Test Catalog for Clinicians

Newborn Screening ................................................................. 2
  Biochemical Genetic Testing ............................................. 2
  Molecular Genetic Testing ................................................. 4

Genetic Testing ........................................................................ 5
  Molecular Genetic Testing ................................................... 5
  Biochemical Genetic Testing .............................................. 7

How to order? ........................................................................... 8
### Fatty Acid Oxidation Disorders
- Carnitine/Acylcarnitine Translocase Deficiency
- Carnitine Palmitoyl Transferase Deficiency Type I
- 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency
- 3-Octanoyl-CoA Dehydrogenase Deficiency
- 2,4-Dienoyl-CoA Reductase Deficiency
- 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
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- Glutaric Acidemia Type I
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### Amino Acid Disorders
- Arginemia
- Argininosuccinic Aciduria
- 5-Oxoprolinuria
- Carbamoyl Phosphate Synthetase Deficiency
- Citrullinemia
- Homocystinuria
- Hypermethioninemia
- Hyperammonemia, Hyperornithinemia, Homocitrullinuria Syndrome
- Hyperornithinemia with Gyral Atrophy
- Maple Syrup Urine Disease
- Phenylketonuria
- Classical/Hyperphenylalaninemia
- Bioprotein Cofactor Deficiencies
- Tyrosinemia
- Transient Neonatal Tyrosinemia
- Tyrosinemia Type I
- 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

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### StepOne + SCID + LSDs

#### StepOne (Comp + SCID)
(All disorders listed above)

#### Lysosomal Storage Disorders (LSDs)

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Details</th>
</tr>
</thead>
<tbody>
<tr>
<td>Fabry</td>
<td></td>
</tr>
<tr>
<td>Gaucher</td>
<td></td>
</tr>
<tr>
<td>Krabbe Disease</td>
<td></td>
</tr>
<tr>
<td>Mucopolysaccharidosis Type I (MPS-I)</td>
<td></td>
</tr>
<tr>
<td>Niemann-Pick A/B</td>
<td></td>
</tr>
<tr>
<td>Pompe</td>
<td></td>
</tr>
</tbody>
</table>

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

#### StepOne (Comp + SCID)
(All disorders listed above)

#### Lysosomal Storage Disorders (LSDs)

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<tr>
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<td></td>
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<tr>
<td>Niemann-Pick A/B</td>
<td></td>
</tr>
<tr>
<td>Pompe</td>
<td></td>
</tr>
</tbody>
</table>

#### X-Linked Adrenoleukodystrophy (X-ALD)

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Newborn Screening > Biochemical Genetic Testing

LSDs Panel Only

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

SCID only

X-ALD only

Specialty Testing

Post Mortem Screens
PKU Clinical Monitoring
Newborn Screening > Molecular Genetic Testing

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

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Testing Approach</th>
</tr>
</thead>
<tbody>
<tr>
<td>Congenital Adrenal Hyperplasia</td>
<td>First Tier; 17-OH P Second Tier; Extracted 17-OH P on all elevated.</td>
</tr>
<tr>
<td>Congenital Hypothyroidism</td>
<td>First Tier; either T4 or TSH. Second Tier TSH with a primary T4.</td>
</tr>
<tr>
<td>Galactosemia</td>
<td>First Tier; Total Galactose plus quantitative Uridyltransferase. Second Tier; Fractionated Galactose.</td>
</tr>
</tbody>
</table>

DNA Second Tier Testing

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Mutations Detected</th>
</tr>
</thead>
<tbody>
<tr>
<td>Galactosemia</td>
<td>N314D (Duarte)</td>
</tr>
<tr>
<td></td>
<td>Q188K, S135L, K285N, and L195P (Classical)</td>
</tr>
<tr>
<td>Hemoglobinopathies</td>
<td>Hb S (173A&gt;T), Hb C (172G&gt;A), Hb E (232G&gt;A), Hb D (121G&gt;C) and Hb O (121G&gt;A)</td>
</tr>
<tr>
<td></td>
<td>β Thalassaemias -29A&gt;G, -88C&gt;T, and IVS1+6T&gt;C</td>
</tr>
<tr>
<td>Cystic Fibrosis</td>
<td>This chart contains the 23 mutations recommended by the ACOG/ACMG:</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Mutations</th>
<th>ACOG/ACMG</th>
</tr>
</thead>
<tbody>
<tr>
<td>ΔF508</td>
<td>1717-1G&gt;A</td>
</tr>
<tr>
<td>ΔI507</td>
<td>R560T</td>
</tr>
<tr>
<td></td>
<td>N1303K</td>
</tr>
<tr>
<td></td>
<td>Y1092X</td>
</tr>
<tr>
<td>G542X</td>
<td>R553X</td>
</tr>
<tr>
<td></td>
<td>394delTT</td>
</tr>
<tr>
<td></td>
<td>M1101K</td>
</tr>
<tr>
<td>G85E</td>
<td>G551D</td>
</tr>
<tr>
<td></td>
<td>Y122X</td>
</tr>
<tr>
<td></td>
<td>S1255X</td>
</tr>
<tr>
<td>R117H</td>
<td>1898+1G&gt;A</td>
</tr>
<tr>
<td></td>
<td>R347H</td>
</tr>
<tr>
<td></td>
<td>3876delA</td>
</tr>
<tr>
<td>621+1G&gt;T</td>
<td>2184delA</td>
</tr>
<tr>
<td></td>
<td>V520F</td>
</tr>
<tr>
<td></td>
<td>3905insT</td>
</tr>
<tr>
<td>711+1G&gt;T</td>
<td>2789+5G&gt;A</td>
</tr>
<tr>
<td></td>
<td>A559T</td>
</tr>
<tr>
<td></td>
<td>5/7/9T</td>
</tr>
<tr>
<td>1078delT</td>
<td>3120+1G&gt;A</td>
</tr>
<tr>
<td></td>
<td>S549N</td>
</tr>
<tr>
<td></td>
<td>F508C</td>
</tr>
<tr>
<td>R334W</td>
<td>R1162X</td>
</tr>
<tr>
<td></td>
<td>S549R</td>
</tr>
<tr>
<td></td>
<td>I507V</td>
</tr>
<tr>
<td>R347P</td>
<td>3659delC</td>
</tr>
<tr>
<td></td>
<td>1898+5G&gt;T</td>
</tr>
<tr>
<td></td>
<td>I506V</td>
</tr>
<tr>
<td>A455E</td>
<td>3849+10kbC&gt;T</td>
</tr>
<tr>
<td></td>
<td>2183AA&gt;G</td>
</tr>
</tbody>
</table>
Second Tier Reflex Testing Menu

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

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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.
Genetic Testing > Molecular Genetic Testing

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)

<table>
<thead>
<tr>
<th>Test Description</th>
<th>Gene Sequencing</th>
</tr>
</thead>
<tbody>
<tr>
<td>Fabry GLA gene sequencing</td>
<td>Morquio syndrome type A (MPS IVA) GALNS gene sequencing</td>
</tr>
<tr>
<td>Gaucher GBA gene sequencing</td>
<td>Beta-1 Galactosidase (Morquio syndrome type B, MPS IVB) GLB1 gene sequencing</td>
</tr>
<tr>
<td>Pompe GAA gene sequencing</td>
<td>Maroteaux-Lamy (MPS-VI) ARSB gene sequencing</td>
</tr>
<tr>
<td>Krabbe GALC gene sequencing</td>
<td>Mucopolysaccharidosis VII GUSB gene sequencing</td>
</tr>
<tr>
<td>Niemann-Pick A/B SMPD1 gene sequencing</td>
<td></td>
</tr>
<tr>
<td>Hurler Syndrome (MPS-I) IDUA gene sequencing</td>
<td></td>
</tr>
<tr>
<td>Hunter syndrome (MPS-II) IDS gene sequencing</td>
<td></td>
</tr>
</tbody>
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# Genetic Testing

## Biochemical Genetic Testing

### StepOne® (Comp + SCID)

#### Fatty Acid Oxidation Disorders
- Carnitine/ Acylcarnitine Translocase Deficiency
- Carnitine Palmitoyl Transferase Deficiency Type I
- 3-Hydroxy-Long Chain Acyl-CoA Dehydrogenase Deficiency
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- Short Chain Acyl-CoA Dehydrogenase Deficiency
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#### Organic Acid Disorders
- 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency
- Glutaric Acidemia Type I
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- 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
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- Hyperalimentation
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- 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
  - Simple Virilizing
- Congenital Hypothyroidism

### X-ALD only

#### X-Linked Adrenoleukodystrophy (X-A LD)

#### Other Disorder
- Biotinidase Deficiency
  - Complete Deficiency
  - Partial Deficiency
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- Cystic Fibrosis (not valid after 90 days of age)*
- Galactosemia
  - Galactokinase Deficiency
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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
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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