

Mount Sinai Genetic Testing Laboratories Clinical Tests

Molecular Genetics

Test Name	Gene / mutations
Ashkenazi Jewish Panel	
Bloom syndrome	BLM (1 mutation)
Canavan disease	ASPA (4 mutations)
Cystic Fibrosis	CFTR (71 mutations)
Familial dysautonomia	IKBKAP (2 mutations)
Familial hyperinsulinism	ABCC8 (2 mutations)
Fanconi anemia	FANCC (2 mutations)
Gaucher disease	GBA (8 mutations)
Glycogen Storage Ia	G6PC (2 mutations)
LAD deficiency (E3)	LAD (2 mutations)
Maple Syrup Urine Disease	BCKHDB (3 mutations)
Mucopolidosis IV	MCOLN1 (2 mutations)
Nemaline myopathy	NEB (1 mutation)
Niemann-Pick	SMPD1 (4 mutations)
Tay-Sachs disease	HEXA (7 mutations)
Usher type III	USH3A (1 mutation)
Usher type I	PCDH15 (1 mutation)
Fragile X	
Noonan syndrome, blood	PTPN11 (Exons 3 and 8, more if nec.)
Noonan syndrome, prenatal	PTPN11 (Exons 3 and 8, more if nec.)
Y Microdeletion	
Maternal Cell Contamination Studies	
Warfarin Genotyping	
Cytochrome P450 (Tamoxifen) Genotyping	CYP2C19 (7 mutations) / CYP2D6 (17 mutations)

Cytogenetics

Test Name

Chromosome Analysis - Amnio
Chromosome Analysis - CVS
Chromosome Analysis - Peripheral Blood
Stat Chromosome Analysis - Peripheral Blood
Chromosome Analysis - Skin biopsy
Chromosome Analysis - Products of Conception
Fetal blood
High Resolution blood
Mosaicism Study, Peripheral Blood

Molecular Cytogenetics

Test Name

Region

FISH - Aneuploidy	
FISH - Chromosome Analysis	
Aneuvision / STAT FISH	chromosomes 13, 21, 18, X and Y
FISH - Microdeletion/Microduplication	
Single Probe FISH - Microdeletion	
SR Y	Yp11.3

FISH - Microdeletion Panel (8 Syndromes)	
DiGeorge syndrome	22q11.2 deletion
Williams syndrome	7q11.23 deletion
Cri du Chat syndrome	5p15.2 deletion
Wolf-Hirschhorn syndrome	4p16.3 deletion
Smith-Magenis syndrome	17p11.2 deletion
Miller-Dieker syndrome	17p13.3 deletion
Prader-Willi syndrome	15q12 deletion
Angelman syndrome	15q12 deletion

Marker chromosome identification using C-banding and chromosome specific FISH centromere panel (CEP probes).

Biochemical Genetics

Disease/Syndrome	Test	Notes
Gaucher Disease	Acid Beta-Glucosidase enzyme analysis	Affected Status only (no carrier detection by enzyme testing)
Gaucher Disease - biomarker	Chitotriosidase analyses	Enzyme and DNA analyses (common dup24 allele)
Fabry Disease	Alpha-Galactosidase A	Enzyme analysis
Schindler Disease	Alpha-Galactosidase B	Enzyme analysis
Tay-Sachs Disease	Hexosaminidase A	Enzyme analysis (carrier screening and prenatal analysis)
Sandhoff Disease	Hexosaminidase A and B	Enzyme analysis
Amino Acids (quantitative)		Plasma
	Full Profile	
	Branched chain amino acids	
	PKU panel: Phenylalanine and tyrosine	
Organic Acid analysis (qualitative)		Urine, plasma
Organic Acid analysis (quantitative)		Urine, plasma
	Methylmalonic Acid (MMA)	
	Orotic Acid	
	Succinylacetone	

Porphyria Testing Laboratory

Test Name	Gene
Total Porphyria Panel (AIP, HCP and VP)	
Acute Intermittent Porphyria (AIP)	HMBS
Hereditary Coproporphyrin (HCP)	CPOX
Variegate Porphyria (VP)	PPOX
Congenital Erythropoietic Porphyria (CEP)	UROS
Familial Porphyria Cutanea Tarda (fPCT)	UROD
Erythropoietic Protoporphyrin (EPP)	FECH

Lysosomal Diagnostic Laboratory

Test	Gene	Notes
Fabry Disease	<i>GLA</i>	Sequence analysis, enzyme assay
Gaucher Disease	<i>GBA</i>	Targeted mutation analysis, enzyme assay
Gaucher Disease biomarker (Chitotriosidase)	<i>CHIT1</i>	Enzyme assay
Hexosaminidase A deficiency	<i>HEXA</i>	Targeted mutation analysis, enzyme assay
Sandhoff Disease	<i>HEXB</i>	Enzyme assay
Schindler Disease	<i>NAGA</i>	Sequence analysis, enzyme assay