PerkinElmer
Genetics

Test Catalog for Clinicians

StepOne® (Comprehensive with SCID)

Fatty Acid Oxidation Disorders
Carnitine/Acylcarnitine Translocase Deficiency
Carnitine Palmitoyl Transferase Deficiency Type I1
3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency
2,4-Dienoyl-CoA Reductase Deficiency1
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 Disorders
Argininemia
Argininosuccinic Aciduria
5-Oxoprolinuria1
Carbamoyl Phosphate Synthetase Deficiency1
Citrullinemia
Homocystinuria
Hypermethioninemia
Hyperammonemia, Hyperornithinemia, Homocitrullinuria Syndrome1
Hyperornithinemia with Gyral Atrophy1
Maple Syrup Urine Disease
Phenylketonuria
  Classical/Hyperphenylalaninemia
  Biopterin Cofactor Deficiencies
Tyrosinemia
Transient Neonatal Tyrosinemia
  Tyrosinemia Type I2
  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

Endocrine Disorder
Congenital Adrenal Hyperplasia
  Salt Wasting 21-Hydroxylase Deficiency
  Simple Virilizing 21-Hydroxylase Deficiency
Congenital Hypothyroidism

Other Disorder
Biotinidase Deficiency
  Complete Deficiency
  Partial Deficiency
Glucose-6-Phosphate Dehydrogenase Deficiency
Cystic Fibrosis (not valid after 90 days of age)*
Galactosemia
  Galactokinase Deficiency
  Galactose-1-Phosphate Uridyltransferase Deficiency
  Galactose-4-Epimerase Deficiency
Severe Combined Immunodeficiency (SCID)

Hemoglobin Disorder
Sickle Cell & other Hemoglobinopathies
  Hemoglobin S, S/C, S/Beta-Thalassemia, C, & E Diseases

StepOne + SCID + LSDs

StepOne (Comp + SCID)

(All disorders listed above)

Lysosomal Storage Disorders (LSDs)
Fabry
Gaucher
Krabbé Disease
Mucopolysaccharidosis Type I (MPS-I)
Niemann-Pick A/B
Pompe

StepOne + SCID + LSDs + X-ALD (full panel)

StepOne (Comp + SCID)

(All disorders listed above)

Lysosomal Storage Disorders (LSDs)
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X-Linked Adrenoleukodystrophy (X-ALD)

LSDs Panel Only

Lysosomal Storage Disorders (LSDs)
Fabry
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SCID only

X-ALD only

Specialty Testing

Post Mortem Screens

PKU Clinical Monitoring

Second Tier Reflex Testing Menu

PerkinElmer Genetics uses combinations of assays in a multi-tier approach that optimizes detection of abnormal results. Positive DNA identification for many disorders further speeds definitive diagnosis and implementation of critical therapies.

Biochemical Second Tier Testing

Disorder	**Testing Approach**
Congenital Adrenal Hyperplasia	First Tier; 17-OH P Second Tier; Extracted 17-OH P on all elevated.
Congenital Hypothyroidism	First Tier; either T4 or TSH. Second Tier TSH with a primary T4.
Galactosemia	First Tier; Total Galactose plus quantitative Uridyltransferase. Second Tier; Fractionated Galactose.

DNA Second Tier Testing

Disorder	**Mutations Detected**
Galactosemia	N314D (Duarte) Q188R, S135L, K285N, and L195P (Classical)
Hemoglobinopathies	Hb S (173A>T), Hb C (172G>A), Hb E (232G>A), Hb D (121G>C) and Hb O (121G>A) β Thalassaemias -29A>G, -88C>T, and IVS1+6T>C
Cystic Fibrosis	This chart contains the 23 mutations recommended by the ACOG/ACMG:

ΔF508	1717-1G>A	W1282X	2307insA
ΔI507	R560T	N1303K	Y1092X
G542X	R553X	394delTT	M1101K
G85E	G551D	Y122X	S1255X
R117H	1898+1G>A	R347H	3876delA
621+1G>T	2184delA	V520F	3905insT
711+1G>T	2789+5G>A	A559T	5/7/9T
1078delT	3120+1G>A	S549N	F508C
R334W	R1162X	S549R	I507V
R347P	3659delC	1898+5G>T	I506V
A455E	3849+10kbC>T	2183AA>G	

Second Tier Reflex Testing Menu

Disorder	Mutations Detected
Biotinidase Deficiency	G98:d7i3, Q456H, R157H, R538C, D252G and D444H; D444H;A171T, D444H; F403V, D444H;R157H
MCAD	A985A>G, 199T>C
LCHAD	1528G>C
Glutaric Acidemia 1	A421V (Amish) R402W (Caucasian)
Propionic Acidemia	E168K (Spanish) 1218del14/ins12 (Caucasian) 1170insT
Methylmalonic Acidemia	N219Y (Caucasian) G717V (African American)
3-Methylcrotonyl-CoA Carboxylase Def.	518insT (Mennonite)
Maple Syrup Urine Disease	Y438N (previously known as Y393N)
Isovaleric Acidemia	A282V

Clinical Whole Genome

Contact us on 1-866-463-6436 for details

Clinical Exome

Contact us on 1-866-463-6436 for details

NeoSeq (NGS)

Contact us on 1-866-463-6436 for details

LSD NGS panel (single gene or full panel sequencing)

Fabry *GLA* gene sequencing

Gaucher *GBA* gene sequencing

Pompe *GAA* gene sequencing

Krabbe *GALC* gene sequencing

Niemann-Pick A/B *SMPD1* gene sequencing

Hurler Syndrome (MPS-I) *IDUA* gene sequencing

Hunter syndrome (MPS-II) *IDS* gene sequencing

Morquio syndrome type A (MPS IVA) *GALNS* gene sequencing

Beta-1 Galactosidase (Morquio syndrome type B, MPS IVB) *GLB1* gene sequencing

Maroteaux-Lamy (MPS-VI) *ARSB* gene sequencing

Mucopolysaccharidosis VII *GUSB* gene sequencing

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Citrullinemia
Homocystinuria
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X-ALD only

X-Linked Adrenoleukodystrophy (X-ALD)

Disclaimer
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 the patient at the time of specimen collection, quality of the specimen, health status of the patient, and other variables, such conditions may not be detected in all affected patients.

Pursuant to applicable federal and/or state laboratory requirements, PerkinElmer® Genetics has established and verified the accuracy and precision of its laboratory developed testing services. Tests are developed and performance characteristics are determined by PerkinElmer Genetics.

How to order?

Ordering - Clinicians

In this section, you can learn more about our testing services:

Step-1: Select the list of disorders from list above

Step-2: Download our test requisition form

Step-3: Call us on 1-866-463-6436 to obtain sample collection material

More about Newborn Screening services

Testing Services
PerkinElmer Genetics offers several comprehensive newborn screening panels as well as Specialty Testing, Post Mortem and DNA Carrier Screening.

PerkinElmer Genetics has made numerous innovations resulting from the efforts of the laboratory's dedicated professionals which include: the advent of tandem mass spectrometry; the use of DNA technologies as both primary and secondary means of disease detection; the advent of unique testing algorithms to improve the accuracy of newborn screening; and the development and implementation of screening assays for Severe Combined Immunodeficiency (SCID) and Lysosomal Storage Disorders (LSD).

New Service Offering
PerkinElmer Genetics is positioned to offer three new disorders that were added to the Recommended Uniform Screening Panel (RUSP). Pompe disease was added in March 2015 followed by Mucopolysaccharidosis Type 1 (MPS-1 or Hurler Syndrome) and X-linked Adrenoleukodystrophy (X-ALD) in February 2016.

Severe Combined Immunodeficiency (SCID)
Severe Combined Immunodeficiency (SCID) is a group of disorders characterized by the absence of both humoral and cellular immunity. The defining characteristic for SCID is always a severe defect in T-cell production and function, with defects in B-lymphocytes as a primary or secondary problem and, in some genetic types, in natural killer (NK) cell production as well.

SCID screening is available as stand-alone testing or included in our StepOne Comprehensive + SCID panel.

Lysosomal Storage Disorders (LSDs) (click to download sell sheet)
PerkinElmer Genetics offers screening for six (6) Lysosomal Storage Disorders (LSD); Fabry, Gaucher, Krabbe Disease, Mucopolysaccharidosis Type I (MPS-1), Niemann-Pick A/B and Pompe. Lysosomal storage disorders develop as a result of an enzyme deficiency or malfunction that causes cell waste to build up within the cell instead of being excreted.

LSD screening is available as stand-alone testing or included in our StepOne Comprehensive + SCID + LSD panel.

X-Linked Adrenoleukodystrophy (X-ALD) (click to download sell sheet)
X-ALD is a serious progressive genetic disorder caused by an abnormality in the ABCD1 gene on the X chromosome and affects roughly one out of every 17,000 to 20,000 births.

X-ALD screening is available as stand-alone testing or included in our StepOne Comprehensive +SCID + LSD + X-ALD panel.

If you have questions on our testing services please contact Client Services.

Normal Business Hours
Client Services Department
Monday thru Friday 8 AM Eastern to 5 PM Eastern
Phone: 1-412-220-2300
Toll Free: 1-866-463-6436
Fax: 1-412-220-0784
Contact us via web form

How to order?

Results

Results
PerkinElmer Genetics' laboratory works collaboratively with clinicians providing accurate and timely test results necessary for quality care of patients. PerkinElmer Genetics receives samples 6 days a week – Monday through Saturday (excluding courier holidays). Testing occurs 7 days per week. The Genetic Counseling Staff is available 24 hours per day, 7 days per week to facilitate short-term follow-up, communication and education to health care providers worldwide.

Click here to visit Results center

To obtain an ID to download results from our secure portal, please contact Client Services to obtain the appropriate form.

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