Department of Pathology and Laboratory Medicine at Children's Hospital of Colorado

Lab Client Services:
LabClientServices@childrenscolorado.org

Test Directory:
https://childrenscolorado.testcatalog.org/

Biochemical Genetics Laboratory

Scientific Director: Tim Wood, PhD FACMG, Timothy.Wood@childrenscolorado.org

Test/Assay	Condition or Indication	What the Biomarker Reflects
VLCAD Enzyme Assay	Positive NBS for VLCAD deficiency/Family history of VLCAD deficiency	Accumulation patterns indicating impaired very-long-chain fatty acid β-oxidation
3-Analyte MMA Group	Positive NBS for elevated C3/Treatment monitoring for Propionic or methylmalonic acidemia	**MMA:** Propionate/cobalamin dysfunction **3OH-Propionic Acid:** Propionic acidemia marker **Methylcitric Acid:** Propionyl-CoA shunting due to pathway block
Trimethylamine (TMA)	Concern for trimethylaminuria / Fishy odor	Inability to convert TMA → TMAO, causing odor-producing metabolite buildup

Mitochondrial Laboratory

Scientific Director: Johan Van Hove, MD, Johan.VanHove@childrenscolorado.org
Associate Professor: Marisa W. Friederich, PhD, Marisa.Friederich@childrenscolorado.org

Test/Assay	Purpose	What it Detects
Blue Native Electrophoresis (BN-PAGE)	Separates and visualizes intact mitochondrial respiratory chain enzyme complexes	Defects in activity and/or assembly of oxidative phosphorylation (OXPHOS) complexes
Pyruvate Dehydrogenase (PDH) Enzyme Activity	Measures activity of the PDH complex responsible for converting pyruvate to acetyl-CoA	PDH deficiency, causes of lactic acidosis, mitochondrial energy metabolism disorders
GDF-15 + FGF-21 Biomarker Panel	Combined serum biomarkers that rise in response to mitochondrial dysfunction	Sensitive indicators of mitochondrial disease, including mitochondrial myopathies and systemic OXPHOS defects; useful when enzyme or genetic testing is inconclusive
Complex V ATP Hydrolysis + Mitochondrial Respiratory Chain Enzyme Panel (Complex I–V)	Comprehensive evaluation of the enzymatic activity across all mitochondrial respiratory chain complexes	Identifies ATP synthase–related defects and isolated or combined oxidative phosphorylation abnormalities; supports diagnosis of mitochondrial disorders impacting cellular energy production

The assays listed above showcase just a portion of our specialized metabolic test offerings. Please visit our test catalog for a full list, requisitions, and test codes.

COMING IN 2026!

Pyridoxine Dependent Epilepsy (PDE) biomarkers – 2OPP and 6OP quantitation via LCMSMS

Stay up to date by **signing up for our newsletter**!

Children's Hospital Colorado

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