DISORDERS INCLUDED IN THE STEPONE® NEWBORN SCREENING PANEL

DISORDERS DETECTED BY TANDEM MASS SPECTROMETRY

Acylcarnitine Profile

Fatty Acid Oxidation Disorders
Carnitine/Acylcarnitine Translocase Deficiency
Carnitine Palmitoyl Transferase Deficiency Type I[1]
3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency
2,4-Dienoyl-CoA Reductase Deficiency[1]
Medium Chain Acyl-CoA Dehydrogenase Deficiency
Multiple Acyl-CoA Dehydrogenase Deficiency
Neonatal Carnitine Palmitoyl Transferase Deficiency Type II
Short Chain Acyl-CoA Dehydrogenase Deficiency
Short Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency
Trifunctional Protein Deficiency
Very Long Chain Acyl-CoA Dehydrogenase Deficiency
Organic Acid Disorders
3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency
Glutaric Acidemia Type I
Isobutyryl-CoA Dehydrogenase Deficiency
Isovaleric Acidemia
2-Methylbutyryl-CoA Dehydrogenase Deficiency
3-Methylcrotonyl-CoA Carboxylase Deficiency
3-Methylglutaconyl-CoA Hydratase Deficiency
Methylmalonic Acidemias
Methylmalonyl-CoA Mutase Deficiency
Some Adenosylcobalamin Synthesis Defects
Maternal Vitamin B12 Deficiency
Mitochondrial Acetoacetyl-CoA Thiolase Deficiency
Propionic Acidemia
Multiple CoA Carboxylase Deficiency
Malonic Aciduria

Amino Acid Profile

Amino Acid Disorders	
Argininemia	Hyperornithinemia with Gyral Atrophy[1]
Argininosuccinic Aciduria	Maple Syrup Urine Disease
5-Oxoprolinuria1	Phenylketonuria
Carbamoylphosphate Synthetase Deficiency[1]	Classical/Hyperphenylalaninemia
Citrullinemia	Biopterin Cofactor Deficiencies
Homocystinuria	Tyrosinemia
Hypermethioninemia	Transient Neonatal Tyrosinemia
Hyperammonemia, Hyperornithinemia, Homocitrullinuria Syndrome1	Tyrosinemia Type I[2]
	Tyrosinemia Type II
	Tyrosinemia Type III

Other Observations

Hyperalimentation
Liver Disease
Medium Chain Triglyceride Oil Administration
Presence of EDTA Anticoagulants in blood specimen
Treatment with Benzoate, Pyvalic Acid, or Valproic Acid
Carnitine Uptake Deficiency[1]

DISORDERS DETECTED BY OTHER TECHNOLOGIES

Biotinidase Deficiency	Galactosemia
Complete Deficiency	Galactokinase Deficiency
Partial Deficiency	Galactose-1-Phosphate
Glucose-6-Phosphate Dehydrogenase Deficiency	Uridyltransferase Deficiency
Congenital Adrenal Hyperplasia	Galactose-4-Epimerase Deficiency
Salt Wasting 21-Hydroxylase Deficiency	
Simple Virilizing 21-Hydroxylase Deficiency	
Cystic Fibrosis (not valid after 3 months of age)*	
Congenital Hypothyroidism	
Sickle Cell and Other Hemoglobinopathies	
Hemoglobin S, S/C, S/Beta Thalassemia, C, & E Diseases	

1 - There is a lower probability of detection of this condition during the immediate newborn period.
2 - Succinylacetone (SUAC) is the primary marker for Tyrosinemia Type 1.
* For information on DNA Carrier Testing for children over 3 months of age, please call 866.463.6436.

The analyses conducted by PerkinElmer Genetics produce results that can be used by qualified physicians in the diagnosis of disorders described herein. Evidence of these conditions will be detected in the vast majority of affected individuals; however, due to genetic variability, age of patient at time of specimen collection, quality of specimen, health status of the patient, and other variables, such conditions may not be detected in all affected patients.

ABOUT US

PerkinElmer Genetics, a state-of-the-art newborn screening laboratory, provides one of the world's most comprehensive programs for detecting clinically significant inherited disorders. Our services are designed to identify treatable disorders very early in life before irreversible health damage occurs. Since our founding in 1994, we have analyzed blood samples from over 4 million newborns. We strive to improve the health of the most fragile members of our community, our children.

The mission of PerkinElmer Genetics is to provide high quality newborn screening services to improve and save children's lives through early detection and intervention. We continually emphasize the core values of integrity, respect, dignity, and compassion for each baby in our care.

PerkinElmer Genetics provides screening programs across the Americas and around the world including several state mandated programs in the United States.

Our laboratory is accredited by the College of American Pathologists, the Centers for Medicare and Medicaid Services in compliance with the Clinical Laboratory Improvement Amendments, the Joint Commission of Healthcare Organizations and the Commission for Office Laboratory Accreditation. PerkinElmer Genetics also participates in the Newborn Screening Quality Assurance Program administered by the Centers for Disease Control and Prevention.

PerkinElmer Genetics is a part of PerkinElmer, Inc., a global life-sciences company recognized as the leader in newborn screening research and development worldwide.

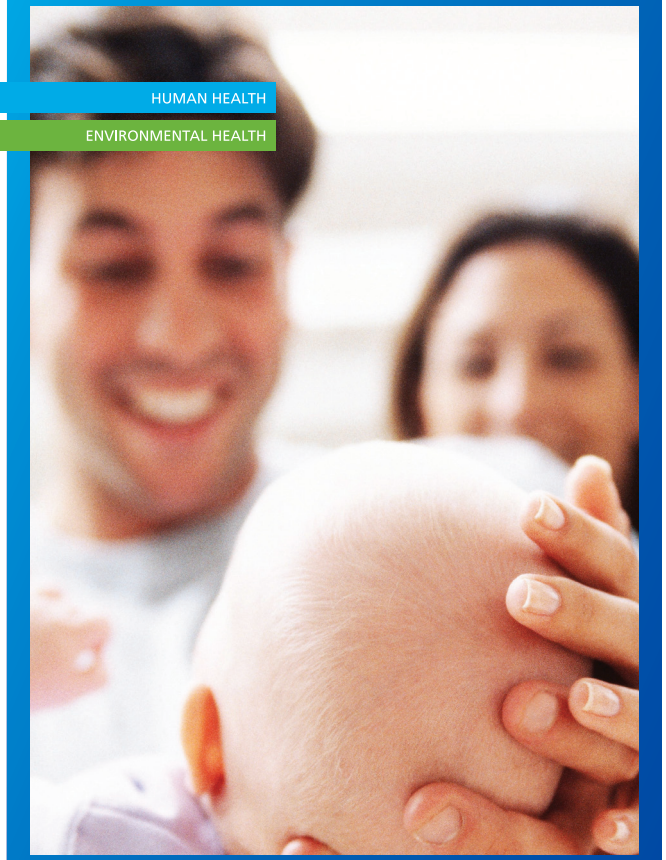
For more information, or to order a screening packet, please call 866.463.6436 or visit www.perkinelmergenetics.com.

PerkinElmer, Inc.
940 Winter Street
Waltham, MA 02451 USA
P: (800) 762-4000 or
(+1) 203-925-4602
www.perkinelmer.com

PerkinElmer®

For a complete listing of our global offices, visit www.perkinelmer.com/ContactUs

009995_01 Printed in USA



StepOne® NEWBORN SCREENING

THE GOLD STANDARD IN NEWBORN SCREENING

Comprehensive Newborn Screening Program

PerkinElmer® *For the Better*

StepOne®
NEWBORN SCREENING

Newborn screening: What you need to know

Within the first few days after a baby is born, he or she is examined by a team of physicians and nurses. Part of the exam includes the collection of blood. A portion of this blood can be used to screen for inherited disorders. While more than 50 inherited disorders are detectable today, the number of disorders tested varies from state to state. The American College of Medical Genetics (ACMG) recommends a core and secondary panel for 54 disorders, which will affect approximately 1 out of 750 babies.

Although rare, inherited disorders are serious and the complications can be devastating. Inherited disorders that go undetected until symptoms develop can cause lifelong complications, including mental retardation, motor impairment and physical disability. Some can lead to death. Newborns that are screened and diagnosed early can receive treatment before symptoms develop. These children can lead normal, healthy lives as a result of early detection.

StepOne® is one of the most comprehensive newborn screening programs available today. You can purchase StepOne online at www.perkinelmergenetics.com or by calling 866.463.6436. The StepOne screening packet contains all of the necessary information and materials to have your baby tested. Take the packet to the hospital when your baby is born and give it to the physician. A healthcare professional will prick your baby's heel before your baby leaves the hospital to collect a blood sample. You or the physician should send it in the prepaid postage envelope to the PerkinElmer Genetics laboratory for processing. The blood sample can also be collected at the first doctor visit. Keep in mind, all babies should receive a blood test during the first few days of life to screen for the disorders that your state requires, but StepOne screens your baby for more than 50 disorders to help ensure a healthy start in life. To learn more about our laboratory operations, see the About Us section of this brochure.

For more information, or to order a screening packet, please call 866.463.6436 or visit www.perkinelmergenetics.com.

- **15 years of experience screening over 4 million babies**
- **More than 50 disorders screened**
- **Genetic counselors on staff**
- **Research scientists on staff**
- **State-of-the-art-technology**
- **Results available to physicians online**
- **Fully accredited laboratory**

Is newborn screening a new procedure?

No. Newborns have been screened for inherited disorders in the U.S. since the early 1960s. Every state currently maintains a screening program, but the number of disorders tested varies from state to state.

What are metabolic and other inherited disorders?

Metabolic and other inherited disorders are flaws in body chemistry. For example, metabolic disorders can cause newborns to have difficulty processing food, placing them at risk for serious health complications such as mental retardation, coma, and even death.

Why would I have my baby tested?

Newborns may not show obvious signs that they have an inherited disorder until after health complications have developed. Early identification can allow your physician to start specialized medical treatment that may improve the long-term health of your baby. While we recommend StepOne® for all newborns, screening can be performed on children of any age.

How many disorders does the StepOne screening include?

StepOne can identify the presence of more than 50 inherited disorders, including Cystic Fibrosis, Maple Syrup Urine Disease, and Congenital Adrenal Hyperplasia as well as many lesser known disorders your state may not include in its screening program.

If my child is over 3 months of age, can he or she be tested for Cystic Fibrosis?

Yes. To order DNA Carrier Testing for Cystic Fibrosis, please call 866.463.6436.

How is my baby tested?

A healthcare professional will draw a small specimen of blood by pricking your baby's heel. The specimen is placed on the absorbent filter paper and sent to PerkinElmer Genetics laboratory to be analyzed.

Can I test my baby myself?

No. Your physician must coordinate the entire process. You should not, under any circumstances, attempt to draw and submit the blood specimen yourself.

When will I know the results?

The testing will be complete approximately three business days after the sample arrives at the PerkinElmer Genetics laboratory. Your child's physician will be notified of any abnormal results by telephone. Results are available to your child's physician by secured Internet access and will be mailed to him or her as well. We are not able to give results directly to parents.

Descriptions of Selected Disorders Included in the StepOne® Newborn Screening Panel

Cystic Fibrosis – Disorder that affects the gastrointestinal tract and the respiratory tract, resulting in poor growth and frequent infections. Treatment typically involves pancreatic enzymes and antibiotics.

Biotinidase Deficiency – Defect in activation of the vitamin biotin, resulting in possible mental retardation or early death. Treatment typically involves vitamin supplementation.

Congenital Adrenal Hyperplasia – Defect in steroid hormone production, possibly resulting in serious illness or early death from loss of body minerals. Treatment typically involves hormone replacement.

Glucose - 6 - Phosphate Dehydrogenase Deficiency (G-6-PD) – Defect in ability to protect red blood cells. Results in anemia or jaundice. Treatment typically involves avoidance of certain common medications and foods.

Maple Syrup Urine Disease (MSUD) – Defect in breakdown of amino acids from proteins. Could result in failure to thrive, neurological complications and death in the newborn. Treatment typically involves restricted protein diet.

Tyrosinemias (Types II, III, and Transient Neonatal) – Defects in the breakdown of an amino acid. Symptoms include liver and kidney failure, poor growth, neurologic problems and may result in early death. Treatment typically involves restricted protein diet.

Homocystinuria – Defect in the breakdown of an amino acid. Symptoms include poor eyesight, mental retardation, and frequent blood clots. Treatment typically involves special diet.

Citrullinemia – Indicated by the accumulation of citrulline. The age of onset is unpredictable but results in poor growth, respiratory failure, and neurological disorders. Treatment typically involves restricted protein diet.

Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency – Defect in the breakdown of fat. Generally presents within the first two years of life with recurrent episodes of low blood sugar after fasting. It is sometimes determined as the cause of previously unexplained infant deaths. Treatment typically involves avoidance of fasting and reduction of dietary fat.

Methylmalonic Acidemia – Defect in the breakdown of organic acids. Symptoms include abnormal blood acid levels and poor growth. Treatment typically involves dietary protein restriction and/or vitamin supplementation.

Propionic Acidemia – Defect in the breakdown of organic acids. Symptoms include not eating, vomiting, seizures, low blood acid, and coma. Treatment typically involves protein restriction and carnitine, glycine or vitamin supplementation.

Isovaleric Acidemia – Defect in the breakdown of organic acids. Symptoms include low blood acid, vomiting, and coma. Treatment typically involves protein restriction and carnitine or glycine supplementation.

The analyses conducted by PerkinElmer Genetics produce results that can be used by qualified physicians in the diagnosis of disorders described herein. Evidence of these conditions will be detected in the vast majority of affected individuals; however, due to genetic variability, age of patient at time of specimen collection, quality of specimen, health status of the patient, and other variables, such conditions may not be detected in all affected patients.