Dried Blood Spot (DBS) Testing

About Emory Genetics Laboratory (EGL)
EGL specializes in genetic diagnostic testing, with 45 years of clinical experience and board-certified laboratory directors and genetic counselors reporting out cases. EGL offers a combined 1100 molecular genetics, biochemical genetics, and cytogenetics tests under one roof and custom testing for all medically relevant genes, for domestic and international clients.

Equally important to improving patient care through quality genetic testing is the contribution EGL makes back to the scientific and medical communities. EGL is one of only a few clinical diagnostic laboratories to openly share data with the NCBI freely available public database ClinVar (>7000 variants on >500 genes) and is also the only laboratory with a free online database (EmVClass), featuring a variant classification search and report request interface, which facilitates rapid interactive curation and reporting of variants.

Benefits of DBS Testing
DBS testing has numerous benefits, including:

- Ideal for NICU and low-birth weight babies, disease monitoring, and newborn screening (NBS) confirmatory testing
- Advantageous for neonates with limited blood volume
- Cost-effective alternative to ordering repeat screens and other tests that require multiple blood draws
- Increases specificity of the NBS result and reduces false positives
- Convenient for patients and families to use as dietary compliance monitoring

DBS for Disease Monitoring
DBS is a simple and convenient method for monitoring phenylalanine and tyrosine concentrations, which help determine dietary compliance in patients with a diagnosis of phenylketonuria. DBS is also appropriate for monitoring allo-isoleucine and branched chain amino acid (leucine, isoleucine, and valine) concentrations to determine dietary compliance in patients with a diagnosis of maple syrup urine disease.

DBS Diagnostic Screening
Congenital Adrenal Hyperplasia (CAH): Confirmatory Testing for Positive NBS
CAH confirmatory testing is recommended as a second tier test to rule out CAH in neonates with elevated cortisol on NBS. It involves rapid, accurate, quantification of 17-hydroxyprogesterone (17-OHP), androstenedione, cortisol, 11-deoxycortisol, and 21-deoxycortisol.

Methylmalonic Acidemia, Propionic Acidemia or Homocystinemia: Confirmatory Testing for Elevated C3 Carnitine or Methionine on NBS
Confirmatory testing is recommended to determine the cause of elevated C3 carnitine or methionine on NBS. One test quantifies methylmalonic acid and methylenecitrinic acid, while the other test quantifies total homocysteine.

These two tests are used to rule in or out possible differential diagnoses: methylmalonic acidemia due to methylmalonyl-CoA mutase deficiency or CbiA/CbiB deficiencies; combined methylmalonic acidemia and homocystinemia due to deficient CbiC, CbiD, or CbiF, or impaired vitamin B12 absorption or malnutrition; and propionic acidemia.

Lysosomal Storage Disorders: Confirmatory Testing for Fabry, Krabbe, and Pompe Disease
Confirmatory testing is recommended to affirm a diagnosis or positive NBS for Fabry, Krabbe, and Pompe disease. Each test is specific to each disease (α-galactosidase activity for Fabry disease; galactocerebrosidase activity for Krabbe disease; and α-glucosidase activity for Pompe disease) and may be utilized to monitor enzyme replacement therapy.
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Why choose DBS testing at EGL?
- DBS testing allows for easier sample collection from infants
- DBS samples are easier to store and ship
- DBS testing is convenient for self-collection by metabolic patients needing dietary monitoring

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<th>Test Code</th>
<th>Test Name</th>
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**CPT® is a registered trademark of the American Medical Association.

For more information about EGL and the >900 tests we offer:
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