

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

Parental Carrier Screening Tests:

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

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

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

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.