Disorders Identified by the New York State Newborn Screening Program

Group		Condition
Endocrinology		Congenital adrenal hyperplasia (CAH)
		Congenital hypothyroidism (CH)
Hematology, Hemoglobinopathies		Hb SS disease (Sickle cell anemia)
		Hb SC disease
		Hb CC disease
		Other hemoglobinopathies
Infectious Diseases		HIV-1 infection (HIV-1)
Inborn Errors of Metabolism	**Amino Acid Disorders**	Homocystinuria (HCY)
		Hypermethioninemia (HMET)
		Maple syrup urine disease (MSUD)
		Phenylketonuria (PKU) and Hyperphenylalaninemia (HyperPhe)
		Tyrosinemia (TYR)
	Fatty Acid Oxidation Disorders	Carnitine-acylcarnitine translocase deficiency (CAT)
		Carnitine palmitoyltransferase I (CPT-I) and II (CPT-II) deficiencies
		Carnitine uptake defect (CUD)
		2,4-Dienoyl-CoA reductase deficiency (2,4DI)
		Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
		Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
		Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)
		Medium/short-chain hydroxyacyl-CoA dehydrogenase deficiency (M/SCHAD)
		Mitochondrial trifunctional protein deficiency (TFP)
		Multiple acyl-CoA dehydrogenase deficiency (MADD) (also known as Glutaric acidemia type II (GA-II))
		Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
		Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)
	Organic Acid Disorders	Glutaric acidemia type I (GA-I)
		3-Hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG)
		Isobutyryl-CoA dehydrogenase deficiency (IBCD)
		Isovaleric acidemia (IVA)
		Malonic acidemia (MA)
		2-Methylbutyryl-CoA dehydrogenase deficiency (2-MBCD)
		3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
		3-Methylglutaconic acidemia (3-MGA)
		2-Methyl-3-hydroxybutyryl-Co-A dehydrogenase deficiency (MHBD)
		Methylmalonyl-CoA mutase deficiency (MUT), Cobalamin A,B (Cbl A,B) and Cobalamin C,D (Cbl C,D) cofactor deficiencies and other Methylmalonic acidemias (MMA)
		Mitochondrial acetoacetyl-CoA thiolase deficiency (beta-ketothiolase deficiency) (BKT)
		Multiple carboxylase deficiency (MCD)
		Propionic acidemia (PA)
	Urea Cycle Disorders	Argininemia (ARG)
		Argininosuccinic acidemia (ASA)
		Citrullinemia (CIT)
		Hyperammonemia/hyperornithinemia/homocitrullinemia (HHH)
Other Genetic Conditions		Biotinidase deficiency (BIOT)
		Cystic Fibrosis (CF)
		Galactosemia (GALT)
		Krabbe Disease

For more information on the New York State Newborn Screening Program and the disorders in the panel please visit our webpage at www.wadsworth.org/newborn/index.htm

The New York State Newborn Screening Program is a service provided by the State Department of Health to families with newborn babies. Comments on this brochure are always welcome. Please write to:

Newborn Screening Program
Wadsworth Center
New York State Department of Health
P.O. Box 509
Albany, NY 12201-0509
www.wadsworth.org/newborn/index.htm

State of New York
George E. Pataki, Governor

Department of Health
Antonia C. Novello, M.D., M.P.H., Dr.P.H., Commissioner

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Why is my baby tested?

To help ensure that your baby will be as healthy as possible. A blood test provides important information about your baby's health that you or even your doctor might not otherwise know. The Newborn Screening Program identifies those few infants who may have one of several rare disorders and infants who have been exposed to HIV, the virus that causes AIDS. With early diagnosis and medical treatment, serious illness can often be prevented.

Is newborn screening a new program?

No. Every state has a newborn screening program. The New York State program began in 1965. Some disorders can affect a child very early in life – even within the first few days. For this reason, prompt testing and diagnosis are important.

For how many disorders is my baby tested?

The number has increased from one in 1965 to more than 40 today. They are listed on the other side of this brochure. Although these disorders are rare, they are usually serious. Some may be life-threatening. Others may slow down the baby's physical development or cause mental retardation or other problems if undetected and untreated.

Early treatment is very important!

But my baby seems very healthy. Are these tests still needed?

Yes. Most infants with a disorder identified by the Newborn Screening Program show no signs of the disorder immediately after birth. With special laboratory tests, the program can identify an infant who may have one of these disorders and alert the baby's doctor of the need for special care. Usually this can be done before the baby becomes ill.

But children in our family have never had any of those health problems.

Parents who have already had healthy children do not expect any problems and they are almost always right. These disorders are quite rare and the chances are excellent that your child will not have one.

How is my baby tested?

All of the tests are performed on a tiny sample of blood obtained by pricking the baby's heel. The sample is usually taken on the day the baby is discharged from the hospital. The sample is sent for testing to the laboratories of the State Department of Health in Albany.

Will I get the test results?

Your baby's doctor or clinic will be informed of the results and will contact you immediately if anything is wrong. But, as a responsible parent, you should ask about the results when you bring your baby to the doctor or clinic for the baby's first check-up. The pink form given to you by the nurse will tell you how to get the test results from your doctor.

If all the tests are screen-negative, does that mean my baby will be healthy?

The Newborn Screening Program screens for only a few of the many disorders a baby could have. In addition, some babies with these disorders may not be identified because of differences in the way the blood is collected or the kinds of tests used. You should bring your baby to the doctor or clinic on a regular basis for care. Always watch your baby for unexpected behavior and call a doctor immediately if things don't seem right.

Does a "retest" mean my baby may have a disorder?

Not necessarily. Retesting may be needed for a number of reasons. The most common is that the first sample contained too little blood to complete all tests. This does not mean there is anything wrong with your baby. It simply means that another sample must be taken so that all the tests can be done.

Often, when first test results suggest a problem, the results are not considered final until the tests are done again. This requires a new blood sample. In general, the doctor will discuss the need for further evaluation only when a baby's test is unusual for a second time. On very rare occasions, because of the potential severity of a particular disorder, the doctor will treat the child immediately while waiting for the results of the second series of tests. If you are asked to have your baby retested, please act quickly, so the repeat test can be done immediately.

What if my baby has one of these disorders? Can it be cured?

None of these disorders can be cured. However, the serious effects can be lessened – and often be prevented completely – if a special diet or other medical treatment is started early. Most of these disorders are very complicated to treat and medical care should be coordinated by a specialist in the specific disorder.

In the case of HIV, less than ten percent of babies who test positive are actually infected and will need treatment.

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

That depends on the disorder. Some disorders are genetic and inherited by children from their parents. Many families seek genetic counseling to better understand how their child got the disorder and to understand risks to their future children and other family members. Other disorders, such as hypothyroidism and HIV, are not inherited. Hypothyroidism has many causes. And HIV infection is caused by a virus, not a gene.

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