# DISEASE:
# Mucolipidosis type III gamma

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| **NAME:** | Mucolipidosis type III gamma |
| **DESCRIPTION:** | Mucolipidosis type III gamma (ML 3 gamma) is a very rare lysosomal disease, that has most often been observed in the Middle East, characterized by a progressive slowing of the growth rate in early childhood; stiffness and pain in shoulders, hips, and finger joints; a gradual, mild coarsening of facial features; and by a slower progression, milder clinical course and longer life expectancy than that seen in mucolipidosis type II and mucolipidosis type III alpha/beta. Cognitive function is normal or only slightly impaired and retinitis pigmentosa has been reported in a few patients. Many survive into early adulthood, but ultimately succumb to cardiorespiratory insufficiency. |
| **ORPHACODE:** | 423470 |
| **SYNONYMS:** | ML 3 gamma<br>ML III gamma<br>Mucolipidosis type 3 gamma |
| **XREF(S):** | Orphanet<br>OMIM<br>ICD-10 |
| **ANALYTE(S):** | GNPTG |
| **CREATED:** | 13 May 2019 - 01:02 |

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| **CHANGED:** | 22 Jun 2023 - 16:14 |

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# RELATED CONTENT

## Related Genetic Tests

- Lysosomal Storage Disease (gene panel)

## Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

## Related Analytes

- N-acetylglucosamine-1-phosphate transferase subunit gamma

## Related Gene Panels

- Lysomal Storage (64 genes) - VUB