

Lysosomal Storage Disorders

At Healthfirst, we are committed to helping providers accurately document and code their patients' health records. This tip sheet is intended to assist providers and coding staff with the documentation and ICD-10-CM selection, on services submitted to Healthfirst—specifically for **Lysosomal Storage Disorders**. It provides information from industry sources about proper coding practice. However, this document does not represent or guarantee that Healthfirst will cover and/or pay for the services outlined. Coverage decisions are based on the terms of the applicable evidence of coverage and the provider's participation agreement. This includes the determination of any amount that Healthfirst or the member owes the provider.

Lysosomal Storage Disorders are a group of rare, inherited metabolic diseases caused by genetic mutations that result in enzyme deficiencies within cell lysosomes. When an enzyme is missing or dysfunctional, fats, sugars, proteins, and other waste materials accumulate, causing toxic buildup. This leads to cell, tissue, and organ damage.

ICD-10 Codes and Descriptions (Most commonly used)

E74.02	Pompe disease Cardiac glycogenosis Type II glycogen storage disease
E74.05	Lysosome-associated membrane protein 2 [LAMP2] deficiency Danon disease
E75.00	GM2 gangliosidosis, unspecified
E75.01	Sandhoff disease
E75.02	Tay-Sachs disease
E75.09	Other GM2 gangliosidosis Adult GM2 gangliosidosis Juvenile GM2 gangliosidosis

Documentation and Coding

ICD-10 Codes and Descriptions (Most commonly used) (cont.)

E75.10	Unspecified gangliosidosis Gangliosidosis NOS
E75.11	Mucopolipidosis IV
E75.19	Other gangliosidosis GM1 gangliosidosis GM3 gangliosidosis
E75.21	Fabry (-Anderson) disease
E75.22	Gaucher disease
E75.23	Krabbe disease
E75.240	Niemann-Pick disease type A Acid sphingomyelinase deficiency type A (ASMD type A) Infantile neurovisceral acid sphingomyelinase deficiency
E75.241	Niemann-Pick disease type B Acid sphingomyelinase deficiency type B (ASMD type B) Chronic visceral acid sphingomyelinase deficiency
E75.242	Niemann-Pick disease type C
E75.243	Niemann-Pick disease type D
E75.25	Metachromatic leukodystrophy
E75.26	Sulfatase deficiency Multiple sulfatase deficiency (MSD)
E75.29	Other sphingolipidosis Farber's syndrome Sulfatide lipidosis

ICD-10 Codes and Descriptions (Most commonly used) (cont.)

E76.01	Hurler's syndrome
E76.02	Hurler-Scheie syndrome
E76.03	Scheie's syndrome
E76.1	Mucopolysaccharidosis, type II Hunter's syndrome
E76.22	Sanfilippo mucopolysaccharidoses Mucopolysaccharidosis, type III (A) (B) (C) (D) Sanfilippo A syndrome Sanfilippo B syndrome Sanfilippo C syndrome Sanfilippo D syndrome
E76.210	Morquio A mucopolysaccharidoses Classic Morquio syndrome Morquio syndrome A Mucopolysaccharidosis, type IVA
E76.211	Morquio B mucopolysaccharidoses Morquio syndrome B Morquio-like mucopolysaccharidoses Morquio-like syndrome Mucopolysaccharidosis, type IVB
E76.29	Other mucopolysaccharidoses beta-Glucuronidase deficiency Maroteaux-Lamy (mild) (severe) syndrome Mucopolysaccharidosis, types VI, VII

Documentation and Coding

ICD-10 Codes and Descriptions (Most commonly used) (cont.)

E77.0	Defects in post-translational modification of lysosomal enzymes Mucopolipidosis II [I-cell disease] Mucopolipidosis III [pseudo-Hurler polydystrophy]
E77.1	Defects in glycoprotein degradation Aspartylglucosaminuria Fucosidosis Mannosidosis Sialidosis [mucopolipidosis I]
E77.8	Other disorders of glycoprotein metabolism

Type	Deficiency in enzymes needed to break down
Sphingolipidoses	Fat Molecules called Sphingolipids
Mucopolysaccharidoses	Sugar Molecules called Glycosaminoglycans (GAGs)
Glycoproteinoses	Protein Molecules called Glycoproteins
Clinical Documentation Recommendations	
Status of Condition	Stable, Improved, or Worsening
Signs & Symptoms	<ul style="list-style-type: none"> • Coarse Facial Features, • Short Stature, and Failure to Thrive in Infants • Developmental Delays, Intellectual Disability, • Seizures, Hypotonia, Joint Pain, Stiffness, Skeletal Deformities, Bone Fractures • Vision Issues, Hearing Loss, Easy Bruising, and Anemia
Diagnostic Test	<ul style="list-style-type: none"> • Enzyme Activity Assay • Genetic Testing • Substrate Analysis • Imaging: X-rays or MRIs • Tissue Biopsy
Complications	<ul style="list-style-type: none"> • Hepatomegaly or Splenomegaly • Chronic Kidney Disease • Respiratory Failure • Arrhythmias • Heart Failure, Heart Valve Disease

Documentation and Coding

Clinical Documentation Recommendations

Treatment	<ul style="list-style-type: none">• Enzyme Replacement Therapy (ERT)• Substrate Reduction Therapy (SRT)• Hematopoietic Stem Cell Transplantation (HSCT)• Gene Therapy
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Coding Tips

- For Lysosome-associated membrane protein 2 [LAMP2] deficiency/ Danon disease, if applicable code also associated manifestations such as:
 - Dilated cardiomyopathy (**I42.0**)
 - Obstructive hypertrophic cardiomyopathy (**I42.1**)

Coding Example

Case 1	A 23-month-old male child of Ashkenazi Jewish descent presents for a follow up visit with a history of progressive developmental regression, losing the ability to sit independently, following a 5-month history of worsening muscle weakness and a diagnosis of Tay-Sachs disease . Physical exam shows poor eye contact, an exaggerated startle response to sound and hypotonia. Ophthalmology reveals pathognomonic bilateral cherry-red spots.
ICD-10-CM Codes	E75.02 - Tay-Sachs disease

References

- [2026 ICD-10-CM Coding Guidelines](#)
- [Codingclinicadvisor.com](#)
- [AAPC.com](#)
- [Cleveland Clinic - lysosomal-storage-diseases](#)
- [Cleveland Clinic - tay-sachs-disease](#)
- [ncbi.nlm.nih.gov - lysosomal-storage-diseases](#)

Questions?

Contact us at [#Risk Adjustments and clinical Documentation@healthfirst.org](#).

For additional documentation and coding guidance, please visit the coding section on [HFproviders.org](#)