

BIOCHEMICAL GENETICS LABORATORY

Acylcarnitines
Creatine/Guadinoacetate
Free and total Carnitine
Orotic Acid

DNA DIAGNOSTIC LABORATORY

Achondroplasia	Huntington Disease
APC I1307K	Incontinentia Pigmenti
ARX Gene	Kennedy Disease
Bloom	Lowe Syndrome
Canavan	MTHFR
CFTR Poly T	Myotonic Dystrophy
CIT	Niemann-Pick Disease A
Cystic fibrosis (51 mutations)	Pelizaeus-Merzbacher disease (PLP1 gene)
DRPLA	Prader-Willi
Duchenne-Becker muscular dystrophy	Prothrombin
Fabry disease (GLA gene)	PTPN11 Sequence Analysis
factor V Leiden	Rett Syndrome
Familial dysautonomia	RhD testing
Fanconi anemia gaa	Sickle Cell disease
Fragile X	Spinal Muscular Atrophy
Gaucher Disease	Spinocerebellar Ataxia (SCA) 1 and 10
GJB2 (Connexin-26)	SRY
Hemochromatosis	Tay-Sachs disease
HNPCC (MLH1, MSH2 and MHS 6)	X-Linked ocular albinism