PerkinElmer
Genetics

Test Catalog for Pharma

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 3 months 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
Krabbe Disease
Mucopolysaccharidosis Type I (MPS-I)
Niemann-Pick A/B
Pompe

StepOne + LSDs + X-ALD (full panel)

StepOne (Comp + SCID)

(All disorders listed above)

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

DMD

LSDs Panel Only

Lysosomal Storage Disorders (LSDs)
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StepOne® SCID only

Specialty Testing

Post Mortem Screens

PKU Clinical Monitoring

Disclaimer
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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.

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

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NeoSeq (NGS)

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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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How to order?

Ordering and Results

Please contact PerkinElmer representative.

Results

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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