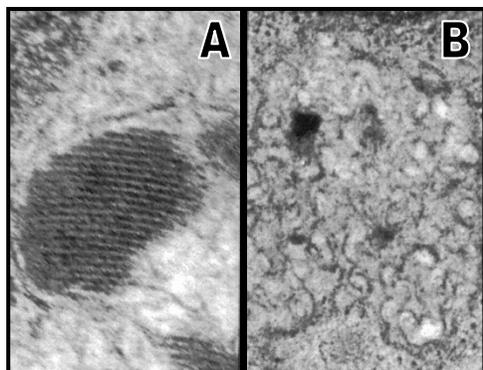


ULTRASTRUCTURAL TESTING

The *IBR Specialty Clinical Laboratories (SCL)* Electron Microscopy Laboratory offers ultrastructural studies for the detection of lysosomal storage disease caused by different genetic disorders. These studies are a valuable diagnostic tool for patients whose enzymatic defects are poorly understood and in cases in which the disease has not been demonstrated by biochemical or molecular genetic means or in which it has been demonstrated but the usual clinical presentation made the diagnosis difficult. Testing for some neurogenetic disorders include (e.g.):



A. Fingerprint (122,000x)
B. Curvilinear Body (34,000x)

Sphingolipidosis
Glycosphingolipidosis
Glycogen Storage Disorders
Mucopolysaccharidosis
Mucopolysaccharidosis
Neuroaxonal Dystrophy
Neuronal Ceroid Lipofuscinosis (NCL)
 Infantile, Late Infantile, Juvenile, Adult form
Leukodystrophies
Peroxisomal Disorders
Mitochondrial Diseases
Neuropathies

Specimen Types

Electron microscopy studies can be performed on lymphocytes (buffy coat) and skin, conjunctiva, rectal and nerve biopsies.

Expertise

SCL's Electron Microscopy Laboratory is directed by a Board-certified pathologist/neuropathologist and a pediatric neurologist.

The SCL is licensed by the New York State Department of Health and is CLIA-certified.

Interpretative Reporting/Consultations

Consultation support is provided to determine the most appropriate tests for the patient. Reports include interpretation and suggestions for further testing. Electron micrographs accompany all positive reports.

Technical Information

Carolyn Salafia, M.D, is available for consultations at 718-494-5202 or the lab at 718-494-5213. See the reverse side for specimen requirements and billing information.

IBR Specialty Clinical Laboratories

NYS Office for People With Developmental Disabilities
Institute for Basic Research in Developmental Disabilities
1050 Forest Hill Road, Staten Island, New York 10314

SPECIALTY CLINICAL LABORATORIES

TEST REQUIREMENTS

TEST:	Electron Microscopy	(Ultrastructural Study) (Lysosomal Storage Diseases)
Specimen(s):	Lymphocytes -5 - 10 ml heparinized whole blood in a green-top tube.	
	<i>or</i>	
	Skin Biopsy -	3-mm skin biopsy immediately immersed in 2.5% glutaraldehyde prepared in 0.1M phosphate buffer. After two hours of glutaraldehyde fixation, change to 0.1M phosphate buffer.
	<i>or</i>	
	Tissue -	Conjunctiva, rectal, nerve in 2.5% buffered glutaraldehyde - see directions for skin biopsy, above.
Special Requirements:		Submit at room temperature within 24 hours. Protect from temperature extremes. Submit clinical summary.
Reference Range:		See laboratory report. Electron micrographs accompany all positive studies.
Methodology:		Electron microscopy
Turnaround Time:		6 weeks
Price:		\$510.00
CPT Code:		88348

Our Laboratory regularly bills the facility sending the samples. Please have your laboratory send the specimens with the following information:

Patient Information:

Patient Name
Date of Birth
Clinical Summary
Patient ID Number

Specimen Information:

Specimen Type
Specimen Collection Date
Specimen ID Number

Billing Information:

Facility Name
Report Address
Billing Address
Phone Number
Fax Number
Billing Authorization

NY #7097M040 • C.L.I.A. #33D0860102

SPECIALTY CLINICAL LABORATORIES

