Genetic Testing:

A Payer's Guide to Partnering with EGL

EMORY UNIVERSITY | Emory Genetics Laboratory

About Emory Genetics Laboratory

Emory Genetics Laboratory (EGL) is a CLIA-certified and CAP-accredited laboratory located in Atlanta, GA. EGL was incorporated into Emory University's Department of Human Genetics in 2002. The department's chair, Dr. Stephen Warren, PhD, FACMG, discovered the fragile X syndrome gene, *FMR1*. Fragile X is one of the leading causes of intellectual disability in the United States and has a prevalence of 1 in 4000 to 1 in 6000 in the general population.

EGL was built upon a solid foundation of research, academia, and cutting-edge technologies; all of which quickly established a foothold for EGL in molecular, biochemical, and cytogenetic testing. EGL formed long-term relationships with clinicians who came to rely on EGL for its expertise and quality of testing and reporting.

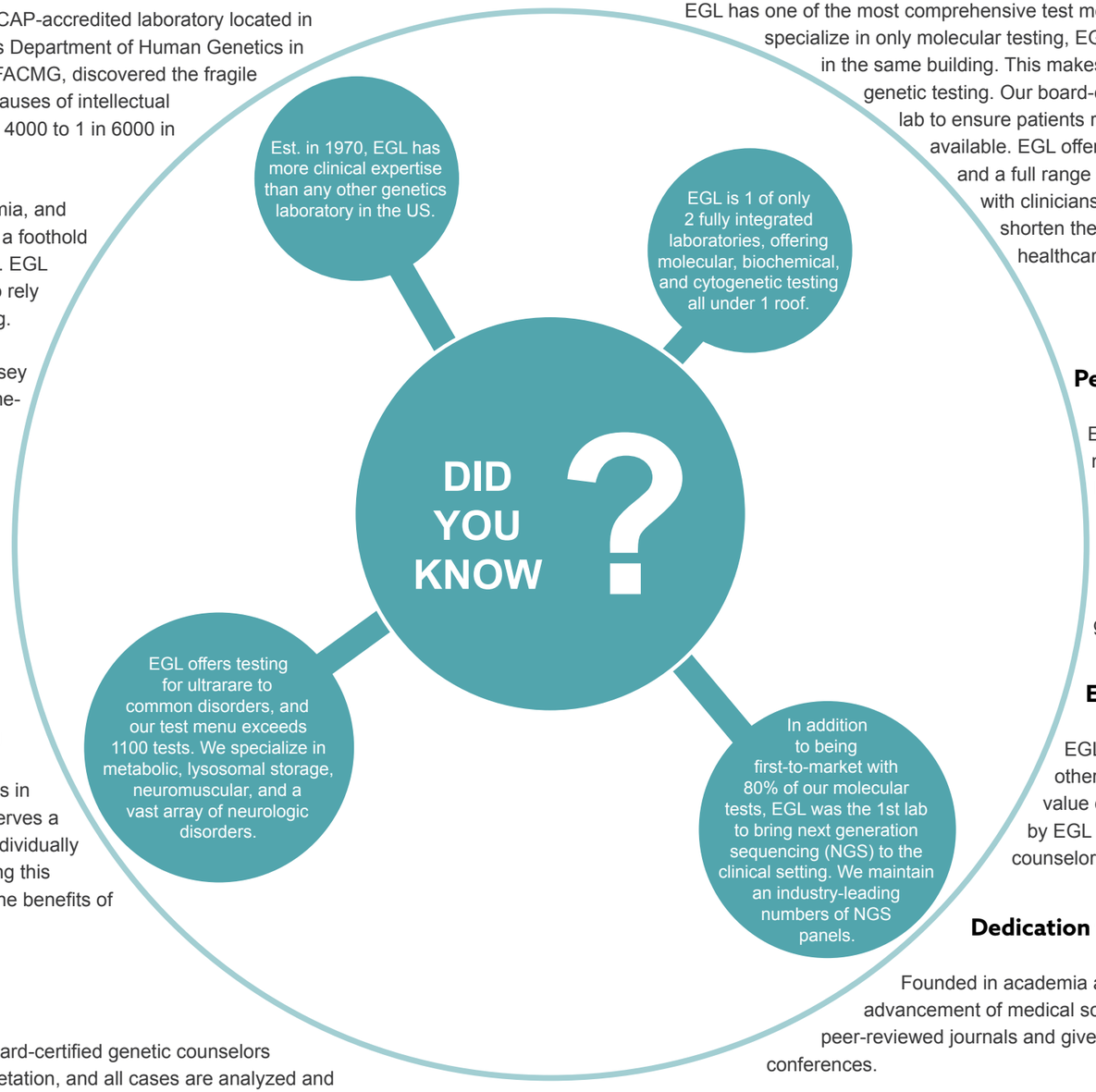
Multi-gene, NGS panels often shorten the diagnostic odyssey for the confirmation of many disorders that is otherwise time-consuming, expensive, and may not lead to confirmation of a clinical diagnosis. EGL was also one of the first laboratories to launch clinical exome sequencing and its Medical EmExome, including the EmExome Boost option and complementary exome deletion and duplication array, provides for the most comprehensive exome testing on the market.

Partnering with EGL

EGL has more than four decades of experience partnering with hospitals, clinicians, and private and public payers to provide comprehensive, cost-effective solutions for patients in need of appropriate and necessary genetic testing. EGL serves a patient population affected by genetic disorders that are individually rare, but collectively common. EGL's commitment to serving this patient population remains the same, as do the following the benefits of partnering with us:

Expertise

ABMGG board-certified laboratory directors and NSGC board-certified genetic counselors are available to assist with test selection and result interpretation, and all cases are analyzed and reported out by board-certified laboratory directors. For more information on EGL laboratory directors, please see page 8 of this brochure.



Cost-effective Testing Options

EGL has one of the most comprehensive test menus on the market. While most genetics laboratories specialize in only molecular testing, EGL offers molecular, biochemical, and cytogenetic testing in the same building. This makes EGL one of only two laboratories offering full-service genetic testing. Our board-certified laboratory directors frequently collaborate cross-lab to ensure patients receive the most streamlined, comprehensive testing available. EGL offers single-gene and multi-gene panels, exome sequencing, and a full range of biochemical and cytogenetic testing. We work closely with clinicians to identify the right test(s) for their patients that will shorten the diagnostic odyssey and lessen the burden on the healthcare system.

Personal Service

EGL reviews test requisitions for appropriateness and necessity based on the information clinicians provide. Board-certified genetic counselors will contact clinicians regarding testing that is ordered out of the normal sequence, testing ordered that doesn't match the clinical information provided, and testing ordered that may not be the best option for confirming a clinical diagnosis of a genetic disorder.

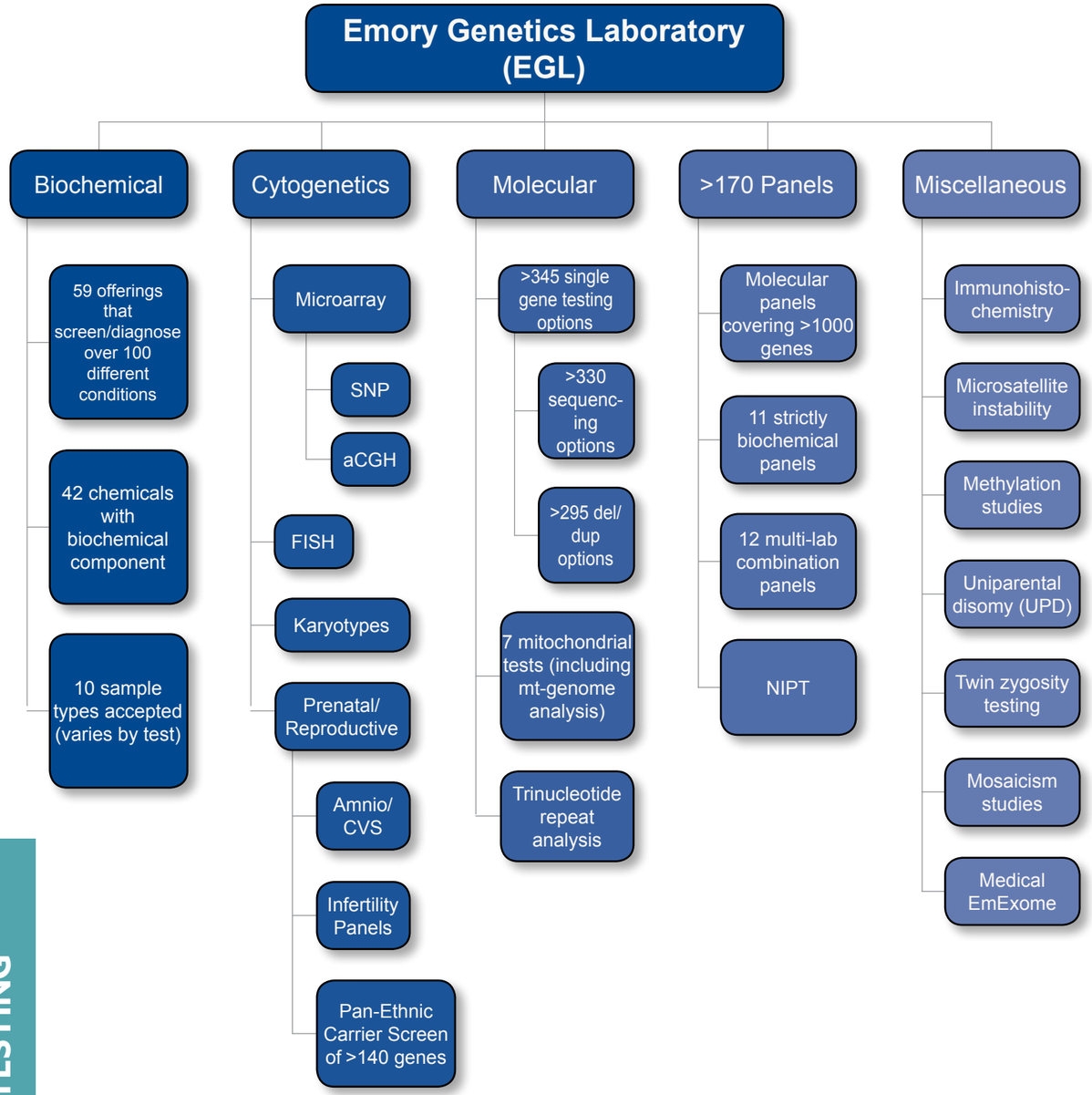
Education and Resources

EGL is committed to providing clinical guides, webinars, and other resources that help educate clinicians and payers on the value of appropriate genetic testing. All materials are developed by EGL board-certified laboratory directors and genetic counselors, who are available to answer questions.

Dedication to Research

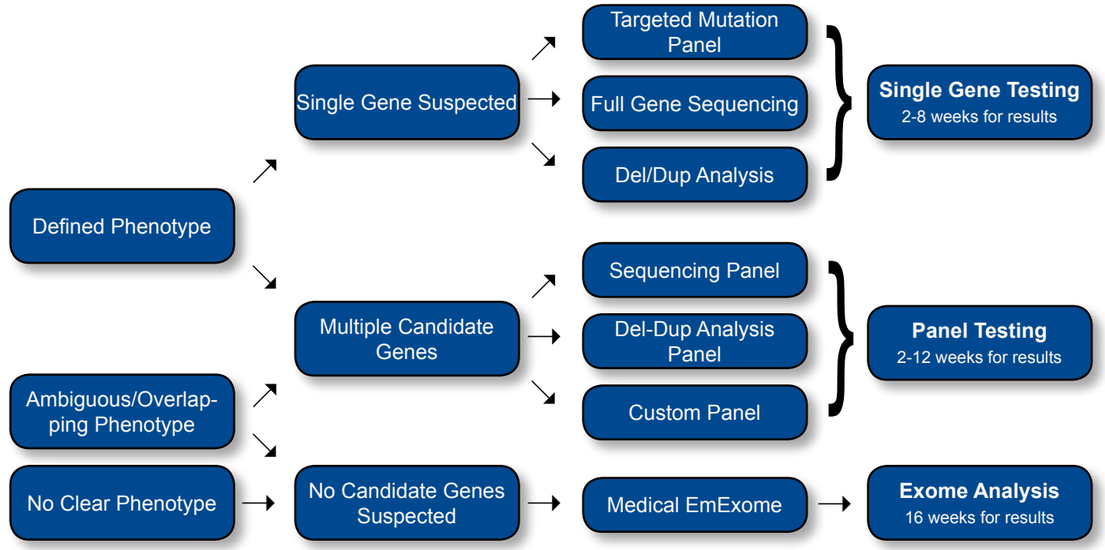
Founded in academia and research, EGL is strongly committed to the advancement of medical science. Laboratory directors frequently publish in peer-reviewed journals and give presentations at regional and national educational conferences.

Testing Overview



Making Sense of the Different Testing Options

There are many testing options to choose from when trying to identify or confirm a genetic condition. The ideal test type for your patient will be determined by various factors including: clinical findings, cost, turnaround time, and why testing is being performed.



Targeted Mutation Analysis vs. Gene Sequencing vs. Deletion/Duplication Analysis

- Targeted mutation analysis – This option only analyzes a specific set of common mutations within a gene. Reasons for choosing this option include:
  - A family history of a specific mutation
  - Looking for most common mutations in certain ethnicities
  - Carrier screening

- Sequencing – This option examines gene(s) from beginning to end for changes and is the most commonly ordered first-tier test. There are certain technological limitations and complexities that prevent sequencing from identifying 100% of mutations within a gene, but it is usually the most comprehensive gene analysis that can be ordered.

- Deletion and duplication array analysis –This is most commonly used as a second-tier testing option, when no mutations (or only 1 mutation in regard to autosomal recessive conditions) have been identified on sequencing. This option looks for large deletions and duplications that may be missed in routine sequencing to allow for a more complete gene analysis.

TESTING

Single-Gene vs. Multi-Gene Panels vs. Exome

Single-gene tests are best used with a more defined phenotype that corresponds to 1 condition/gene. These tests help identify causative mutations and confirm the suspected diagnosis.

Example: Ordering beta-hemoglobin gene (HBB) analysis for someone who has clinical features with complete blood count or hemoglobin electrophoresis results consistent with beta-thalassemia and confirmation of causative mutations is desired.

In contrast, multi-gene panels are used to help narrow down a diagnosis in a more cost-effective and timely manner than testing one gene after another sequentially.

Example: Ordering a panel for congenital disorders of glycosylation (including 66 genes) on a patient with suspected clinical features of this type of disorder. Since the phenotypes can overlap, it is more cost effective to analyze many genes at once, instead of the top 3 or 4 as single-gene tests first.

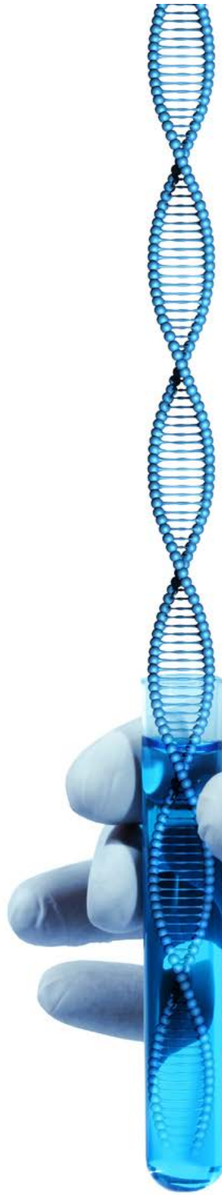
Exome testing is the most comprehensive test available and is often used when there are more complex clinical presentations or when other testing has already shown to be negative. It has an average diagnostic yield of 20-25%, but is more likely to return results of unknown significance than the other 2 types of testing.

Example: Patient symptoms/phenotype does not match any one diagnosis or set of diagnoses so exome testing is ordered to try and find the condition and cause (previous testing may have been done but would be negative).

Specimen Requirements

In an effort to make testing flexible and where possible, less invasive, EGL accepts a variety of specimen types, including:

- Whole blood
- Isolated DNA
- Saliva
- Urine
- Dried blood spot
- Serum
- Plasma
- Cerebral spinal fluid
- Cultured fibroblasts
- Muscle biopsy
- Skin biopsy
- Tissue biopsy
- Amniotic fluid
- Cultured amniocytes
- Bone marrow
- Chorionic villi
- Cord blood
- Products of conception



Customer Service

EGL has streamlined numerous processes to ensure test ordering is efficient and easy for our clients. The following services are available:

- Direct access to EGL board-certified laboratory directors and genetic counselors who assist clients with everything from choosing the right test to understanding the results
- Online test ordering
- Free specimen shipping kits, with prepaid return postage
- Reporting via secure email or fax
- Assistance with insurance pre-verification and pre-authorization

Genetic testing can be complex, but working with EGL isn't.

Certificates & Permits

CAP, ID Number: 7181693

CLIA, ID Number: 11D0683478

Florida Permit Number: 800026872

Georgia Permit Number: 044-170

Maryland Permit Number: 1346

New York Permit Number: 8951

Pennsylvania Permit Number: 031676

NPI Number: 1417330465

Tax ID Number: 47-4383500

More Experience, Better Results

Emory Genetics Laboratory was established in 1970, making it the oldest clinical genetics laboratory in the United States. Our laboratory directors and genetic counselors are board-certified to maintain the quality of the testing and reporting for which EGL is known. As a CLIA-certificatied and CAP-accredited laboratory, EGL maintains the highest of performance and quality standards.

The EGL leadership team includes:

Thomas M Schneider, MD

Medical Director

Michael Gambello, MD, PhD

Medical Director

Madhuri Hegde, PhD, FACMG

Executive Director and Chief Scientific Officer
Board-certified Clinical Molecular Geneticist

Lora H Bean, PhD, FACMG

Chief Quality Officer and Senior Molecular Laboratory Director
Board-certified Clinical Molecular Geneticist

Alice Tanner, PhD, MS, CGC, FACMG

Laboratory Director
Board-certified Clinical Molecular Geneticist

Christin Collins, PhD, FACMG

Laboratory Director
Board-certified Clinical Molecular Geneticist

Arunkanth Ankala, PhD, DABMG

Laboratory Director
Board-certified Clinical Molecular Geneticist

Hussain Askree, MD, PhD, FACMG

Laboratory Director
Board-certified Clinical Biochemical and Molecular Geneticist

John J Alexander, PhD, FACMG

Laboratory Director
Board-certified Clinical Molecular Geneticist

Patricia Hall, PhD, FACMG

Laboratory Director
Board-certified Clinical Biochemical Geneticist

Zunyan Dai, PhD, FACMG

Laboratory Director
Board-certified Clinical Cytogeneticist and Molecular Geneticist

For more information about EGL:

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