GM_2 Gangliosidoses										
Tay-Sachs disease (153)	β-Hexosaminidase A	GM_2 gangliosides	Infantile Juvenile	AR	Mental retardation; seizures; later juvenile form	None	None	Cherry red spot in infantile form	None	Macrocephaly; hyperacusis in infantile form
Sandhoff disease (153)	β-Hexosaminidases A and B	GM_2 gangliosides	Infantile	AR	Mental retardation; seizures	++	±	Cherry red spot	None	Macrocephaly; hyperacusis
Neutral Glycosphingolipidoses										
Fabry disease (150)	α-Galactosidase A [ET]	Globotriaosylceramide	Childhood	X-linked	Painful acroparesthesias	None	None	Corneal dystrophy, vascular lesions	None	Cutaneous angiokeratomas; hypo-hydrosis
Gaucher disease (146)	Acid β-glucosidase [ET, SRT]	Glucosylceramide	Type 1 Type 2 Type 3	AR	None ++++ ++	++++ +++ ++++	++++ + ++++	None Eye movements Eye movements	Gaucher cells in bone marrow; cytopenias	Adult form highly variable
Niemann-Pick disease (144) A and B	Sphingomyelinase [ET—trials]	Sphingomyelin	Neuronopathic, type A Nonneuronopathic, type B	AR	Mental retardation; seizures	++++	None Osteoporosis	Macular degeneration	Foam cells in bone marrow	Pulmonary infiltrates Lung failure
Glycoproteinoses										
Fucosidosis (140)	α-Fucosidase	Glycopeptides; oligosaccharides	Infantile Juvenile	AR	Mental retardation	++	++	None	Vacuolated lymphocytes; foam cells	Coarse facies; angiokeratomas in juvenile form
α-Mannosidosis (140)	α-Mannosidase	Oligosaccharides	Infantile Milder variant	AR	Mental retardation	+++	+++	Cataracts, corneal clouding	Vacuolated lymphocytes, granulated neutrophils	Coarse facies; enlarged tongue
β-Mannosidosis (140)	β-Mannosidase	Oligosaccharides		AR	Seizures; mental retardation		++	None	Vacuolated lymphocytes, foam cells	Angiokeratomas
Aspartylglucosaminuria (141)	Aspartylglucosaminidase	Aspartylglucosamine; glycopeptides	Young adult	AR	Mental retardation	±	++	None	Vacuolated lymphocytes, foam cells	Coarse facies
Sialidosis (140)	Neuraminidase	Sialyloligosaccharides	Type I, congenital Type II, infantile and juvenile	AR	Myoclonus; mental retardation	++, less in type I	++, less in type I	Cherry red spot	Vacuolated lymphocytes	MPS phenotype in type II
Mucolipidoses (ML)										
ML-II, I-cell disease (138)	UDP-*N*-Acetylglucosamine-1-phosphotransferase	Glycoprotein; glycolipids	Infantile	AR	Mental retardation	+	++++	Corneal clouding	Vacuolated and granulated neutrophils	Coarse facies; absence of mucopolysaccharidiuria; gingival hypoplasia
ML-III, pseudo-Hurler polydystrophy (138)	UDP-*N*-Acetylglucosamine-1-phosphotransferase	Glycoprotein; glycolipids	Late infantile	AR	Mild mental retardation	None	+++	Corneal clouding, mild retinopathy, hyperopic astigmatism		Coarse facies; stiffness of hands and shoulders

(*Continued*)