TABLE 432e-1 SELECTED LYSOSOMAL STORAGE DISEASES (*CONTINUED*)

Disorder[a]	Enzyme Deficiency [Specific Therapy]	Stored Material	Clinical Types (Onset)	Inheritance	Clinical Features					
					Neurologic	Liver, Spleen Enlargement	Skeletal Dysplasia	Ophthalmologic	Hematologic	Unique Features
Leukodystrophies										
Krabbe disease (147)	Galactosylceramidase [BMT/HSCT]	Galactosylceramide Galactosylsphingosine	Infantile	AR	Mental retardation	None	None	None	None	White matter globoid cells
Metachromatic leukodystrophy (148)	Arylsulfatase A	Cerebroside sulfate	Infantile Juvenile Adult	AR	Mental retardation; dementia; psychosis in adult	None	None	Optic atrophy	None	Gait abnormalities in late infantile form
Multiple sulfatase deficiency (149)	Active site cysteine to C_{α}-formylglycine-converting enzyme	Sulfatides; mucopoly-saccharides	Late infantile	AR	Mental retardation	+	++	Retinal degeneration	Vacuolated and granu-lated cells	Absent activity of all known cellular sulfatases
Disorders of Neutral Lipids										
Wolman disease (142)	Acid lysosomal lipase [ET—trials]	Cholesteryl esters; triglycerides	Infantile	AR	Mild mental retardation	+++	None	None	None	Adrenal calcification
Cholesteryl ester storage disease (142)	Acid lysosomal lipase [ET—trials]	Cholesteryl esters	Childhood	AR	None	Hepatomegaly	None	None	None	Fatty liver disease; cirrhosis
Farber disease (143)	Acid ceramidase	Ceramide	Infantile Juvenile	AR	Occasional mental retardation	±	None	Macular degeneration	None	Arthropathy, subcutaneous nodules
Disorders of Glycogen										
Pompe disease (135)	Acid α-glucosidase [ET]	Glycogen	Infantile, late onset	AR	Neuromuscular	±	None	None	None	Myocardiopathy
Late -onset GAA deficiency (135)	Acid α-glucosidase [ET]	Glycogen	Variable: juvenile to adulthood	AR	Neuromuscular	None	None	None	None	Respiratory insufficiency; neuromuscular disease
Danon disease (154)	LAMP-2 (lysosomal associated membrane protein-2)	Glycogen	Variable: childhood to adulthood	X-linked (?Dominant)	Cardiomyopathy Neuromuscular Inconsistent mental retardation	None	None	None	None	Myocardial vacuolar degeneration

[a]Numbers in parentheses refer to the chapters in CR Scriver et al: *The Metabolic and Molecular Bases of Inherited Disease,* 9th ed. New York, McGraw-Hill, *www.ommbid.com*, which provide comprehensive reviews.

Abbreviations: AR, autosomal recessive; BMT/HSCT, bone marrow or stem cell transplantation; ET, enzyme therapy; SRT, substrate reduction therapy.