

# Newborn Screening

## POCKET FACTS

As a public health program, the goal of newborn screening is early identification of children at increased risk for selected metabolic or genetic diseases so that medical management can be promptly initiated to avert metabolic crises and prevent irreversible neurological and developmental sequelae. Early identification of these conditions is crucial, as timely intervention can lead to a significant reduction of morbidity, mortality and associated disabilities in affected infants.<sup>1</sup>

This pocket guide will present basic information about the current 29 core conditions recommended for screening by the American College of Medical Genetics (ACMG) with a brief overview of screening protocol, along with talking points for your patients and resources where public health professionals can find additional information about screening for these disorders.

The March of Dimes supports comprehensive newborn screening for all babies in the U.S., regardless of their place of birth. We urge every state to screen every baby for at least 29 core conditions, including hearing loss. For each of these conditions, screening has a documented benefit to the child, and a reliable test that enables early detection is available. The March of Dimes also urges states to provide test results for additional conditions that are reliably revealed during screening for the core conditions even when efficacious treatment is not yet documented.<sup>2</sup>



Table 1. Recommended Newborn Screening Tests: 29 Core Disorders

Name	Abbreviation	Incidence <sup>1</sup>	Description/Presentation Without Treatment	With Treatment
<b>3 hemoglobinopathies associated with Hb/S allele</b>				
Sickle Cell Anemia	Hb SS	>1 in 5,000; 1 in 400 in African-Americans <sup>2</sup>	Inherited anemia that can cause pain, damage to vital organs, stroke and sometimes death.	Beginning in infancy: vigilant medical care, all regular childhood vaccinations and begin treatment with penicillin to reduce the risk of infections such as pneumonia and meningitis. Treatment may include intermittent pain medications and blood transfusions depending on symptoms.  Routine treatment with penicillin may not be universally recommended.
HB S/Beta-Thalassemia <sup>3</sup>	Hb S/BTH	>1 in 50,000	A form of sickle cell anemia, the child inherits one sickle cell gene and one gene for beta thalassemia, another inherited anemia. Symptoms are often milder than Hb SS, but severity varies.	Routine treatment with penicillin may not be universally recommended.
HB S/C Disease <sup>3</sup>	Hb S/C	>1 in 25,000	Another form of sickle cell anemia, the child inherits one sickle cell gene and one gene for another hemoglobin variant called HbC. This form is often milder than Hb SS.	
<b>6 amino acid disorders</b>				
Phenylketonuria	PKU	>1 in 25,000	Individuals cannot process phenylalanine, which can accumulate and cause severe mental retardation if not detected and treated soon after birth.	Adherence to a low-phenylalanine diet at least throughout childhood and adolescence and for females during pregnancy.
Maple Syrup Urine Disease	MSUD	<1 in 100,000	MSUD has a wide spectrum of presentations, from mild to severe, although it can be lethal if untreated. Babies with MSUD appear normal at birth then quickly deteriorate neurologically, and the urine smells of maple syrup.	A special, low-protein diet, sometimes supplemented by thiamin, continued indefinitely.
Homocystinuria	HCY	<1 in 100,000	Individuals lack an enzyme to convert homocysteine to cystathionine, which is needed for brain development. Without treatment, HCY leads to mental retardation, eye problems, skeletal abnormalities and stroke.	A special diet, vitamins B6 or B12, and betaine.
Citrullinemia	CIT	<1 in 100,000	Untreated individuals with CIT have a build-up of citrulline and ultimately ammonia during the newborn period or infancy that can result in seizures, coma, brain damage and death.	Normal development is possible with early detection and treatment of a low-protein diet, medications to prevent ammonia build-up and nutritional supplements.
Argininosuccinic Acidemia	ASA	<1 in 100,000	Symptoms commonly begin in the first few days of life (sometimes later in infancy or childhood) with a build-up of argininosuccinic acid and ultimately, ammonia resulting in brain swelling, coma and sometimes death. Early treatment can be lifesaving, but all individuals with ASA have episodes of ammonia build-up and most have some degree of brain damage.	A low-protein diet, avoiding fasting, medications to prevent ammonia build-up, nutritional supplements and sometimes liver transplant.
Tyrosinemia Type I	TYR I	<1 in 100,000	The absence of an enzyme causes build-up of succinylacetone, a byproduct of tyrosine, in the liver. Without treatment, symptoms begin in the first weeks or months of life and progress to liver or kidney failure, nerve damage and death.	Drug treatment, sometimes with a low-protein diet, can prevent liver and kidney damage in most patients. Hepatic transplant may be considered.
<b>5 fatty oxidation disorders</b>				
Medium-Chain Acyl-CoA Dehydrogenase Deficiency	MCAD	>1 in 25,000	Infants seem well, then suddenly develop seizures caused by low blood sugar, liver failure, coma and death.	Treatment consists of avoidance of fasting and the addition of nutritional supplements.
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	VLCAD	>1 in 75,000	Symptoms can present at any age from the newborn period to adulthood, but tend to be more severe in infants. Left untreated, infants develop heart and liver failure and can die during the first year of life.	Treatment includes a high-carbohydrate/low-fat diet, nutritional supplements, and the avoidance of fasting and prolonged exercise.
Long-Chain 3-OH Acyl-CoA Dehydrogenase Deficiency	LCHAD	>1 in 75,000	Symptoms resulting in heart, lung, or liver failure and death can present soon after birth, or in other cases low muscle tone, developmental delay, heart, lung or liver failure may develop later in infancy or childhood, often following an illness. Early treatment can prevent death, but some children still develop symptoms.	Treatment includes a high-carbohydrate/low-fat diet, nutritional supplements and avoidance of fasting. Women pregnant with fetus with LCHAD are at an increased risk of developing acute fatty liver of pregnancy and other complications.
Trifunctional Protein Deficiency	TFP	<1 in 100,000	A seemingly well infant can die of what appears to be sudden infant death syndrome or develop low muscle tone, seizures, heart failure and coma, often following illness.	Treatment includes a strict avoidance of fasting, a low-fat diet and nutritional supplements.
Carnitine Uptake Defect	CUD	<1 in 100,000	In this condition, cells are unable to bring in carnitine (necessary to transport fatty acids across mitochondrial membranes) from the blood. Symptoms include episodes of hypoglycemia and sudden unexpected death in infancy. Older children may present with progressive heart failure.	Early diagnosis and treatment with carnitine permits normal development.
<b>9 organic acid disorders</b>				
Isovaleric Acidemia	IVA	<1 in 100,000	Individuals with IVA cannot process leucine, which can cause coma, permanent neurological damage and death in the newborn period. Later onset cases can develop in infancy or childhood following an infectious illness.	Early treatment with a low-protein diet and nutritional supplements can result in normal development.
Glutaric Acidemia Type I	GAI	>1 in 75,000	GAI is characterized by normal development up to 18 months until a trigger such as a viral illness leads to brain damage, seizures, low muscle tone, cerebral palsy-like symptoms, and death within the first decade of life. Some babies with GAI are born with an enlarged head.	Treatment with dietary protein restriction and L-carnitine supplement along with the prompt treatment of illness and fever can prevent brain damage.
Hydroxymethyl-Glutaric Aciduria or HMG-CoA Lyase Deficiency or 3-OH 3-CH3 Glutaric Aciduria	HMG	<1 in 100,000	An inability to process leucine results in low blood sugar and the accumulation of several organic acids especially after illness or fasting and can result in brain damage, mental retardation, coma and death without treatment.	Avoiding fasting and a diet low in leucine and fat and high in carbohydrates can lead to normal development.
Methylmalonic Acidemia due to Mutase Deficiency	MUT	>1 in 75,000	A lack of enzymes required to activate several biotin-dependent enzymes results in a build-up of lactic acid and other organic acids. Symptoms usually begin before 15 months of age and may include skin rashes, hair loss, and brain damage, coma and death without treatment.	Biotin can result in normal growth and development.
3-Methylcrotonyl-CoA Carboxylase Deficiency <sup>†</sup>	3MCC	>1 in 75,000	Severity of symptoms varies greatly, but a processing defect of four essential amino acids and other substances results in illness in the first week of life and can result in death during the first month of life.	Treatment with a special metabolic formula/low-protein diet and medication minimizes but cannot prevent central nervous system dysfunction.
Methylmalonic Acidemia CblA and CblB forms	Cbl A,B	<1 in 10,000	The inability to process leucine can lead to brain damage, seizures, liver failure and death in infancy or no symptoms at all into adulthood. Symptoms often develop after childhood illness.	Treatment with a low-protein diet and, in some cases, nutritional supplements, may be helpful.
Propionic Acidemia	PROP	>1 in 75,000	A defect of vitamin metabolism can lead to build-up of organic acids in the blood and result in brain damage, seizures, paralysis, coma and death. Symptoms can begin in the first week of life, though most affected individuals remain symptom-free.	Treatment of vitamin B12 injections and a special metabolic formula/low-protein diet can often prevent serious problems.
Beta-Kerohiolase Deficiency	BKT	<1 in 100,000	This defect in the processing of four amino acids leads to brain damage, coma and death in the newborn period without treatment.	Treatment includes a low-protein diet and nutritional supplements. Even with treatment, some children experience developmental delays, frequent infections and heart problems.
<b>6 unrelated conditions</b>				
Congenital Hypothyroidism	CH	>1 in 5,000	A deficiency of thyroid hormone severely retards growth and brain development.	With prompt intravenous treatment to keep blood sugar levels up and blood acid levels down during illness, children can develop normally. Parents must be alert to early signs of illness. Additional treatments may include avoidance of protein-rich diets and long-term treatment with bicarbonate.
Biotinidase Deficiency	BIOT	>1 in 75,000	The deficiency of biotinidase, the enzyme that recycles the vitamin biotin, may cause frequent infections, alopecia, skin rash, uncoordinated movement, hearing loss, seizures and mental retardation. Left untreated, the deficiency can lead to coma and death.	With prompt intravenous treatment to keep blood sugar levels up and blood acid levels down during illness, children can develop normally. Parents must be alert to early signs of illness. Additional treatments may include avoidance of protein-rich diets and long-term treatment with bicarbonate.
Congenital Adrenal Hyperplasia	CAH	>1 in 25,000	CAH includes a set of conditions resulting from defects in the synthesis of hormones produced by the adrenal gland. In female infants, CAH can result in masculinization of the genitals (ambiguous genitalia). Severe forms of CAH cause life-threatening salt loss from the body if untreated.	Treatment includes salt and hormone replacement.
Hearing Loss	HEAR	>1 in 5,000; up to 3-4 in 1,000 newborns <sup>4</sup>	Without early testing, hearing loss is usually not diagnosed until 3-4 years of age, and children often have delayed speech and language development.	Early diagnosis allows hearing aids by 6 months of age and helps prevent speech and language problems.
Cystic Fibrosis	CF	>1 in 5,000	Abnormalities in the CF protein result in lung and digestive problems, often beginning in early infancy. Recurrent pneumonia and failure to thrive are common and death occurs at an average of 30-35 years.	Early diagnosis and treatment may improve the growth of babies and children with CF. Treatment varies depending on severity, but may include a high-calorie diet supplemented with vitamins and medications to improve digestion, respiratory therapy to help clear mucus from lungs, and medications to improve breathing and prevent lung infections.
Classical Galactosemia	GALT	>1 in 50,000	A missing enzyme prevents the conversion of galactose (sugar from the breakdown of lactose in milk) to glucose, another simple sugar. Galactose then accumulates and damages vital organs, leading to blindness, severe mental retardation, infection and death.	Milk and dairy products must be eliminated from the diet for life. Though treatment dramatically improves symptoms, there continues to be a chance of developmental delays.

<sup>†</sup> 3-Methylcrotonyl-CoA carboxylase deficiency: An abnormal result on newborn screening may be due to abnormal metabolites in the mother, not the baby and can be clarified by further testing of the infant.

**NOTE:** Currently each state operates by law its own newborn screening (NBS) program. Thus, individual programs vary widely in the number and types of conditions for which they screen.<sup>4,5</sup> Furthermore, the list of disorders for which screening tests are available is expanding. For current information on tests for 29 core conditions, visit the March of Dimes Peristats Web site where NBS data is compiled in collaboration with National Newborn Screening & Genetics Resource Center.

Go to [www.marchofdimes.com/peristats](http://www.marchofdimes.com/peristats)

## Overview of screening protocol

Specimens are samples of blood collected from infants by saturating marked areas of a filter paper card with blood obtained by skin puncture of the heel. Many factors determine the acceptability and quality of a specimen: the timing of collection, the completeness and accuracy of patient and specimen information, adequacy of the blood specimen and its subsequent handling. Problems with any of these may render a specimen invalid.

### Timing of specimen

- The American Academy of Pediatrics recommends that the specimen **not** be collected until the newborn is at least 24 hours old.<sup>1</sup>
- Analysis of screening results suggests that a specimen taken on the second day of life is usable for testing, with a slightly increased risk of not detecting an abnormal condition.<sup>2-6</sup>
- Discharging an infant without collecting a specimen, with intent to collect it later, greatly increases the risk of missing an infant with one of the screened conditions.<sup>7</sup>

Table 2. Summary for Specimen Collection Timing

Infant Status	Time of Collection
Normal, Healthy	Day 1: If to be discharged from hospital or birthing center & repeated on day 3-5. Specimen should be collected as close to time of discharge as possible. Day 2: Acceptable Day 3-5: Optimum
Transfused	Prior to transfusion; or if no pretransfusion collection was taken, three days after most recent transfusion; with repeat three months after final transfusion.
Total Parenteral Nutrition (TPN)	Prior to initiation of TPN; or if no pre-TPN collection was taken, three days after most recent TPN; with repeat three months after final TPN.
Premature, Sick or Extended Stay	Prior to transfusion — any age & on day 3-5 or three days after most recent transfusion & at discharge or at one month of age, whichever comes first.

### GO TO...

→ [Kaye CI and Committee on Genetics, American Academy of Pediatrics. Table 2. "Effect of sample timing, preterm birth, diet, transfusion and total parenteral nutrition on newborn screening results."](#)<sup>1</sup>

→ [March of Dimes "Timing of specimen collection" www.marchofdimes.com/professionals/24279\\_9622.asp](#)

### Collection of blood specimen: resources

→ [The Clinical and Laboratory Standards Institute \(formerly NCCLS\) has produced a video "Making a Difference Through Newborn Screening: Blood Collection on Filter Paper, demonstrating proper heel stick techniques."](#)<sup>1</sup>

→ [Whatman, Inc. and the New York State Department of Health](#)

+ "Neonatal Screening: Blood Specimen Collection and Handling Procedure": [www.marchofdimes.com/files/Neonatal.pdf](#)

- Illustration of proper collection techniques

+ "Simple Spot Check": [www.marchofdimes.com/files/Simple\\_Spot.pdf](#)

- Illustrates examples of improper specimen collection

→ [March of Dimes](#) Web page includes an equipment checklist, skin puncture site information and a detailed eight step procedure. Since laboratory analysis of the specimen depends on an assumed amount of blood in the filter paper circle, it is imperative to carefully follow established procedures. Numerous studies have shown the variability occurring due to improper technique in specimen collection.<sup>2-5</sup> [www.marchofdimes.com/professionals/24279\\_9623.asp](#).

### Specimen handling

Contamination of the filter paper (e.g., by skin oils, latex and powder) or the areas within the circle of the filter paper (e.g., by feeding formulas, antiseptic solutions, water, lotions, powder, etc.) should be avoided since test results can be affected or invalidated, respectively. The perfect specimen:

- Has all information legibly and accurately recorded on the blood collection form required by your state.
- Is collected from an infant between 48 and 120 hours of age (third to fifth day of life).
- No foreign substances contaminate the filter paper.
- The blood completely fills all printed circles and is applied evenly on one side of the filter paper, free of layering and clots.
- Is dried for at least four hours on a flat, non-absorbent surface, away from direct heat and sunlight.
- Within 24 hours of collection, is sent to your state's Newborn Screening Program as specified.

### Invalid specimens — when to repeat collection procedure

Certain types of specimens are known to give erroneous laboratory results. In accordance with laboratory regulations, these cannot be tested and are termed invalid. This delays the screening of the newborn, and requires that the submitter repeat the collection procedure. (The greatest number of requests for repeat specimens results from an invalid specimen.)<sup>1</sup> The New York State testing laboratory classifies unsuitable specimens into eight categories:<sup>2</sup>

- Quantity of blood insufficient
- Blood spots appear scratched or abraded
- Blood spots wet or discolored
- Blood spots appear supersaturated
- Blood spots appear diluted
- Blood spots exhibit "serum rings"
- Blood spots appear clotted or layered
- Specimen delivered to the laboratory more than 14 days after collection

### Results and follow-up

"Newborn screening is more than testing — it always should be part of a system that includes screening tests, follow-up, diagnosis, treatment and evaluation as necessary."<sup>1</sup> The American Academy of Pediatrics Newborn Screening Task Force published a report in 2000 listing follow-up procedures as an important part of developing a public health infrastructure for newborn screening systems.

### GO TO...

→ [American Academy of Pediatrics National Center of Medical Home Initiatives for Children with Special Needs, Metabolic/Genetic Screening Activities: www.medicalhomeinfo.org/screening/newborn.html](#)

- Information about disparities, early intervention
- Links to fact sheets for providers and families, policy statements, periodicals/articles, reports/documents, tools and additional resources

→ [American College of Medical Genetics Newborn Screening ACT Sheets and Confirmatory Algorithms: www.acmg.net/resources/policies/ACT/condition-analyte-links.htm](#)

- Table describes "the interrelationships between the conditions screened in newborn screening laboratories and the markers (analytes) used for screening."
- ACTion (ACT) sheet for each marker — short term actions to help communicate with family and determine appropriate follow-up steps for an infant that has screened positive

• [Algorithm for each marker — overview of the basic steps involved in determining the final diagnosis](#)

To find out about your state's regulations on disseminating results and follow-up protocols, contact your state newborn screening program. Go to the resource section in this guide to find out how.

### What abnormal results mean:

#### Talking points regarding a "positive test," "not-normal screening result" or "screen-positive infants"

"An abnormal result does not mean that the baby has the indicated illness. Overall, most babies who have an abnormal screening result turn out not to have the illness. However, an abnormal result does mean that the child should have additional testing to confirm or rule out the condition."<sup>2</sup>

#### A positive test result DOES NOT MEAN:

- That the baby has the indicated condition (most babies who have a positive test result turn out *not* to have the condition).
- That the test was diagnostic. The initial screening provides only preliminary information that must be followed up with more specific diagnostic testing.
- That it is a false positive because the baby appears to be in good health. The screening process is successful when affected babies are identified before onset of symptoms.

#### A positive test result MEANS:

- The test result was out of the normal range.
- The baby should have additional testing to confirm or rule out the condition.
- The newborn screening test may be repeated or another type of test may be requested.

### Resources for Newborn Screening Information

The full breadth of information surrounding the components of newborn screening (NBS) cannot be sufficiently presented in a pocket guide format. Below is a list of selected online resources where additional information can be obtained. Icons adjacent to the URL indicate whether available information and resources are written for health professionals (P) or for consumers and patients (C).

#### American Academy of Pediatrics:

Committee on Genetics, Newborn Screening Introduction and Fact Sheets: [http://aappolicy.aappublications.org/cgi/reprint/pediatrics;118/3/1304.pdf](#) P

- Introduction [http://aappolicy.aappublications.org/cgi/reprint/pediatrics;118/3/e934.pdf](#) P

• Fact sheets to assist pediatricians "in understanding the individual tests, their characteristics, and their strengths and weaknesses"

#### March of Dimes:

[www.marchofdimes.com/professionals/24279.asp](#) P

- What caregivers need to know, an online course by Region 4 Genetics Collaborative
- Clinical issues and considerations
- What providers can do
- Resources for families [www.marchofdimes.com/nbs/](#) C

• Fact sheets for patients: "What you need to know," "What you can do," "Caring for your baby: Testing the Newborn for Metabolic Birth Defects"

- Link to video "A Parent's Guide to Newborn Screening" (English and Spanish)

[www.marchofdimes.com/peristats](#) P C

- Presents state or condition specific tables and maps that can be printed or downloaded into PowerPoint® slides
- Peristats information system compiles data from NNSGRC

#### National Center for Hearing Assessment and Management:

[www.infanthearing.org/legislative/summary/index.html](#) P

- A state-specific summary on universal newborn hearing screening

#### National Coordinating Center for the Genetics and Newborn Screening Regional Collaborative Groups:

[www.nccrcg.org](#) P C

- Links to seven regional collaborative groups
- Project descriptions and resources for patients

#### National Newborn Screening and Genetics Resource Center (NNSGRC):

[http://genes-r-us.uthscsa.edu/](#) P C

- Information about your state's NBS program
- Information about disorders that your state currently screens for
- Calendar of events; U.S. state map with NBS and genetics program links [http://genes-r-us.uthscsa.edu/state\\_contacts.pdf](#) P C
- State NBS program contacts

#### Save Babies Through Screening Foundation:

[www.savebabies.org/index.php](#) C

- Screening FAQ's
- Disease descriptions with support group info and references
- Resource library (items can be ordered from the Web site)
- How to advocate for and promote screening
- List of awareness projects

#### U.S. National Library of Medicine and the National Institutes of Health, Medline Plus:

[www.nlm.nih.gov/medlineplus/newbornscreening.html](#) C

- Latest NBS news, overviews, tutorials, links to information about specific conditions from Genetics Home Reference
- Find related issues, genetics research articles, glossary, directories and organization links

### References:

#### Introduction

- American Academy of Pediatrics. Serving the family from birth to the medical home — A report from the Newborn Screening Task Force. *Pediatrics* 2000;106(suppl):383-427.
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#### Table 1. Recommended Newborn Screening Tests: 29 Core Disorders

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#### Collection of blood specimen: resources

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#### Invalid specimens

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#### Results and follow-up

- Serving the family from birth to the medical home. Newborn screening: a blueprint for the future — a call for a national agenda on state newborn screening programs. *Pediatrics* 2000;106(2 pt 2): 389-427.
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### Acknowledgments

For more information, contact the March of Dimes via e-mail at: [askus@marchofdimes.com](mailto:askus@marchofdimes.com)

To order our catalog or multiple copies of our materials, call 1-800-367-6630.

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