

TABLE 432e-1 SELECTED LYSOSOMAL STORAGE DISEASES

Disorder ^a	Enzyme Deficiency [Specific Therapy]	Stored Material	Clinical Types (Onset)	Inheritance	Clinical Features					
					Neurologic	Liver, Spleen Enlargement	Skeletal Dysplasia	Ophthalmologic	Hematologic	Unique Features
Mucopolysaccharidoses CTMucopolysaccharidoses (MPS)										
MPS I H, Hurler (136)	α -L-Iduronidase [ET, BMT]	Dermatan sulfate Heparan sulfate	Infantile Intermediate	AR	Mental retardation	+++	++++	Corneal clouding	Vacuolated lymphocytes	Coarse facies; cardiovascular involvement; joint stiffness
MPS I H/S, Hurler/ Scheie			Adult		Mental retardation					
MPS I S, Scheie					None					
MPS II, Hunter (136)	Iduronate sulfatase [ET]	Dermatan sulfate Heparan sulfate	Severe infantile Mild juvenile	X-linked	Mental retardation, less in mild form	+++	++++	Retinal degeneration, no corneal clouding	Granulated lymphocytes	Coarse facies; cardiovascular involvement; joint stiffness; distinctive pebbly skin lesions
MPS III A, Sanfilippo A (136)	Heparan-N-sulfatase	Heparan sulfate	Late infantile	AR	Severe mental retardation	+	+	None	Granulated lymphocytes	Mild coarse facies
MPS III B, Sanfilippo B (136)	N-Acetyl- α -glucosaminidase	Heparan sulfate	Late infantile	AR	Severe mental retardation	+	+	None	Granulated lymphocytes	Mild coarse facies
MPS III C, Sanfilippo C (136)	Acetyl-CoA: α -glucosaminide N-acetyltransferase	Heparan sulfate	Late infantile	AR	Severe mental retardation	+	+	None	Granulated lymphocytes	Mild coarse facies
MPS III D, Sanfilippo D (136)	N-Acetylglucosamine- 6-sulfate sulfatase	Heparan sulfate	Late infantile	AR	Severe mental retardation	+	+	None	Granulated lymphocytes	Mild coarse facies
MPS IV A, Morquio A (136)	N-Acetylgalactosamine- 6-sulfate sulfatase [ET—trials]	Keratan sulfate Chondroitin-6 sulfate	Childhood	AR	None	+	++++	Corneal clouding	Granulated neutrophils	Distinctive skeletal deformity; odontoid hypoplasia; aortic valve disease
MPS IV B, Morquio (136)	β -Galactosidase		Childhood	AR	None	\pm	++++			
MPS VI, Maroteaux- Lamy (136)	Arylsulfatase B [ET, BMT]	Dermatan sulfate	Late infantile	AR	None	++	++++	Corneal clouding	Granulated neutrophils and lymphocytes	Coarse facies; valvular heart disease
MPS VII (136)	β -Glucuronidase	Dermatan sulfate Heparan sulfate	Neonatal Infantile Adult	AR	Mental retardation, absent in some adults	+++	+++	Corneal clouding	Granulated neutrophils	Coarse facies; vascular involvement; hydrops fetalis in neonatal form