DISEASE	AFFECTED PATHWAY	CLINICAL EXAMPLE
SPHINGOLIPIDOSES	Degradation of ceramide-containing membrane lipids	Gaucher disease, Fabry disease, Niemann-Pick A/B
MUCOPOLYSACCHARIDOSES	Glycosaminoglycane degradation	MPS type I (Hurler disease)
OLIGOSACCHARIDOSES	Degradation of complex carbohydrate side-chains of glycoproteins	a -Mannosidasis
MUCOLIPIDOSES	Deficiency of several lysosomal enzymes, many pathways affected	Mucolipidos type II
LIPID STORAGE DISEASE	Degradation of lipid compounds other than sphingolipids	Cholesterol ester storage disease
GLYCOGEN STORAGE DISEASE TYPE II	Intralysosomal glycogen breakdown due to deficiency of lysosomal acid maltase	Pompe disease
LYSOSOMAL TRANSPORT DEFECTS	Failure to transport certain compounds across the lysosomal membrane	Sialic acid transport defect

Tabella 1. Classificazione delle malattie da accumulo lisosomiale (*Modified according to van Dahl S, Mengel E-2010)*

Disease	Incidence	Prevalence
Cystinosis	1/281,000	1/192,000
Anderson–Fabry disease	1/117,000	1/117,000
Gaucher disease	1/59,000	1/57,000
GM_1 gangliosidosis	1/422,000	1/384,000
Krabbe disease	1/201,000	1/141,000
α-mannosidosis	1/1,056,000	1/1,056,000
MPS I	1/111,000	1/88,000
MPS II	1/162,000	1/136,000
ASMD	1/264,000	1/248,000
Niemann–Pick C	1/211,000	1/211,000
Pompe disease	1/201,000	1/146,000
Sandhoff disease	1/422,000	1/384,000
Tay-Sachs disease	1/222,000	1/201,000
Wolman disease	1/704,000	1/528,000
All LSDs	1/9000	1/7700

Legenda: ASMD, acid sphingomyelinase deficiency (formerly Niemann–Pick A/B); LSD, lysosomal storage disorder; MPS, mucopolysaccharidosis

Tabella 2. Incidenza e prevalenza delle malattie da accumulo lisosomiale (adapted from Meikle PJ, et al. 1999)

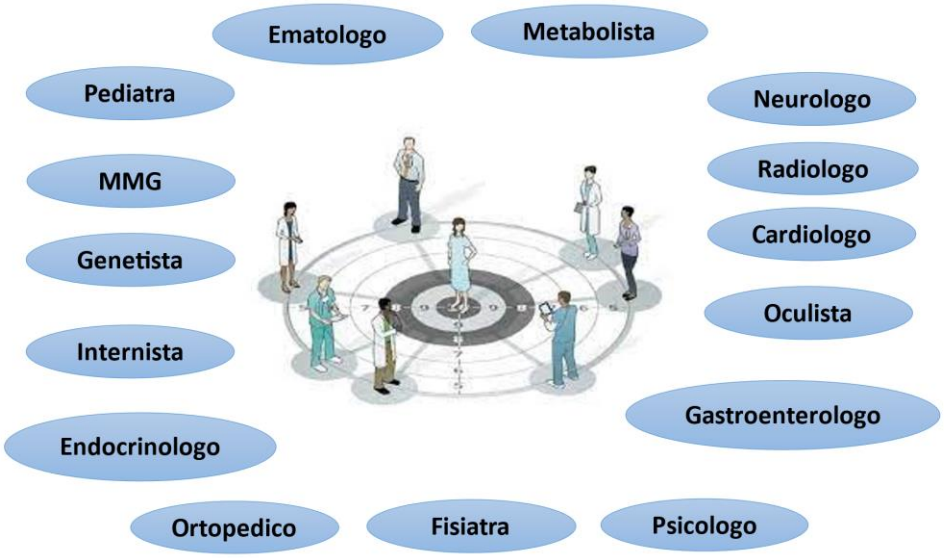

Figura 1. La gestione multidisciplinare delle malattie da accumulo lisosomiale