Baylor College of Medicine

PRENATAL TESTS

Medical Genetics Laboratories

INFORMATION FOR MEDICAL PROFESSIONALS

Prenatal Tests Offered

The Medical Genetics Laboratories perform a full range of genetic testing for prenatal diagnosis and parental screening assays, including cytogenetic analyses and multiple biochemical and DNA-based studies, listed below. The laboratory staffs four certified genetic counselors available for consultation prior to submitting prenatal cases for genetic testing. This consultation process insures that appropriate samples are submitted to complete prenatal diagnosis as rapidly and comprehensively as possible. Our specimen requirements and quality control measures are compliant with American College of Medical Genetics (ACMG) Standards and Guidelines for Clinical Genetics Laboratories.

Available Cytogenetic Testing:

High Resolution Chromosomes with AFP and ACHE (if indicated)
Chromosomal Microarray Analysis (CMA) Aneuploidy FISH

CMA-TARGETED

This Prenatal array is very similar to what was used in the NIH trial and is ideal for physicians, counselors and patients who want detection of all well characterized, deletion and duplication syndromes. It also includes additional deletion/duplication syndromes that were not yet characterized when the NIH array was designed.This array includes 180,000 oligonucleotides, and SNP probes targeted for chromosomes 6, 7, 11, 14, 15, and 20 for detection of uniparental disomy (UPD).

CMA-TARGETED + LIMITED KARYOTYPE ANALYSIS

This option includes the Limited array as described above plus a 5 cell chromosome analysis to rule out rearrangements not detected by microarray such as balanced translocations, inversions and triploidy. The benefits of the limited karytoype are lower cost and a quicker turn around time.

CMA –EXPANDED

Baylor MGL also offers an EXPANDED prenatal array. This array offers exon by exon coverage for over 1700 genes. In Baylor MGL's experience, this array does not detect significantly more cases of uncertain significance that lead to difficult counseling situations. The EXPANDED array is recommended for families who want the highest level of analysis possible (understanding no single test can assess for every possible genetic outcome).
This array includes 400,000 oligonucleotides and SNP probes for all chromosomes.

CMA-EXPANDED+LIMITED KARYOTYPE ANALYSIS

This option includes the Expanded array as described above, plus a 5 cell chromosome analysis to rule out rearrangements not detected by microarray such as balanced translocations, inversions and triploidy. The benefits of the limited karytoype are lower cost and a quicker turn around time.

PARENTAL CARRIER SCREENING TESTS:

General Population: CF and Fragile X
Jewish Genetic Diseases
Family History: CF and below when known familial mutation
Universal Carrier Screening

Notice for Prenatal Biochemical and DNA Tests

Please be aware that our specimen requirements and quality control measures are compliant with American College of Medical Genetics (ACMG) Standards and Guidelines for Clinical Genetics Laboratories. While these requirements are intended to provide the highest level of assurance that a single laboratory can offer, the ideal practice to assure the accuracy of prenatal diagnostic testing is through duplicate testing conducted by independent laboratories. We recommend that referring medical professionals make the necessary arrangements for these two independent analyses for their patients prior to performing the prenatal diagnostic procedure.

Turnaround Time Notice

Samples received after 3 pm may be subject to an increase in turnaround time.

Reflex Policy

The following will be performed by reflex at no additional charge; AChE when AF-AFP is elevated; Fetal HGB when AF-AFP is elevated and amniotic fluid is bloody; CF5T when R117H CF mutation is present.

Notice: *The laboratory provides highly trained and knowledgeable genetic councelsors. Please contact one of our councelors prior to ordering specific disorder testing, to discuss clinical and sample requirements.*

Specific Disorder Tests Available

- ❒ ad-PEO 2 ANT1/SLC25A4
- ❒ ad-PEO 3 TWINKLE/PEO1
- ❒ Adenosine Deaminase Deficiency
- ❒ Angelman Syndrome UBE3A
- ❒ APECED AIRE
- ❒ Argininosuccinic Aciduria
- ❒ Arginosuccinic Aciduria ASL
- ❒ ARX Related Disorders
- ❒ BCS1L Related Complex III Deficiency BCS1L
- ❒ Carbamoyl Phosphate Synthetase I Deficiency CPS1
- ❒ Cartilage Hair Hypoplasia RMRP
- ❒ CDKL5 Related Atypical Rett Syndrome CDKL5/STK9
- ❒ Charge Syndrome CHD7
- ❒ Citrin Deficiency SLC25A13
- ❒ Citrullinemia I ASS
- ❒ Cleidocranial Dysplasia RUNX2
- ❒ Coenzyme Q10 Deficiency COQ2
- ❒ Coenzyme Q10 Deficiency PDSS2
- ❒ COX10
- ❒ Cystic Fibrosis CFTR
- ❒ DGUOK (Deoxyguanosine Kinase)
- ❒ Diamond-Blackfan Anemia RPS19
- ❒ Fabry Disease GLA
- ❒ Familial Adenomatous Polyposis APC
- ❒ Fatal Infantile Lactic Acidosis w/mtDNA Depletion SUCLG1
- ❒ Focal Dermal Hypoplasia PORCN
- ❒ Fragile X Syndrome
- ❒ Guanidinoacetate Methyltransferase Deficiency GAMT
- ❒ Hereditary Fructose Intolerance ALDOB
- ❒ Huntington Disease
- ❒ Incontinentia Pigmentia
- ❒ L-Arginine:Glycine Amidinotransferase Deficiency GATM
- ❒ Lesch-Nyhan Disease HPRT1
- ❒ Leukoencephalopathy VWM EIF2B5
- ❒ Lowe Syndrome OCRL1
- ❒ MNGIE Syndrome (Thymidine Phosphorylase) TP
- ❒ MPV17
- ❒ Myotonic Dystrophy
- ❒ Nail-Patella Syndrome LMX1B
- ❒ Noonan Syndrome PTPN11
- ❒ X-linked Ichthyosis (STS Deficiency)
- ❒ Optic atrophy type 1 OPA1
- ❒ Ornithine Transcarbamylase (OTC) Deficiency OTC
- ❒ Osteogenesis Imperfecta, AR CRTAP
- ❒ Osteogenesis Imperfecta, AR LEPRE1
- ❒ Pelizaeus-Merzbacher PLP1
- ❒ POLG1 Related Disorders
- ❒ Purine Nucleoside Phosphorylase Deficiency
- ❒ Pyruvate Dehydrogenase Deficiency PDHA1
- ❒ Rett Syndrome MECP2
- ❒ RHD Molecular Typing
- ❒ Rothmund-Thomson Syndrome RECQL4
- ❒ SCO1
- ❒ SCO2
- ❒ SMCD COL10A1
- ❒ Spinocerebellar Ataxia 10 (SCA10)
- ❒ SURF1
- ❒ Thymidine Kinase TK2
- ❒ Wolman Disease
- ❒ X-linked Ocular Albinism GPR143