EGL Genetics | Eurofins Clinical Diagnostics

Test Directory

SUMMARY

EGL Genetics specializes in genetic diagnostic testing, with nearly 50 years of clinical experience and board-certified laboratory directors and genetic counselors reporting out cases. EGL Genetics offers a combined 1000 molecular genetics, biochemical genetics, and cytogenetics tests under one roof and custom testing for all medically relevant genes, for domestic and international clients. EGL Genetics is led by a team of laboratory directors with expertise spanning the fields of rare-disease testing (including metabolic and neuromuscular disease), genomic variant interpretation and test development research. In addition to the clinical offerings and internal research, EGL Genetics also collaborates on various external clinical and technology research projects. EGL Genetics receives samples from 49 states and more than 45 countries, and is also the follow-up laboratory for the State of Georgia Newborn Screening Program. As a CLIA-licensed and CAP-accredited laboratory, EGL Genetics is dedicated to providing superior, cutting-edge genetic testing for use in improving patient care.

EGL Genetics is committed to ensuring clients and their patients are kept as up-to-date as possible concerning the classification of sequence variants. As the first laboratory to contribute to the "free the data" movement, EGL Genetics has contributed (>35,000 submissions on >1700 genes) to ClinVar, the NCBI-sponsored variant database. EGL Genetics was also the first laboratory to develop its own free, online, no registration required public variant classification catalog. This catalog, called EmVClass, provides current classification status of all sequence variants detected by EGL Genetics. When new data emerges to support a variant classification change, EGL Genetics will issue amended reports for each patient with that variant, upon request.

We welcome feedback and suggestions, and encourage visiting the website (eglgenetics.com) for detailed descriptions of all available testing. For any question as to which type of testing to order, call 470-378-2200 to speak with our genetic counselors.

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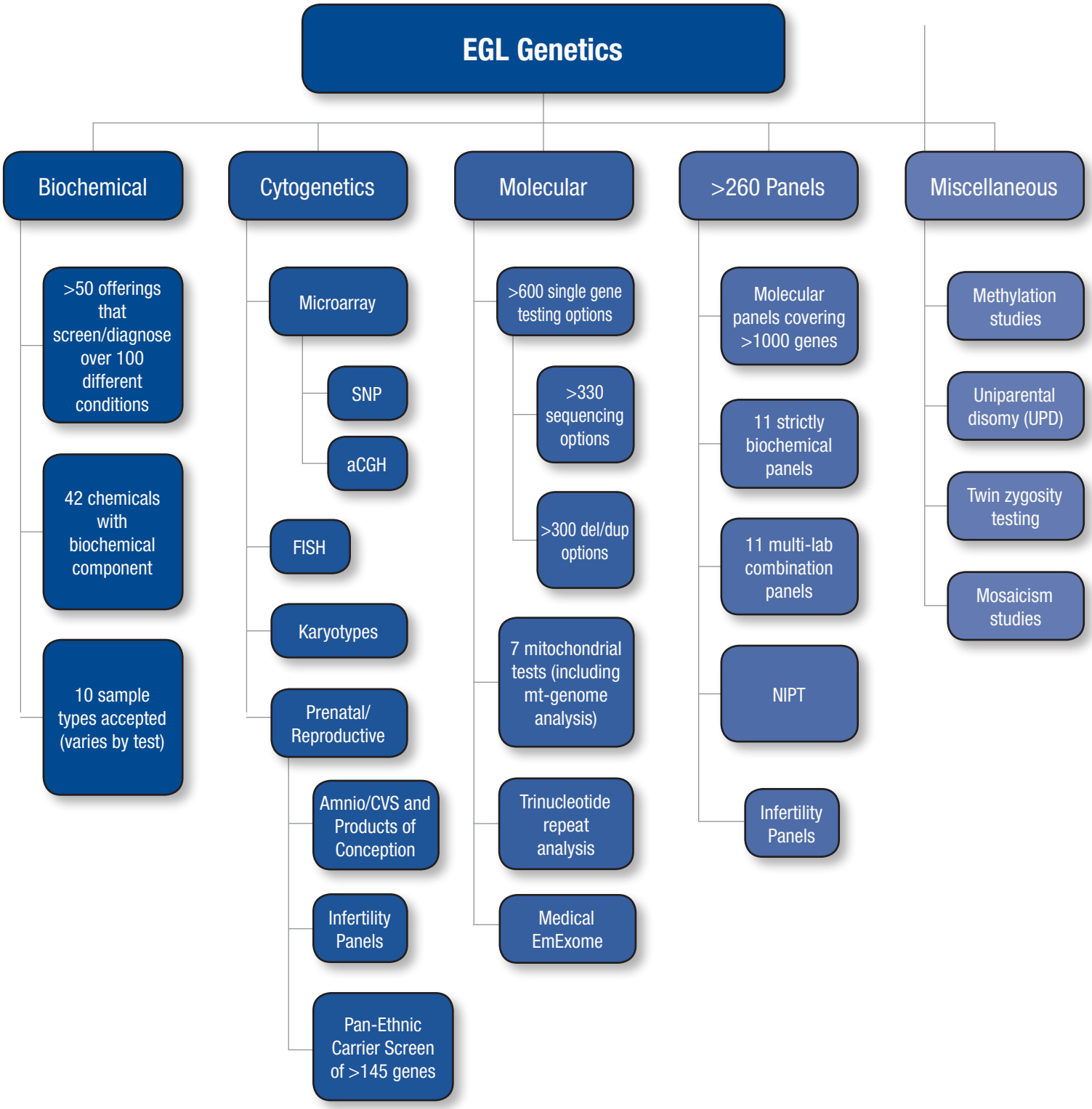


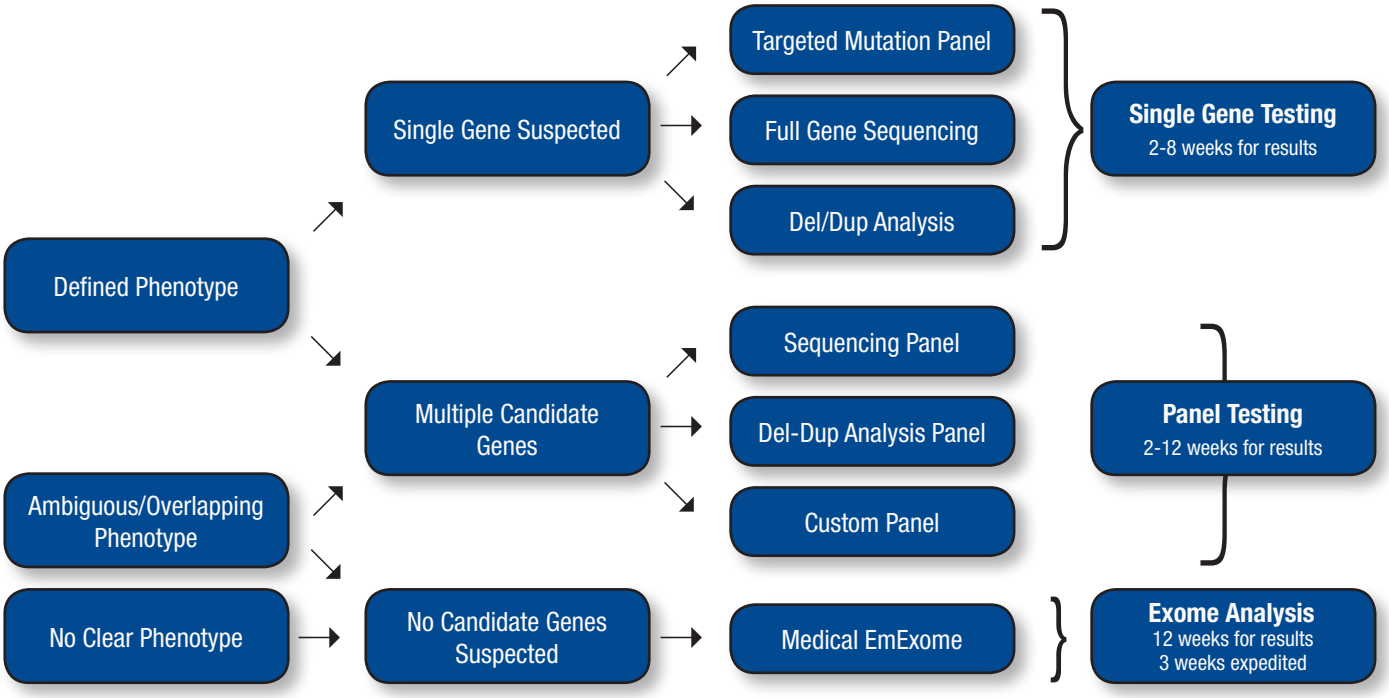
For more information about EGL Genetics:

CALL
470.378.2200

WEB
eglgenetics.com

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. This page will help you work through the different options available.



If the mutation for a particular condition is already known within a family, targeted mutation analysis is the best option. Please contact the laboratory to discuss ordering known mutation testing.

TARGETED MUTATION ANALYSIS vs. 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.
  - NOTE: Some conditions (e.g. Duchenne muscular dystrophy) are largely caused by deletions and thus ordering this array would make a better first-tier option.

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).

EXOME SEQUENCING

What is the Medical EmExome? The Medical EmExome sequencing design provides >97% coverage of 22,000 genes, with a mean read depth of 100X. Of the ~5000 disease-associated genes analyzed, 3000 have 100% coverage (≥20X) of all exons; twice the number of genes with complete coverage offered by competitors, making it the most comprehensive exome sequencing test available. This is usually the first-tier when ordering exome testing. If necessary this can be followed up with a Medical EmExome array which is a comprehensive deletion/duplication analysis of the exome.

What are the ordering options? It is best to perform exome testing on family trios (the patient and usually the patient's parents), as the additional information is used to help interpret some of the variants seen during analysis. Having additional affected or unaffected family members tested may also help achieve a diagnosis, which is why EGL Genetics has added the option of additional family member testing to any family trio. If additional family members are not available, proband only testing is also an option.

What is the EmExome Boost option? This option allows clinicians to choose a gene panel relevant to the patient's phenotype to ensure coverage of ALL exons in that panel (some of which may have been less than 100% on the exome itself), at no additional cost.

Variant Interpretation Updates-- As more information from human exome and genome sequencing projects becomes available, and as more research is conducted on previously reported DNA variants, knowledge of variant classification increases. This knowledge can allow variants previously classified as variants of uncertain clinical significance to be reclassified as pathogenic variants or benign polymorphisms. Reanalysis can be requested.

Other exome services include interpretation only, confirmation testing and interpretation, and exome sequencing without interpretation. Please refer to the website for more information.

For more information about EGL Genetics:
CALL 470.378.2200
WEB eglgenetics.com

TEST OFFERINGS

Here is a listing of testing available at EGL Genetics, including biochemical, molecular, and cytogenetics. Specific availability of single-gene sequencing and/or deletion and duplication analysis is noted. If the condition can be assessed through a panel, in addition to a single-gene test, this is noted in the PANEL column. Other test types (e.g. methylation, trinucleotide repeat analysis, and small targeted mutation panels) are also listed. If panel or biochemical testing is marked, more details on those offerings can be found in the corresponding Panel or Biochemical Testing sections of this directory.

If a particular gene or condition of interest is not listed, please contact EGL Genetics as custom testing is available for most other genes/conditions.

NOTE: Laboratory offerings are subject to change. Please visit eglgenetics.com for the most current testing information.

Name	Gene	Molecular Del/Dup	Molecular Sequencing	Biochemical	Cytogenetics	Cytogenetics STAT	Available on Panel	Other
22q11.2 Deletion Syndrome (DiGeorge)	*22q11.2*				•	•		Done as FISH analysis
3-Hydroxy-3-Methylglutaryl (HMG) CoA Lyase Deficiency	*HMGCL*	•	•	•			•	
Aarskog-Scott Syndrome	*FGD1*	•	•				•	
Acyl-CoA Dehydrogenase 9 Deficiency	*ACAD9*	•	•	•			•	
Adenosine Monophosphate Deaminase 1 Deficiency	*AMPD1*	•	•				•	Targeted 2 mutations (Q12X, P48L)
Adenosine Monophosphate Deaminase 3 Deficiency, Erythrocytic	*AMPD3*	•	•				•	
Adrenoleukodystrophy, X-linked	*ABCD1*	•	•	•				
Allan-Herndon-Dudley Syndrome	*SLC16A2*	•	•				•	
Alpha-Mannosidosis	*MAN2B1*	•	•	•			•	
Alpha-N-Acetylgalactosaminidase Deficiency	*NAGA*		•	•			•	
Alpha-Thalassemia	*HBA1 & HBA2*							HBA1 & HBA2 deletions
Alpha-Thalassemia X-linked Intellectual Disability Syndrome	*ATRX*	•	•				•	
Angleman Syndrome	*UBE3A*	•	•				•	
Angleman-like Syndrome	*SLC9A6*	•	•				•	
Argininosuccinate Lyase Deficiency	*ASL*	•	•	•			•	
Arthrogryposis, Distal, Type 2B	*TNNI2*	•	•				•	
Aspartylglucosaminuria	*AGA*		•	•			•	
Ataxia with Oculomotor Apraxia, Type 2	*SETX*		•					
Autism Susceptibility, X-linked 1	*NLGN3*	•	•				•	
Autism Susceptibility, X-linked 2	*NLGN4X*	•	•				•	
Autism Susceptibility, X-linked 5	*RPL10*	•	•				•	
Bamforth Lazarus Syndrome	*FOXE1*	•	•				•	
Beckwith-Wiedemann Syndrome (H19)	*H19*						•	Methylation
Beckwith-Wiedemann Syndrome (LIT1)	*LIT1*						•	Methylation
BEST1-related disorders	*BEST1*	•	•				•	
Beta-Ketothiolase Deficiency	*ACAT1*	•	•	•			•	
Beta-Mannosidosis	*MANBA*	•	•	•			•	
Biotinidase Deficiency	*BTD*	•	•	•			•	
Birt-Hogg-Dube Syndrome	*FLCN*	•	•				•	
Bloom Syndrome	*BLM*	•	•				•	
Borjeson-Forssman-Lehman Syndrome	*PHF6*	•	•				•	
BRAF-related disorders	*BRAF*	•	•				•	
Brody Myopathy	*ATP2A1*		•					
Brugada Syndrome	*CACNA1C*	•	•				•	
Brunner Syndrome	*MAOA*	•	•				•	
Cardiac Disorders, Congenital, Isolated Nonsyndromic	*NKX2-5*	•	•				•	
Cardiofaciocutaneous Syndrome, Type 3	*MAP2K1*	•	•				•	
Cardiofaciocutaneous Syndrome, Type 4	*MAP2K2*	•	•				•	
Cardiomyopathy, TPM1-related	*TPM1*	•	•				•	
Carnitine Palmitoyltransferase 1A Deficiency	*CPT1A*	•	•	•			•	
Carnitine Palmitoyltransferase 2 Deficiency	*CPT2*	•	•	•			•	
Carnitine-Acylcarnitine Translocase Deficiency	*SLC25A20*	•	•	•			•	
Carnnitine Deficiency, Primary	*SLC22A5*	•	•	•			•	

Name	Gene	Molecular Del/Dup	Molecular Sequencing	Biochemical	Cytogenetics	Cytogenetics STAT	Available on Panel	Other
CHARGE Syndrome	*CHD7*	•	•				•	
CHILD Syndrome	*NSDHL*	•	•				•	
Childhood Absence Epilepsy	*GABRB3*	•	•					
Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter 1	*EIF2B1*	•	•				•	
Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter 2	*EIF2B2*	•	•				•	
Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter 3	*EIF2B3*	•	•				•	
Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter 4	*EIF2B4*	•	•				•	
Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter 5	*EIF2B5*	•	•				•	
Choroideremia	*CHM*	•	•				•	
Citrullinemia	*ASS1*	•	•	•			•	
CK Syndrome	*NSDHL*	•	•				•	
CNTNAP2-related disorders	*CNTNAP2*	•	•				•	
Coffin-Lowry Syndrome	*RPS6KA3*	•	•				•	
Cohen Syndrome	*VPS13B*	•	•				•	
Congenital Disorder of Glycosylation, Type 1a	*PMM2*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 1b	*MPI*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 1c	*ALG6*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 1d	*ALG3*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 1e	*DPM1*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 1f	*MPDU1*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 1g	*ALG12*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 1h	*ALG8*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 1i	*ALG2*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 1j	*DPAGT1*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 1k	*ALG1*		•	•			•	
Congenital Disorder of Glycosylation, Type 1L	*ALG9*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 1m	*DOLK*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 1n	*RFT1*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 1o	*DPM3*		•	•			•	
Congenital Disorder of Glycosylation, Type 1p	*ALG11*		•	•			•	
Congenital Disorder of Glycosylation, Type 1q	*SRD5A3*		•	•			•	
Congenital Disorder of Glycosylation, Type 1r	*DDOST*		•	•			•	
Congenital Disorder of Glycosylation, Type 1s	*ALG13*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 1v	*NGYL1*		•	•			•	
Congenital Disorder of Glycosylation, Type 2a	*MGAT2*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 2b	*GCS1*	•	•	•				
Congenital Disorder of Glycosylation, Type 2c	*SLC35C1*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 2d	*B4GALT1*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 2e	*COG7*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 2f	*SLC35A1*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 2g	*COG1*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 2h	*COG8*	•	•	•			•	
Congenital Disorder of Glycosylation, Type 2i	*COG5*		•	•			•	
Congenital Disorder of Glycosylation, Type 2j	*COG4*		•	•			•	
Congenital Disorder of Glycosylation, Type 2L	*COG6*		•	•			•	
Congenital Hypothyroidism with Spiky Hair and Cleft Palate	*FOXE1*	•	•				•	
Cornelia de Lange Syndrome, Type 1	*NIPBL*	•	•				•	
Cornelia de Lange Syndrome, Type 2	*SMC1A*	•	•				•	
Costello Syndrome	*HRAS*	•	•				•	

Name	Gene	Molecular Del/Dup	Molecular Sequencing	Biochemical	Cytogenetics	Cytogenetics STAT	Available on Panel	Other
Cowden Syndrome	*PTEN*	•	•				•	
Cutis Laxa, Type IIA	*ATP6V0A2*	•	•	•			•	
Cystic Fibrosis	*CFTR*	•	•				•	
Danon Disease	*LAMP2*	•	•				•	
Deafness-Dystonia, Optic Neuronopathy Syndrome	*TIMM8A*	•	•				•	
Diffuse Gastric Cancer	*CDH1*		•				•	
Dihydrolipoamide Dehydrogenase Deficiency (a.k.a MSUD III)	*DLD*	•	•	•			•	
Duchenne/Becker Muscular Dystrophy	*DMD*	•	•				•	
Dyserythropoietic Anemia, Congenital, Type II	*SEC23B*		•				•	
Dyskeratosis Congenita, X-linked	*DKC1*	•	•				•	
ELN-related disorders	*ELN*		•				•	
Epidermolysis Bullosa, PLEC-related	*PLEC*	•	•				•	
Epilepsy, X-linked, with Variable Learning Disabilities and Behavior Disorders	*SYN1*	•	•				•	
Epileptic Encephalopathy, Early Infantile, Type 10	*PNKP*	•	•				•	
Epileptic Encephalopathy, Early Infantile, X-linked Female-Limited, Type 9	*PCDH19*	•	•				•	
Fabry Disease	*GLA*	•	•	•			•	
Familial Adenomatous Polyposis (FAP)	*APC*	•	•				•	
FISH, Chromosome 13					•	•	•	
FISH, Chromosome 18					•	•	•	
FISH, Chromosome 21					•	•	•	
FISH, Chromosomes X & Y					•	•	•	
FISH, Other					•			Various options are available. Please call to discuss with laboratory genetic counselor
FLNA-related disorders	*FLNA*	•	•				•	
Focal Dermal Hypoplasia	*PORCN*	•	•				•	
Formiminotransferase Deficiency	*FTCD*	•	•	•				
Fragile X Syndrome	*FMR1*	•	•				•	CGG Repeat Analysis
Fragile XE Syndrome	*AFF2*	•	•				•	CCG Repeats (females) CCG Repeats & Methylation (males)
Fucosidosis	*FUCA1*	•	•				•	
Galactosemia, Classic, Galactose-1-Phosphate Uridylyltransferase Deficiency	*GALT*	•	•	•			•	
Galactosemia, Epimerase Deficiency	*GALE*	•	•					
Galactosemia, Galactokinase Deficiency	*GALK1*	•	•	•				
Gaucher Disease	*GBA*		•	•			•	
Glucose Transporter Type 1 (GLUT 1) Deficiency Syndrome	*SLC2A1*	•	•				•	
Glucose-6-Phosphate (G6PD) Deficiency	*G6PD*	•	•				•	
Glutaric Aciduria, Type I	*GCDH*	•	•	•			•	
Glycerol Kinase Deficiency	*GK*	•	•	•			•	
Glycogen Storage Disease V (McArdle)	*PYGM*	•	•				•	Targeted 3 mutations (R49X, G204S, K542T)
GM1-Gangliosidosis	*GLB1*	•	•	•			•	
GM2-Gangliosidosis, AB Variant	*GM2A*		•	•			•	
Hearing Loss, Non-syndromic, (a.k.a. Connexin 26)	*GJB2*		•				•	
Hearing Loss, Non-syndromic, (a.k.a. Connexin 30)	*GJB6*		•				•	
Hereditary Hemochromatosis	*HFE*							Targeted Analysis
Hermansky-Pudlak Syndrome, Type 1	*HPS1*		•				•	
Hermansky-Pudlak Syndrome, Type 4	*HPS4*	•	•				•	
Holocarboxylase Synthetase Deficiency	*HLCS*	•	•	•			•	
Homocystinuria, CBS-deficient	*CBS*	•	•	•			•	
HSD17B10-related disorders (17-beta-hydroxysteroid dehydrogenase)	*HSD17B10*	•	•				•	
Huntington Disease	*HTT*							CAG Repeat Analysis

Name	Gene	Molecular Del/Dup	Molecular Sequencing	Biochemical	Cytogenetics	Cytogenetics STAT	Available on Panel	Other
Hydrocephalus with Aqueductal Stenosis, X-linked	*L1CAM*	•	•				•	
Hyperekplexia, ARHGEF9-related	*ARHGEF9*	•	•				•	
Hyperinsulinemic Hypoglycemia	*HADH*	•	•				•	
Hyperuricemic Nephropathy, Familial Juvenile 1	*UMOD*	•	•				•	
Hypophosphatasia	*ALPL*	•	•					
Hypothyroidism, Congenital, due to thyroid dysgenesis or hypoplasia	*PAX8*	•	•				•	
Ichthyosis Follicularis with Atrichia and Photophobia Syndrome	*MBTPS2*	•	•				•	
Inclusion Body Myopathy 2	*GNE*	•	•				•	
Intellectual Disability with Language and Impairment and Autistic Features	*FOXP1*	•	•				•	
Intellectual Disability, ARX-related disorders	*ARX*	•	•				•	
Intellectual Disability, Autosomal Dominant 1	*MBD5*	•	•				•	
Intellectual Disability, Autosomal Recessive, Type 7	*TUSC3*	•	•				•	
Intellectual Disability, Stereotypic Movements, Epilepsy, and/or Cerebral Malformation	*MEF2C*	•	•				•	
Intellectual Disability, X-linked, CASK-related disorders	*CASK*	•	•				•	
Intellectual Disability, X-linked, Claes-Jensen Type	*KDM5C*	•	•				•	
Intellectual Disability, X-linked, Hedera type	*ATP6AP2*	•	•				•	
Intellectual Disability, X-linked, KLF8-related	*KLF8*	•	•				•	
Intellectual Disability, X-linked, Nascimento Type	*UBE2A*	•	•				•	
Intellectual Disability, X-linked, PTCHD1-related	*PTCHD1*	•	•				•	
Intellectual Disability, X-linked, Raymond Type	*ZDHHC9*	•	•				•	
Intellectual Disability, X-linked, Siderius Type	*PHF8*	•	•				•	
Intellectual Disability, X-linked, Snyder Robinson Type	*SMS*	•	•				•	
Intellectual Disability, X-linked, Stocco Dos Santos Type	*SHROOM4*	•	•				•	
Intellectual Disability, X-linked, Turner Type	*HUWE1*	•	•				•	
Intellectual Disability, X-linked, Type 14	*UPF3B*	•	•				•	
Intellectual Disability, X-linked, Type 15 Cabezas	*CUL4B*	•	•				•	
Intellectual Disability, X-linked, Type 21/34	*IL1RAPL1*	•	•				•	
Intellectual Disability, X-linked, Type 30/47	*PAK3*	•	•				•	
Intellectual Disability, X-linked, Type 41	*GDI1*	•	•				•	
Intellectual Disability, X-linked, Type 58	*TSPAN7*	•	•				•	
Intellectual Disability, X-linked, Type 59	*AP1S2*	•	•				•	
Intellectual Disability, X-linked, Type 63	*ACSL4*	•	•				•	
Intellectual Disability, X-linked, Type 72	*RAB39B*	•	•				•	
Intellectual Disability, X-Linked, Type 9	*FTSJ1*	•	•				•	
Intellectual Disability, X-linked, Type 90	*DLG3*	•	•				•	
Intellectual Disability, X-linked, Type 91	*ZDHHC15*	•	•				•	
Intellectual Disability, X-linked, Type 93	*BRWD3*	•	•				•	
Intellectual Disability, X-linked, Type 94	*GRIA3*	•	•				•	
Intellectual Disability, X-linked, Type 96	*SYP*	•	•				•	
Intellectual Disability, X-linked, Type 97	*ZNF711*	•	•				•	
Intellectual Disability, X-linked, Type 98	*KIAA2022*	•	•				•	
Intellectual Disability, X-linked, with Agenesis of the Corpus Callosum, Ocular Coloboma, and Micrognathia	*IGBP1*	•	•				•	
Intellectual Disability, X-linked, with Cerebellar Hypoplasia and Distinctive Facial Appearance	*OPHN1*	•	•				•	
Intellectual Disability, X-linked, with Isolated Growth Hormone Deficiency	*SOX3*	•	•				•	
Isobutyrl Co-A Dehydrogenase Deficiency	*ACAD8*	•	•	•				
Isovaleric Acidemia	*IVD*	•	•	•			•	
Jalili Syndrome	*CNNM4*	•	•				•	
Juvenile Polyposis	*SMAD4*	•	•				•	
Kabuki Syndrome, Type 1	*KMT2D*	•	•				•	
Kabuki Syndrome, Type 2	*KDM6A*	•	•				•	

Name	Gene	Molecular Del/Dup	Molecular Sequencing	Biochemical	Cytogenetics	Cytogenetics STAT	Available on Panel	Other
Karyotype					•		•	Prenatal (Amnio, CVS, PUBS) Products of Conception (POC) Blood, skin or tissue
Kleefstra Syndrome	*EHMT1*	•	•				•	
Krabbe Disease	*GALC*	•	•	•			•	
KRAS-related disorders	*KRAS*	•	•				•	
Leber Hereditary Optic Neuropathy (LHON)	*Mitochondrial*							3460G>A in MT-ND1 11778G>A in MT-ND4 14459G>A & 14484T>C in MT-ND6
Legius Syndrome	*SPRED1*		•				•	
Leigh Syndrome, mitochondrial	*Mitochondrial*							9176T>C & 8993T>C in MT-ATP6 14459G>A in MT-ND6 3243A>G in MT-TL1
Leiomyomatoisis and Renal Cell Cancer	*FH*	•	•				•	
Lesch-Nyhan Syndrome	*HPRT1*	•	•				•	
Li-Fraumeni Syndrome	*TP53*	•	•				•	
Limb-Girdle Muscular Dystrophy Type 1A	*MYOT*	•	•				•	
Limb-Girdle Muscular Dystrophy Type 1B	*LMNA*	•	•				•	
Limb-Girdle Muscular Dystrophy Type 1C	*CAV3*	•	•				•	
Limb-Girdle Muscular Dystrophy Type 2A	*CAPN3*	•	•				•	
Limb-Girdle Muscular Dystrophy Type 2B	*DYSF*	•	•				•	
Limb-Girdle Muscular Dystrophy Type 2C	*SGCG*	•	•				•	
Limb-Girdle Muscular Dystrophy Type 2D	*SGCA*	•	•				•	
Limb-Girdle Muscular Dystrophy Type 2E	*SGCB*	•	•				•	
Limb-Girdle Muscular Dystrophy Type 2F	*SGCD*	•	•				•	
Limb-Girdle Muscular Dystrophy Type 2G	*TCAP*	•	•				•	
Limb-Girdle Muscular Dystrophy Type 2H	*TRIM32*	•	•				•	
Limb-Girdle Muscular Dystrophy Type 2L	*ANO5*	•	•				•	
Lissencephaly 1 /Subcortical Laminal Heterotopia	*PAFAH1B1*	•	•				•	
Lissencephaly, Type 2	*RELN*	•	•				•	
Lissencephaly, X-linked /Subcortical Laminal Heterotopia	*DCX*	•	•				•	
Long Chain 3-Hydroxy Acyl-CoA Dehydrogenase Deficiency	*HADHA*	•	•	•			•	
Lowe Syndrome	*OCRL*	•	•				•	
Lynch Syndrome, HNPCC Type 1	*MSH2*	•	•				•	
Lynch Syndrome, HNPCC Type 2	*MLH1*	•	•				•	
Lynch Syndrome, HNPCC Type 4	*PMS2*		•				•	
Lynch Syndrome, HNPCC Type 5	*MSH6*	•	•				•	
Lynch Syndrome, HNPCC Type 8	*EPCAM*	•					•	
Lysosomal Acid Lipase Deficiency	*LIPA*		•				•	
Malonyl-CoA Decarboxylase Deficiency	*MLYCD*		•	•			•	
Mandibuloacral Dysplasia with Type B Lipodystrophy, & Restrictive Dermopathy, Lethal	*ZMPSTE24*	•	•				•	
Marinesco-Sjogren Syndrome	*SIL1*	•	•				•	
MED12-related Disorders	*MED12*	•	•				•	
Medium Chain Acyl-CoA Dehydrogenase Deficiency	*ACADM*	•	•	•			•	Targeted 2 mutations (K304E, Y42H)
Medullary Cystic Kidney Disease, Type 2	*UMOD*	•	•				•	
Melanoma-Pancreatic Cancer	*CDKN2A*	•	•				•	
MELAS (Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-Like Episodes)	*Mitochondrial*							3243A>G in MT-TL1
Menkes Disease	*ATP7A*	•	•				•	
MERRF (Myoclonic Epilepsy and Ragged-Red Fiber Disease)	*Mitochondrial*							8344A>G & 8356T>C in MT-TK

Name	Gene	Molecular Del/Dup	Molecular Sequencing	Biochemical	Cytogenetics	Cytogenetics STAT	Available on Panel	Other
Metachromatic Leukodystrophy	*ARSA*	•	•	•			•	
Methylmalonic Aciduria (MUT)	*MUT*	•	•	•			•	
Methylmalonic Aciduria and Homocystinuria, cblC Type	*MMACHC*	•	•	•			•	
Microarray, CytoScan SNP					•		•	Prenatal (amnio, CVS, PUBS) Products of Conception (POC) Blood, tissue, or skin Also available on certain panels
Microarray, EmArray (CGH)					•		•	Prenatal (amnio, CVS, PUBS) Products of Conception (POC) Blood, tissue, or skin Also available on certain panels
Microphthalmia with Linear Skin Defects	*HCCS*	•	•				•	
Microphthalmia, Syndromic 2	*BCOR*	•	•				•	
Mitochondrial Complex I Deficiency	*NDUFA1*	•	•				•	
Mitochondrial Genome	*Mitochondrial*	•	•					
Mosaicism Study, Peripheral Blood					•			
Mowat-Wilson Syndrome	*ZEB2*	•	•				•	
Mucolipidosis, Type II	*GNPTAB*	•	•	•			•	
Mucolipidosis, Type III Gamma	*GNPTG*	•	•	•			•	
Mucolipidosis, Type IIIA	*GNPTAB*	•	•	•			•	
Mucopolysaccharidosis, Type I (Hurler, Scheie)	*IDUA*	•	•	•			•	
Mucopolysaccharidosis, Type II (Hunter)	*IDS*	•	•				•	
Mucopolysaccharidosis, Type IIIA (Sanfilippo A)	*SGSH*	•	•				•	
Mucopolysaccharidosis, Type IIIB (Sanfilippo B)	*NAGLU*	•	•				•	
Mucopolysaccharidosis, Type IIIC (Sanfilippo C)	*HGSNAT*	•	•				•	
Mucopolysaccharidosis, Type IIID (Sanfilippo D)	*GNS*	•	•				•	
Mucopolysaccharidosis, Type IVA (Morquio A)	*GALNS*	•	•				•	
Mucopolysaccharidosis, Type IVB (Morquio B)	*GLB1*	•	•	•			•	
Mucopolysaccharidosis, Type VI (Maroteaux-Lamy)	*ARSB*	•	•	•			•	
Mucopolysaccharidosis, Type VII (Sly)	*GUSB*	•	•	•			•	
Multiple Endocrine Neoplasia, Type 1	*MEN1*	•	•				•	
Multiple Endocrine Neoplasia, Type 2	*RET*	•	•				•	
Multiple Sulfatase Deficiency	*SUMF1*	•	•	•			•	
Muscle-Eye-Brain Disease	*POMGNT1*	•	•				•	
Muscular Dystrophy, Bethlem Myopathy (COL6A1)	*COL6A1*	•	•				•	
Muscular Dystrophy, Bethlem Myopathy (COL6A2)	*COL6A2*	•	•				•	
Muscular Dystrophy, Bethlem Myopathy (COL6A2)	*COL6A3*	•	•				•	
Muscular Dystrophy, Congenital, Fukuyama	*FKTN*	•	•				•	3kb retrotransposon 3' UTR insertion assay
Muscular Dystrophy, Congenital, Ullrich (COL6A1)	*COL6A1*	•	•				•	
Muscular Dystrophy, Congenital, Ullrich (COL6A2)	*COL6A2*	•	•				•	
Muscular Dystrophy, Congenital, Ullrich (COL6A3)	*COL6A3*	•	•				•	
Muscular Dystrophy, Congenital, with Integrin Alpha-7 Deficiency	*ITGA7*	•	•				•	
Muscular Dystrophy, Congenital, with Rigid Spine	*SEPN1*	•	•				•	
Muscular Dystrophy, Emery-Dreifus, X-linked	*EMD*	•	•				•	
Muscular Dystrophy, Merosin-Deficient Congenital 1A	*LAMA2*	•	•				•	
Muscular Dystrophy, Merosin-Deficient Congenital 1C	*FKRP*	•	•				•	
Muscular Dystrophy, Merosin-Deficient Congenital 1D	*LARGE*	•	•				•	
Muscular Dystrophy, Oculopharyngeal	*PABPN1*						•	GCN Repeat Analysis
Myoclonus-Dystonia	*SGCE*	•	•				•	
Myofibrillar Myopathy 2	*DES*	•	•				•	
Myoglobinuria, Acute Recurrent, Autosomal Recessive	*LPIN1*		•				•	
Myotonic Dystrophy, Type 1	*DMPK*						•	CTG Repeat Analysis
Myotubular Myopathy, X-linked	*MTM1*	•	•				•	

Name	Gene	Molecular Del/Dup	Molecular Sequencing	Biochemical	Cytogenetics	Cytogenetics STAT	Available on Panel	Other
Nance-Horan Syndrome	*NHS*	•	•				•	
NARP (Neuropathy, Ataxia, and Retinitis Pigmentosis)	*Mitochondrial*							8993T>G & 8993T>C in MT-ATP6
Nemaline Myopathy 1	*TPM3*	•	•				•	
Nemaline Myopathy 2	*NEB*	•	•				•	
Nemaline Myopathy 3	*ACTA1*	•	•				•	
Nemaline Myopathy 4	*TPM2*	•	•				•	
Nemaline Myopathy 5	*TNNT1*	•	•				•	
Nephronophthisis 1, Juvenile	*NPHP1*	•	•				•	
Nephronophthisis 2, Infantile	*INVS*	•	•				•	
Nephronophthisis 3	*NPHP3*	•	•				•	
Nephronophthisis 4	*NPHP4*	•	•				•	
Neurodegeneration due to Cerebral Folate Transport Deficiency	*FOLR1*	•	•				•	
Niemann-Pick Disease, Type A & B (a.k.a. Acid Sphingomyelinase Deficiency)	*SMPD1*	•	•				•	
Noonan Syndrome, Type 1	*PTPN11*	•	•				•	
Noonan Syndrome, Type 2	*RAF1*	•	•				•	
Noonan Syndrome, Type 4	*SOS1*	•	•				•	
Noonan Syndrome, Type 6	*NRAS*	•	•				•	
Noonan-like Syndrome with Loose Anagen Hair	*SHOC2*	•	•				•	
Norrie Disesase	*NDP*	•	•				•	
Oculo-Facio-Cardio-Dental (OFCD) Syndrome	*BCOR*	•	•				•	
Opitz GBBB Syndrome, X-linked	*MID1*	•	•				•	
Optic Atrophy, Autosomal Dominant	*OPA3*	•	•				•	
Optic Atrophy, Autosomal Dominant 1, Kjer Type	*OPA1*	•	•				•	
Oral-Facial-Digital Syndrome	*OFD1*	•	•				•	
Ornithine Transcarbamylase (OTC) Deficiency	*OTC*	•	•	•			•	
Papillary Renal Carcinoma	*MET*	•	•				•	
Paranganglioma-Pheochromacytoma Syndrome	*SDHB*		•				•	
PAX6-related Disorders	*PAX6*	•	•				•	
Pearson Marrow-Pancreas Syndrome	*Mitochondrial*	•						
Pelizaeus-Merzbacher Disease	*PLP1*	•	•				•	
Peters Plus Syndrome	*B3GALTL*	•	•				•	
Peutz-Jeghers Syndrome	*STK11*	•	•				•	
Phenylketonuria	*PAH*	•	•	•			•	
Phosphoglycerate Kinase-1 Deficiency	*PGK1*	•	•	•			•	
Pitt-Hopkins Syndrome	*TCF4*	•	•				•	
Pitt-Hopkins-like Syndrome 2	*NRXN1*	•	•				•	
Polycystic Kidney Disease, Autosmal Recessive	*PKHD1*	•	•				•	
Polycystic Kidney Disease, Autosomal Dominant, Type 2	*PKD2*	•	•				•	
Polycystic Liver Disease (PRKCSH)	*PRKCSH*	•	•					
Polycystic Liver Disease (SEC63)	*SEC63*	•	•				•	
Polyposis, MUTYH-Associated	*MUTYH*	•	•				•	Targeted 2 mutations (Y179C, G396D)
Pompe Disease	*GAA*	•	•	•			•	
Prader-Willi/Angelman Syndrome	*15q11.2*				•		•	Methylation
PRPS1-related Disorders	*PRPS1*	•	•				•	
PSAP-related Disorders	*PSAP*	•	•				•	
Pyruvate Dehydrogenase Deficiency	*PDHA1*	•	•	•			•	
Renpenning Syndrome 1	*PQBP1*	•	•				•	
Retinitis Pigmentosa 59	*DHDDS*		•				•	
Retinitis Pigmentosa, X-Linked, Type 2	*RP2*	•	•				•	
Retinitis Pigmentosa, X-Linked, Type 3	*RPGR*	•	•				•	
Retinoblastoma	*RB1*	•	•				•	

Name	Gene	Molecular Del/Dup	Molecular Sequencing	Biochemical	Cytogenetics	Cytogenetics STAT	Available on Panel	Other
Rett Syndrome	*MECP2*	•	•				•	
Rett Syndrome, Atypical	*CDKL5*	•	•				•	
Rett Syndrome, Congenital Variant	*FOXG1*	•	•				•	
Rothmund-Thomson Syndrome	*RECQL4*	•	•				•	
Rubinstein-Taybi Syndrome, Type 1	*CREBBP*	•	•				•	
Rubinstein-Taybi Syndrome, Type 2	*EP300*		•				•	
RYR1-related disorders	*RYR1*	•	•				•	
RYR2-related disorders	*RYR2*	•	•				•	
Sandhoff Disease	*HEXB*	•	•				•	
SCN1A-related disorders	*SCN1A*	•	•				•	
SHOX-related disorders	*SHOX*		•				•	
Sialic Acid Storage (Salla) Disease	*SLC17A5*		•	•			•	
Sialidosis	*NEU1*		•	•			•	
Sickle Cell Disease, including Hemoglobin C	*HBB*						•	E6V (Hgb S) & E6K (Hgb C)
Simpson-Golabi-Behmel Syndrome, Type 1	*GPC3*	•	•				•	
Smith-Lemli-Opitz Syndrome	*DHCR7*	•	•	•			•	
Smith-Magenis Syndrome	*RAI1/17p11.2*	•	•		•			
Sotos Syndrome	*NSD1*	•	•				•	
Speech-Language Disorder 1	*FOXP2*	•	•				•	
Spinal Muscular Atrophy	*SMN1*						•	Diagnostic Deletion & Carrier Dosage
Spinocerebellar Ataxia Type 1	*ATXN1*							CAG Repeat Analysis
Sterol-C4-Methyl Oxidase (SC4MOL) Deficiency	*MSMO1*		•					
Succinyl-CoA: 3-Oxoacid CoA Transferase (SCOT) Deficiency	*OXCT1*		•	•				
TAR (Thrombocytopenia-Absent Radius) Syndrome	*1q21.1/ RBM8A*		•		•		•	
Tay-Sachs Disease	*HEXA*	•	•	•			•	
Timothy Syndrome	*CACNA1C*	•	•				•	
TTN-related disorders	*TTN*	•	•				•	
Twin Zygosity: DNA-Based Testing								Zygosity testing
Tyrosinemia, Type I	*FAH*	•	•	•			•	
Uniparental Disomy (UPD) 14							•	Methylation
Uniparental Disomy (UPD) 6								Methylation
VACTERL-Hydrocephalus Syndrome	*FANCB*	•	•				•	
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	*ACADVL*	•	•	•			•	
Von Hippel-Lindau Syndrome	*VHL*	•	•				•	
Walker-Warburg Syndrome, Type 1	*POMT1*	•	•				•	
Walker-Warburg Syndrome, Type 2	*POMT2*	•	•				•	
Walker-Warburg Syndrome, Type 4	*FKTN*	•	•				•	
Wilson Disease	*ATP7B*		•				•	
Wolman Disease	*LIPA*		•				•	
Y-Chromosome: Microdeletion Analysis	*Yq11*						•	Deletion analysis of AZF regions a-d (usually undetected by cytogenetics)
Y-Chromosome: SRY Analysis	*SRY*				•			
Zellweger Syndrome Spectrum (PEX 1)	*PEX1*	•	•				•	
Zellweger Syndrome Spectrum (PEX 2)	*PEX2*	•	•				•	
Zellweger Syndrome Spectrum (PEX 3)	*PEX3*	•	•				•	
Zellweger Syndrome Spectrum (PEX 5)	*PEX5*	•	•				•	
Zellweger Syndrome Spectrum (PEX 6)	*PEX6*	•	•				•	
Zellweger Syndrome Spectrum (PEX12)	*PEX12*	•	•				•	
Zellweger Syndrome Spectrum (PEX14)	*PEX14*	•	•				•	
Zellweger Syndrome Spectrum (PEX26)	*PEX26*	•	•				•	

PANELS

Here is a listing of panels (multi-gene or condition-specific) available at EGL Genetics. In addition to the number of genes analyzed on each panel, the specific availability of sequencing, deletion/duplication analysis, or combined sequencing and deletion/duplication analysis options is noted. Some genes are not included in deletion/duplication analyses, and thus a gene number in parentheses denotes the number of genes in the deletion and duplication option versus the sequencing option. A '+' in the NUMBER OF GENES column denotes biochemical and/or cytogenetics testing is included in the test (also noted as "Combination" in the OTHER column). If you do not find a panel that fits your specific needs, please note that custom panels can be created. In addition, all genes on next generation sequencing panels may be ordered individually, even if the genes are not listed in the main directory chart.

NOTE: Laboratory offerings and genes included on panels are subject to change. Please visit eglgenetics.com for the most current testing information.

Panel Name	# of Genes	Deletion/ Duplication	Sequencing	Sequencing with Deletions/ Duplications	Other
3-Methylcrotonyl-CoA Carboxylase (3-MCC) Deficiency Panel	2	•	•		
ACOG/ACMG Carrier Screen: Gene Sequencing Panel	10		•		
ACOG/ACMG Carrier Screen: Targeted Mutation Panel	10				Targeted 192 mutations
Achromatopsia, Cone, and Cone-rod Dystrophy Panel	36	•	•		
Albinism Panel	7 (5)	•	•		
Anophthalmia/Microphthalmia/Anterior Segment Dysgenesis/Anomaly Panel	23	•	•		
Achalasia Panel	6		•		
Arrhythmia Panel	37	•	•		
Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy Panel	8	•	•		
Ashkenazi Jewish Carrier Screen: Gene Sequencing Panel	20		•		
Ashkenazi Jewish Carrier Screen: Targeted Mutation Panel	20				Targeted 223 mutations
Autism Spectrum Disorders Panel: Complete Tier 1	1+				Combination
Autism Spectrum Disorders Panel: Tier 1	1+				Combination
Autism Spectrum Disorders Panel: Tier 2 (Molecular)	63 (61)	•	•		
Autism Spectrum Disorders Panel: Tier 1 (Biochemical)					Biochemical
Bardet-Biedl Syndrome Panel	18	•	•		
Beckwith-Wiedemann Syndrome Methylation Panel					Methylation
Bethlem Myopathy and Ullrich Congenital Muscular Dystrophy Panel	3	•	•		
Brain and Nervous System Cancer Panel	16 (15)	•	•		
Brain Malformation Panel	50	•	•		
BRCA Ashkenazi Jewish Targeted Mutation Panel	2				Targeted 3 mutations
Breast and Ovarian Cancer Panel	24 (22)			•	
Breast Cancer High Risk Panel	7(5)			•	
Bronchiectasis Panel	16	•	•		
Brugada Syndrome Panel	8	•	•		
Cardiomyopathy (Dilated) Panel	39 (37)	•	•		
Cardiomyopathy (Hypertrophic) Panel	20 (19)	•	•		
Cardiomyopathy Panel	65 (62)	•	•		
Cardiovascular Comprehensive Panel	117 (106)	•	•		
Central Hypoventilation Congenital Syndrome Panel	7	•	•		
Cerebral Cavernous Malformation Panel	3	•	•		
Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing White Matter Panel	5	•	•		
Cholestasis Panel	57	•	•		
Ciliopathy Panel	112 (96)	•	•		
Colorectal and Gastrointestinal Cancer Panel	19	•	•		
Colorectal Cancer High Risk Panel	15	•	•		
Congenital Disorder of Glycosylation: Biochemical Panel					Biochemical
Congenital Disorders of Glycosylation Panel	66 (65)	•	•		

Panel Name	# of Genes	Deletion/ Duplication	Sequencing	Sequencing with Deletions/ Duplications	Other
Congenital Muscular Dystrophy Panel	24	•	•		
Congenital Obesity Panel	29		•		
Connective Tissue Disorder Panel	29	•	•		
Cornelia de Lange Syndrome Panel	5	•	•		
Cystic Fibrosis Common Mutation Panel	1				Targeted 39 mutations
Cystic Fibrosis Expanded Mutation Panel	1				Targeted 142 mutations
Cystic Lung Disease Panel	8	•	•		
Dystonia Panel	80		•		
Endocrine Cancer Panel	15 (13)	•	•		
Endocrine Disorder Panel	57	•	•		
Epilepsy and Seizure Disorder Panel	110 (107)	•	•		
Eye Disorder Panel	210 (207)	•	•		
FISH, Prenatal, Chromosomes 13, 18, 21, X & Y					Cytogenetic
Flecked-Retina Disorder Panel	6	•	•		
Galactosemia, Classic Biochemical Panel					Biochemical
Gaucher Disease Biomarker Screening Panel					Biochemical
Glycogen Storage Disorder Comprehensive Panel	20	•	•		
Glycogen Storage Disorder, Liver Panel	11	•	•		
Glycogen Storage Disorder, Muscle Panel	12	•	•		
Hearing Loss, Common Panel	2+				Combination
Hearing Loss, Connexin Panel	2	•	•		
Hearing Loss, Expanded Panel	131	•	•		
Hearing Loss, Mitochondrial Panel	2				Targeted 4 mutations
Hemophagocytic Lymphohistiocytosis Panel	16		•		
Hereditary Breast and Ovarian Cancer Syndrome Panel	2	•	•	•	
Hereditary Cancer Syndrome Panel	60 (55)	•	•		
Hereditary Hemorrhagic Telangiectasia Panel	5	•	•		
Hermansky-Pudlak Syndrome (Pulmonary Fibrosis) Panel	17	•	•		
Hypercholesterolemia Panel	24		•		
Hyper IgE Syndrome Panel	4	•	•		
Hypohidrotic Ectodermal Dysplasia Panel	3	•	•		
Hypothyroidism Congenital Panel	2	•	•		
Hypotonia Congenital Panel	4+				Combination
Infertility Panel: Female	6				Combination
Infertility Panel: Male	5				Combination
Inflammatory Bowel Disease Panel	26	•	•		
Interstitial Lung Disease	7 (5)	•	•		
Intellectual Disability, X-linked Panel	91	•	•		
Joubert Syndrome Panel	18	•	•		
Kabuki Syndrome Panel	2	•	•		
Leber Congenital Amaurosis Panel	23	•	•		
Limb Malformation Panel	46	•	•		
Limb-Girdle Muscular Dystrophy Panel	35 (33)	•	•		
Long and Short QT Syndrome Panel	13 (12)	•	•		
Lynch Syndrome (HNPCC) Panel 1	3	•	•		
Lynch Syndrome (HNPCC) Panel 2	4		•		
Lynch Syndrome (HNPCC) Panel 3	4			•	
Lynch Syndrome (HNPCC) Panel 4	4(4)			•	
Lysosomal Storage Disease: 13 Enzyme Panel					Biochemical
Lysosomal Storage Disorder Panel	55(54)	•	•		
Lysosomal Storage Disorder: Biochemical Screening Panel					Biochemical

Panel Name	# of Genes	Deletion/ Duplication	Sequencing	Sequencing with Deletions/ Duplications	Other
Mabry Syndrome	6		•		
Macrocephaly and Overgrowth Syndromes	23 (11)	•	•		Includes methylation of H19 and LIT1
Macular Dystrophy/Degeneration/Stargardt Disease Panel	15	•	•		
Maple Syrup Urine Disease Panel (BCKD Complex)	3	•	•		
Marfan Syndrome and Related Disorders Panel	17	•	•		
Maturity-Onset Diabetes of the Young Panel	4	•	•		
Melanoma Panel	13	•	•		
Metabolic Disease Biochemical Panel					Biochemical
Metabolic Disorder Panel	103 (100)	•	•		
Methylmalonic Aciduria (cblA & cblB) Panel	2	•	•		
Mitochondrial Diseases - Nuclear Genes Only	44	•	•		
Mucopolysaccharidosis Type III Panel (Sanfilippo A-D)	4	•	•		
Mucopolysaccharidosis: Biochemical Screening Panel					Biochemical
Multiple Acyl-CoA Dehydrogenase Deficiency Panel	3	•	•		
Multiple Epiphyseal Dysplasia Panel	7	•	•		
Myasthenic Congenital Syndrome Panel	11	•	•		
Nephronophthisis Panel	4	•	•		
Neurological Disease Panel	167 (163)	•	•		
Neuromuscular Disorder Expanded Panel	79 (78)	•	•		
Neuromuscular Disorder Panel	46 (45)	•	•		
Neuronal Ceroid-Lipofuscinosis Panel	11	•	•		
Neuropathy Panel	90	•	•		
Niemann-Pick Disease, Type C Panel	2	•	•		
Noonan Spectrum Disorder Panel	13		•		
Optic Atrophy Panel	5	•	•		
Osteogenesis Imperfecta and Decreased Bone Density Disorder Panel	36 (34)	•	•		
Pancreatic Cancer Panel	14	•	•		
Pan-Ethnic Carrier Screen: Gene Sequencing Panel	>145	•	•		Includes del/dup analysis of 6 genes
Pan-Ethnic Carrier Screen: Targeted Mutation Panel	>145	•			722 targeted mutations and del/dup analysis of 6 genes
Paraganglioma-Pheochromocytoma Panel	9	•	•		
Periodic Fever Syndrome Panel	7	•	•		
Phenylketonuria Biochemical Monitoring Panel					Biochemical
Premature Ovarian Failure Panel	21		•		FMRICGG-repeat analysis
Rhabdomyolysis Panel	25		•		
Propionic Acidemia Panel	2	•	•		
Pulmonary Disease Panel	52	•	•		
Pulmonary Fibrosis and Hermansky-Pudlak Syndrome Panel	16	•	•		
Pulmonary Hypertension Panel	8	•	•		
Renal Cancer Panel	23 (22)	•	•		
Retina/Photoreceptor Dystrophy Panel	121	•	•		
Retinitis Pigmentosa Panel	66	•	•		
Russell-Silver Syndrome					Combination
Sarcoglycanopathy Panel	4	•	•		
Senior Loken Syndrome Panel	7	•	•		
Severe Combined Immunodeficiency (SCID) B- Panel	7	•	•		
Severe Combined Immunodeficiency (SCID) B+ Panel	14	•	•		
Severe Combined Immunodeficiency (SCID) B+/B- Panel	21	•	•		
Skeletal Dysplasia Comprehensive Panel	173 (162)	•	•		
Skeletal Dysplasia, Disproprotionate Short Stature Panel	85 (76)	•	•		

Panel Name	# of Genes	Deletion/ Duplication	Sequencing	Sequencing with Deletions/ Duplications	Other
Skeletal Dysplasia with Increased Bone Density Panel	22	•	•		
Skeletal Dysplasia, Proportionate Short Stature/Small for Gestational Age Panel 1	45+				Combination
Skeletal Dysplasia, Proportionate Short Stature/Small for Gestational Age Panel 2	45+				Combination
Skeletal Dysplasia, Proportionate Short Stature/Small for Gestational Age Panel 3	45	•	•		
Skeletal Dysplasia, Proportionate Short Stature/Small for Gestational Age Panel 4	45+				Combination
Stationary Night Blindness, Congenital Panel	15	•	•		
Steroid-resistant Nephrotic Syndrome Panel	27		•		
Stickler Syndrome Panel	5	•	•		
Sudden Cardiac Arrest Disorder Panel	11 (10)	•	•		
TAR (Thrombocytopenia-Absent Radius) Syndrome Panel	1+				Combination
Trifunctional Protein Deficiency Panel	2	•	•		
Tuberous Sclerosis Panel	2	•	•		
Usher Syndrome Panel	12	•	•		
Vitreoretinopathy Panel	9	•	•		
Wilms Tumor Panel	2	•	•		
X-Linked Intellectual Disability Panel	92 (91)	•	•		
Zellweger Syndrome Spectrum Panel	14	•	•		

BIOCHEMICAL TESTING

Here is a listing of biochemical testing performed at EGL Genetics and the various sample types accepted for each test.

NOTE: Laboratory offerings are subject to change. Please visit eglgenetics.com for the most current testing information.

Name	Available on a Panel	Cerebral Spinal Fluid	Dried Blood Spot	Plasma	Red Blood Cells	Serum	Urine	White Blood Cells	Other Sample Types & Other Details
7-dehydrocholesterol	•			•					
Acetylcholinesterase (ACHE)									amniotic fluid
Acylcarnitine Profile	•			•					STAT testing available
Allo-isoleucine & Branched Chain Amino Acids			•						
Alpha-Fetoprotein, Amniotic Fluid (AFAFP)									amniotic fluid
Alpha-Fucosidase	•							•	
Alpha-Galactosidase								•	
Alpha-L-Iduronidase	•							•	
Alpha-Mannosidase	•							•	
Amino Acid Profile	•	•		•			•		STAT testing available
Angiotensin Converting Enzyme (ACE)	•					•			
Arylsulfatase A	•							•	
Arylsulfatase B	•							•	
Autism Spectrum Disorder Panel: Complete Tier 1	•								
Autism Spectrum Panel: Tier 1 (Biochemical)	•								
Beta-Galactosidase								•	
Beta-Glucosidase	•							•	
Beta-Glucuronidase	•							•	
Beta-Mannosidase	•							•	
Biotinidase						•			
Carbohydrate Deficient Transferrin	•			•					
Carnitine Profile	•			•			•		
Chitotriosidase (CHITO)	•					•			
Coenzyme Q10				•					
Congenital Disorder of Glycosylation: Biochemical Panel	•								
Galactitol							•		
Galactokinase					•				
Galactose-1-Phosphate	•				•				
Galactose-1-Phosphate Uridyltransferase	•				•				carrier enzyme testing also available
Galactosemia, Classic Biochemical Panel	•								
Gaucher Disease Biochemical Screening Panel	•								
Gaucher Disease Biomarker Screening Panel	•								
Globotriaosylceramine (Gb3)							•		
Glycosaminoglycans (GAGs)	•						•		
Hexosaminidase A	•							•	NOTE: this is not appropriate for carrier testing or Sandhoff disease testing
Homocysteine	•		•	•					
Lysosomal Storage Disease: 13 Enzyme Panel	•								
Lysosomal Storage Disorder: Biochemical Screening Panel	•								
Metabolic Disease Biochemical Panel	•								
Methylmalonic Acid	•			•			•		
Methylmalonic Acid and Methylcitric Acid			•						

Name	Available on a Panel	Cerebral Spinal Fluid	Dried Blood Spot	Plasma	Red Blood Cells	Serum	Urine	White Blood Cells	Other Sample Types & Other Details
Mucopolysaccharidosis: Biochemical Screening Panel	•								
Newborn Screening Follow-up Panel	•								
N-Glycan Profile	•					•			
O-Glycan Analysis						•			
Oligosaccharide Screen	•						•		
Organic Acid Profile	•						•		STAT testing available
Orotic Acid							•		
Phenylketonuria Biochemical Monitoring Panel	•								
Pyruvic Acid		•							whole blood (special prep needed)
Rhabdomyolysis: Tier 1 Panel	•								
Rhabdomyolysis: Tier 2 Panel	•								
Sterols				•					
Sialic Acid (free)	•						•		
Tartrate Resistant Acid Phosphatase (TRAP)	•					•			

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