DISEASE:
Mucolipidosis type II

NAME:	Mucolipidosis type II
DESCRIPTION:	A rare, severe form of mucolipidosis characterized by growth retardation, skeletal abnormalities (dysostosis multiplex, craniosynostosis, contractures of the joints and osteopenia), facial dysmorphism, stiff skin, obstructive airway, cardiomegaly and severe global developmental delay.
ORPHACODE:	576
SYNONYMS:	I-cell disease Mucolipidosis type II alpha/beta N-acetylglucosamine 1-phosphotransferase deficiency
XREF(S):	Orphanet OMIM ICD-10 MeSH
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