EGL Genetics has nearly 50 years of genetic testing history built upon a strong academic foundation. Our expertise spans common and rare genetic disease testing, genomic variant interpretation, test development and research. As we have grown, we have evolved into a high-science and high-performing CLIA-certified and CAP-accredited laboratory with over 1,100 test offerings across biochemical genetics, cytogenetics, and molecular genetic testing.

NEWBORN SCREENING OFFERINGS

Newborn screening (NBS) is a public health program designed to identify infants with treatable conditions, before they become clinically evident. There are two main laboratory components to NBS – the screening assay programs and confirmatory testing. EGL Genetics has expertise and testing options for both components to meet your needs.

Screening Assays:
- Experience working with public health laboratories
- Able to utilize the same dried blood spot sample collected for standard NBS
- Flexibility and experience bringing up new assays as they are added to state screening panels, including liquid chromatography assays for targeted second tier testing
- High capacity and competitive service

Service Offering:
- MS/MS screening for 6 lysosomal storage disorders [Pompe, MPSI, Krabbe, Gaucher, Fabry, Niemann-Pick A/B]
  - Two tier screening approaches available
  - Post-analytical tools for interpretation minimize false positive screens
- MS/MS screening for X-linked adrenoleukodystrophy
  - Second tier testing by LC-MS/MS
  - Post-analytical tools for interpretation minimize false positive screens

Confirmatory Testing:
- Experience working closely with state NBS programs to confirm or rule out disorders suggested by screening
- Expedited testing available when clinically indicated
- ABMGG boarded laboratory directors available for consultation about abnormal results or testing algorithms
- Molecular testing for most NBS disorders can also be performed at EGL Genetics; reflex testing options available

Service Offering:
- Diagnostic profiles:
  - Amino Acids
  - Plasma Acylcarnitines
  - Urine organic acids
  - Carnitine profile
- Enzyme assays:
  - Classic galactosemia
  - Biotinidase deficiency
  - Pompe disease
  - Mucopolysaccharidosis Type I
- If a diagnosis is confirmed, EGL Genetics is able to provide many options for monitoring of dietary compliance
EXPERIENCE MATTERS

Nearly 50 years of comprehensive clinical genetic testing by board-certified ABMGG laboratory directors reporting over 675,000 results—because experience matters. One of the first laboratories in the genetics industry with exome sequencing, gene targeted array CGH, constitutional microarrays, congenital disorders of glycosylation, and rare disease testing.

ORDER THE RIGHT GENETIC TESTS FOR YOUR PATIENTS

EGL Genetics can customize NBS options according to the requirements of your program. We can offer complete first and second tier options to add new conditions to your screening program while minimizing false positives. We also have the expertise and capabilities to offer targeted second tier testing designed to support the first tier options selected by your program.

To find additional test offerings, please visit our website (http://www.egl-eurofins.com) to search our comprehensive test menu. Laboratory directors and genetics counselors are available to discuss testing options:

Phone: (470) 378-2200
Email: eglgc@egl-eurofins.com

CONTRIBUTIONS TO THE GENETICS COMMUNITY

EGL’s service to the newborn screening and genetics communities extends beyond our testing menu. Our team has a scientific history focused on performance improvement and high quality results. We are engaged with the NBS community at the state and federal level.

WE HAVE THE ANSWERS.

Consolidate your testing needs with our industry-leading expertise in molecular genetics, cytogenetics, and biochemical genetic testing with a focus in rare disease testing.