Test Catalog for Patients and Parents

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Newborn Screening > Biochemical Genetic Testing

StepOne® (Comprehensive with SCID)

**Fatty Acid Oxidation Disorders**
- Carnitine/Acylcarnitine Translocase Deficiency
- Carnitine Palmitoyl Transferase Deficiency Type I1
- 3-Hydroxy-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**
- 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 Acidemia
- 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
- S-0xoprolinuria1
- Carbamoyl Phosphate Synthetase Deficiency
- Citrullinemia
- Homocystinuria
- Hypermethioninemia
- Hyperammononemia, Hyperornithinemia, Homocitrullinuria Syndrome1
- Hyperornithinemia with Gyral Atrophy1
- Maple Syrup Urine Disease
- Phenylketonuria
- Classical/Hyperphenylalaninemia
- Bioprotein 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

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**StepOne + SCID + LSDs**

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

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

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

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

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

**X-Linked Adrenoleukodystrophy (X-ALD)**
LSDs Panel Only

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

StepOne® SCID only

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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.
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.

## Second Tier Reflex Testing Menu

**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>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>ΔF508</th>
<th>1717-1G&gt;A</th>
<th>W1282X</th>
<th>2307insA</th>
</tr>
</thead>
<tbody>
<tr>
<td>ΔI507</td>
<td>R560T</td>
<td>N1303K</td>
<td>Y1092X</td>
</tr>
<tr>
<td>G542X</td>
<td>R553X</td>
<td>394delTT</td>
<td>M1101K</td>
</tr>
<tr>
<td>G85E</td>
<td>G551D</td>
<td>Y122X</td>
<td>S1255X</td>
</tr>
<tr>
<td>R117H</td>
<td>1898+1G&gt;A</td>
<td>R347H</td>
<td>3876delA</td>
</tr>
<tr>
<td>621+1G&gt;T</td>
<td>2184delA</td>
<td>V520F</td>
<td>3905insT</td>
</tr>
<tr>
<td>711+1G&gt;T</td>
<td>2789+5G&gt;A</td>
<td>A559T</td>
<td>5/7/9T</td>
</tr>
<tr>
<td>1078delT</td>
<td>3120+1G&gt;A</td>
<td>S549N</td>
<td>F508C</td>
</tr>
<tr>
<td>R334W</td>
<td>R1162X</td>
<td>S549R</td>
<td>I507V</td>
</tr>
<tr>
<td>R347P</td>
<td>3659delC</td>
<td>1898+5G&gt;T</td>
<td>I506V</td>
</tr>
<tr>
<td>A455E</td>
<td>3849+10kbC&gt;T</td>
<td>2183AA&gt;G</td>
<td></td>
</tr>
</tbody>
</table>
## Newborn Screening > Molecular Genetic Testing

### 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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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>Fabry GLA gene sequencing</th>
<th>Morquio syndrome type A (MPS IVA) GALNS gene sequencing</th>
</tr>
</thead>
<tbody>
<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>Maroteaux-Lamy (MPS-VI) ARSB gene sequencing</td>
</tr>
<tr>
<td>Niemann-Pick A/B SMPD1 gene sequencing</td>
<td>Maroteaux-Lamy (MPS-VI) ARSB gene sequencing</td>
</tr>
<tr>
<td>Hurler Syndrome (MPS-I) IDUA gene sequencing</td>
<td>Mucopolysaccharidosis VII GUSB gene sequencing</td>
</tr>
<tr>
<td>Hunter syndrome (MPS-II) IDS gene sequencing</td>
<td>Mucopolysaccharidosis VII GUSB gene sequencing</td>
</tr>
</tbody>
</table>

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Pompe

SCID only

X-ALD only

X-Linked Adrenoleukodystrophy (X-ALD)
Genetic Counseling

All Tests

Contact us for details

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How to order?

Ordering – Patients and Parents

Here’s the process

**Step-1:** Newborn screening tests - Order StepOne®
For Genetic Testing Services and all other services, please reach out to us on 1-866-463-6436

Read more about the Newborn screening services order process below

**Ensure a healthy start in life with StepOne®**

**Prevention**
StepOne® Newborn Screening can detect more than **50 disorders** in newborns from just a few drops of blood. This screening gives parents the opportunity to protect your baby from the preventable complication of undiagnosed disorders. Many of these disorders are manageable if treatment begins early.
When it comes to the health of your child, any information is valuable information. StepOne® determines the risk your baby is at for a metabolic or other inherited disorder. An abnormal result indicates the need for diagnostic testing to confirm the presence of a disorder. If your baby is diagnosed with one of these disorders, early medical intervention can play a key role in helping your child to lead a normal life and StepOne® is the first step in this process.

**What is the process?**

1. Our StepOne® parent packets may be ordered through this web site at [Order StepOne®](#) or by calling 866-463-6436;
2. The packet will be shipped directly to you and will contain all of the information and materials required to have your newborn screened;
3. Screening your baby is simple, but it must be coordinated by a physician.
4. Take your StepOne® packet with you to the hospital at the time of delivery. A physician letter and the filter paper for the blood specimen collection will have instructions on the back side on how to coordinate, handle and submit the filter paper for analysis;
5. Specimens should be drawn between 24 and 48 hours after birth. Please ensure all demographic fields are completed; A healthcare professional will take a small sample of blood by pricking the baby’s heel and place it on the absorbent filter paper located in the packet;
6. After the sample dries, the healthcare provider will return it to you the parent for shipment.
7. For your convenience, a prepaid UPS envelope is provided in the packet. Please return the filter paper with the blood spots and the Physicians Form for Results to our lab where professionals use different screening methods and equipment to analyze the sample.
8. The results are given directly to your baby’s physician you provided on the Form for Results.

If your baby’s physician denies your request for screening, you will be provided with a full refund of payment. Always seek the advice of your baby’s physician if you have questions or before you stop, start, or change any treatment plan for your baby. PerkinElmer Genetics services and materials are not as substitute for medical advice, diagnosis, or treatment.

**Key points**

1. While more than 50 inherited disorders are detectable today, the number of disorders tested varies from state to state by your Department of Health;
2. The American College of Medical Genetics and Genomics has proposed a Recommended Uniform Screening Panel (RUSP) for 58 disorders, which will affect approximately 1 out of 750 babies;
3. PerkinElmer Genetics has 20 years of experience screening over 6.5 million babies;
4. Genetic Counselors and Research Scientists on staff;
5. State-of-the-art-technology;
6. Turn-around Time - 72 hours from receipt of specimen;
7. Results available to physicians on a secure Results Portal;
8. Fully accredited laboratory.

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Results

Contact your provider