another child that will have sickle cell disease.

Other variant Hb-pathies (including Hb E)

AMINO ACID DISORDERS *Argininemia, Arginosuccinic Acidemia, Benign Hyperphenylalaninemia, Citrullinemia types 1 & 2, Homocystinuria, Hypermethioninemia, Maple Syrup Urine Disease, Phenylketonuria, and Tyrosinemia type 1, 2, & 3* are all disorders



where the body lacks a specific enzyme and cannot process proteins. They require treatment with a special diet. Complications of these disorders are mental retardation, brain damage, liver damage, eye and circulation problems.

FATTY ACID DISORDERS *Carnitine/acylcarnitine translocase deficiency, Carnitine palmitoyltransferase deficiency types 1 & 2, Carnitine uptake defect, Ethylmalonic encephalopathy, Isobutyryl-CoA dehydrogenase deficiency, Long-chain L-3 hydroxyacyl-CoA dehydrogenase, Medium-chain acyl-CoA dehydrogenase deficiency, Short-chain acyl-CoA dehydrogenase deficiency, Trifunctional protein deficiency, Glutaric acidemia type 2, and Very long-chain acyl-CoA dehydrogenase deficiency* are disorders where the body lacks the proper enzyme to process fats for energy. They require treatment with a special diet. Complications can include problems with the heart, liver, lungs, muscle weakness, seizures, and even death.

ORGANIC ACID DISORDERS *2-Methyl 3-hydroxybutyric aciduria, 3-Methylglutaconic aciduria, 3-methylcrotonyl CoA Carboxylase Deficiency, Beta Ketothiolase Deficiency, Glutaric Acidemia type 1, Hydroxymethylglutaric aciduria, Isovaleric Acidemia, Malonic aciduria, Methylmalonic Acidemia, Methylmalonic Acidemia Mutase Deficiency, Multiple Carboxylase Deficiency, and Propionic Acidemia* are disorders in processing proteins, maintaining the acid-base balance in the blood. Special diets are required. Medication and treatments vary according to disorder. Complications include mental retardation, poor muscle tone, seizures, breathing problems, coma, and possibly death.

OTHER DISORDERS *Biotinidase Deficiency, Cystic Fibrosis, and Galactosemia* are other disorders for which your newborn is screened. *Biotinidase Deficiency* occurs when the body lacks an enzyme to process the essential vitamin, biotin. *Vision, hearing, and skin complications can result from this condition. This disorder is treated with medication. Cystic Fibrosis* is a problem with the way the body handles salt and is handled with treatments and medications. *Galactosemia* occurs when the body lacks an enzyme to process milk sugars. A special diet is required.

CAN MY CHILD'S DOCTOR TAKE CARE OF THIS PROBLEM?

The Kentucky Newborn Screening Program has contracted with both the University of Kentucky and the University of Louisville to provide special consultation if your child needs more testing to determine the presence of one of these disorders. If your child does have one of these disorders, the university specialists will determine proper treatment for your child and will discuss your child's needs with your local doctor.



WHAT CAN YOU DO TO HELP YOUR BABY?

Be sure to give the birthing hospital staff and your child's doctor your correct phone number and address so you can be reached quickly in the event



that your child has a newborn screening test that needs to be repeated. Provide the hospital staff and your child's doctor the name and telephone number of a friend, relative, or neighbor that can reach you quickly in the event that the newborn screening

test needs to be repeated or your child needs to be immediately referred to a specialty clinic. And, if you're contacted about the need for repeat newborn screening to be performed, please respond quickly. Your child's health depends on it!

