

Las principales manifestaciones musculoesqueléticas de MPS por tipo de trastorno.¹

OMIM	Enfermedad	Enzima deficiente	Principales sustancias acumuladas
607014	MPS I (Hurler, Scheie, Hurler/Scheie)	α -L-iduronidase	Dermatan sulfato, heparan sulfato
607015			
607016			
309900	MPS II (Hunter)	Iduronate 2-sulfatase	Dermatan sulfato, heparan sulfato
252900	MPS IIIA (San lippo A)	Heparan N-sulfatase	heparan sulfato
252920	MPS IIIB (San lippo B)	α -N-acetilglucosaminidase	heparan sulfato
252930	MPS IIIC (San lippo C)	Acetyl CoA: α -glucosaminidase N-acetyltransferase	heparan sulfato
252940	MPS IID (San lippo D)	N-acetylglucosamine-6-sulfatase	Queratan sulfato, condroitin 6 sulfato
253000	MPS IVA (Morquio A)	N-acetylgalactosamine-6-sulfatase	Queratan sulfato, condroitin 6 sulfato
253010	MPS IVB (Morquio B)	β -galactosidase	Queratan sulfato
253200	MPS VI (Maroteaux–Lamy)	N-acetyl galactosamine 4-sulfatase (arylsulfatase B)	Dermatan sulfato
253220	MPS VII (Sly)	β -glucuronidase	Dermatan sulfato, heparan sulfato, condroitin 6 sulfato
601492	MPS IX (Natowicz)	Hialuronidase	Hialuronan

Adaptada de Filocamo, *Hum Genomics*, 2011.

Abreviación: MPS, mucopolisacaridosis.

Reference: 1. Filocamo M, Morrone A. Lysosomal storage disorders: molecular basis and laboratory testing. *Hum Genomics*. 2011;5(3):156-169. doi:10.1186/1479-7364-5-3-156.