

GENETIC TESTING

The *IBR Specialty Clinical Laboratories (SCL)* provide a wide spectrum of laboratory tests for the accurate diagnosis of lysosomal storage diseases and carbohydrate, amino acid, organic acid and peroxisomal disorders.

Lysosomal Storage Disorders

CLN2-Late Infantile Neuronal

Ceroid Lipofuscinosis

Fabry

Fucosidosis

Gaucher

GM1 Gangliosidosis

Hurler/Scheie (MPSI)

Krabbe

Mannosidosis

Maroteaux-Lamy (MPSVI)

Metachromatic Leukodystrophy

Mucopolidosis I

Mucopolidosis II, III

Niemann-Pick

Pompe (Skin only)

Sandhoff

Sanfilippo (MPS IIIB)

Sly (MPS VII)

Tay-Sachs

Ganglioside Profile

Amino Acid Disorders

Metabolic Screen

Qualitative Analysis, Urine/Plasma

Quantitative Analysis, Urine/Plasma

Organic Acid Disorders

Organic Acid GCMS Identification

Carbohydrate Disorders

Monosaccharides

Mucopolysaccharide Spot Test

Mucopolysaccharide Quantitation

Mucopolysaccharide Identification

Oligosaccharides

Other (Methylation Testing for Prader-Willi Syndrome, Angelman Syndrome, Fragile X)

Galactose-1-phosphate Uridyltransferase

Tripeptide Peptidase I

Expertise

SCL's Genetic Testing Laboratories are directed by an American Board of Medical Genetics-certified biochemical geneticist. The SCL is fully certified by the New York State Department of Health and CLIA. We participate in proficiency testing of the College of American Pathology and the National Tay-Sachs and Allied Diseases Foundation.

Interpretative Reporting/Consultations

Consultation support is provided to determine the most appropriate tests for the

patient and the family. Reports are accompanied by interpretation and suggestions for further testing to confirm suspected diagnoses.

Technical Information

A complete manual of test requirements is available by phoning 718-494-5345 or by writing to:

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