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
- Mucolipidosis I
- Mucolipidosis 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
- Tripeptidyl 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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