About EGL Genetics
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.

Equally important to improving patient care through quality genetic testing is the contribution EGL Genetics makes back to the scientific and medical communities. EGL Genetics is one of only a few clinical diagnostic laboratories to openly share data with the NCBI freely available public database ClinVar (>35,000 variants on >1700 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.

Prenatal Genetic Testing
EGL Genetics offers numerous testing options for prenatal care, including cytogenetics and molecular testing, designed to provide rapid and accurate diagnoses for patients and families.

CytoScan SNP Array Prenatal
The Chromosomal Microarray, CytoScan SNP Array Prenatal provides high-definition copy number analysis using the most current methods and software. Couples choosing prenatal diagnosis now have the option of microarray analysis to optimize detection of submicroscopic genetic imbalances. As the founding member of the International Standard Cytogenetic Array Consortium (ISCA), Emory Genetics leads the industry in quality improvement efforts in chromosomal microarray testing as well as improved genetic healthcare for patients. Our ABMG-certified cytogeneticists, molecular geneticists, and genetic counselors work with the ordering clinician to assist in clinical correlations with significant array finding(s).

EmArray Cyto Prenatal
The EmArray Cyto Prenatal microarray is a 60K oligonucleotide array that detects copy number aberrations across the genome. With an increased detection rate over standard chromosome analysis, this chromosome microarray is designed according to International Standards of Cytogenomic Arrays Consortium (ISCA) standards and targets ~500 clinically relevant loci, with an average probe spacing of 75 kilobases (kb). The American College of Obstetrics and Gynecology has endorsed microarray analysis for prenatal testing and recommends its use in pregnancies with abnormal ultrasound findings and a normal G-banded chromosome analysis.

Indications:
- Abnormal ultrasound findings
- Abnormal serum screening results
- Familial chromosome rearrangement
- Advanced maternal age
- Suspected deletion/duplication syndrome

FISH and Chromosome Analysis
Rapid Screen
Aneuploidies of chromosomes 13, 18, 21, X, and Y are the most common prenatal chromosome abnormalities. Analysis by rapid screen FISH detects these chromosome aneuploidies within 24-48 hours of sample receipt, providing accurate and fast detection of abnormalities.

Reference:
Chromosome Analysis
The risk for chromosome abnormalities in a pregnancy increases with maternal age. Chromosome analysis by conventional karyotype is the standard of care to detect chromosome aneuploidy, large deletions/duplications of chromosomes, and balanced chromosome rearrangements. For smaller genetic imbalances beyond the resolution of a karyotype, the Prenatal EmArray Cyto is recommended.

Indications:
- Advanced maternal age
- Abnormal serum screen
- Abnormal ultrasound findings

Known Mutation Testing
EGL Genetics offers targeted prenatal mutation analysis for previously identified familial mutations. An affected family member and/or both biological parents must be tested and found to carry the familial mutation. Prenatal testing is not available for variants of unknown clinical significance.

Indications:
- Known familial sequence mutation
- Known familial whole or partial gene deletion/duplication
- Family history of genetic disease

PKHD1 Full Gene Sequencing
Autosomal recessive polycystic kidney disease (ARPKD) is a genetic disease characterized by large, echogenic kidneys, typically presenting in the neonatal period. EGL offers prenatal testing, including full gene sequencing and deletion/duplication analysis for PKHD1, when there is a prenatal indication of ARPKD.

Indications:
- Ultrasound abnormalities suggestive of ARPKD
- Family history of ARPKD

L1CAM Full Gene Sequencing
Mutations in the L1CAM gene result in a phenotypic spectrum of diseases known as L1 syndromes, which include X-linked hydrocephalus with stenosis of the aqueduct of Sylvius (HSAS), MASA syndrome (mental retardation, aphasia, shuffling gait, and adducted thumbs), SPG1 (X-linked complicated hereditary spastic paraplegia type 1), and X-linked complicated corpus callosum agenesis. EGL Genetics offers both full gene sequencing and deletion/duplication analyses when there is a prenatal indication of an L1 syndrome.

Indications:
- Ultrasound abnormalities suggestive of hydrocephalus or an L1 syndrome
- Family history of an L1 syndrome

Prenatal Testing Sample Requirements
EGL Genetics accepts direct or cultured chorionic villus and amniocentesis samples. Backup cultures must always be maintained either at EGL Genetics or the referring lab until testing is complete. Please note that a maternal blood sample (EDTA/purple top) MUST be included for maternal cell contamination studies.

Please be sure to consult a laboratory genetic counselor before ordering a molecular prenatal test.

For more information about EGL Genetics:

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