DISEASE:
Mucolipidosis type III alpha/beta

NAME:	Mucolipidosis type III alpha/beta
DESCRIPTION:	Mucolipidosis III alpha/beta (MLIII alpha/beta) is a lysosomal disorder characterized by progressive slowing of the growth rate from early childhood, stiffness and pain in joints, gradual coarsening of facial features, moderate developmental delay and mild intellectual disability in most patients.
ORPHACODE:	423461
SYNONYMS:	ML 3 alpha/beta ML III alpha/beta Mucolipidosis type 3 alpha/beta
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	GNPTAB
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RELATED CONTENT

Related Genetic Tests

- Lysosomal Storage Disease (gene panel)
- Mucolipidosis II and III

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- N-acetylglucosamine-1-phosphate transferase subunits alpha and beta

Related Gene Panels

- Lysomal Storage (64 genes) - VUB