DISEASE:
Familial medullary thyroid carcinoma

NAME:	Familial medullary thyroid carcinoma
DESCRIPTION:	A rare thyroid tumor characterized by a malignant neoplasm derived from the calcitonin-secreting parafollicular C-cells of the thyroid and occurring familially, but not as a component of multiple endocrine neoplasia syndromes. The commonly multifocal, bilateral nodules are typically located at the junction of the upper and middle thirds of the thyroid lobes. Clinically, patients may present with diarrhea, flushing, or weight loss caused by excessive secretion of calcitonin by the tumor. In rare cases, the tumor can also cause Cushing syndrome due to ectopic corticotropin production.
ORPHACODE:	99361
SYNONYMS:	Familial MTC
XREF(S):	Orphanet MeSH ICD-10 OMIM
ANALYTE(S):	ESR2 NTRK1 RET
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- Medullary thyroid carcinoma
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- Multiple endocrine neoplasia type 2A and 2B / Familial medullary thyroid carcinoma
- Onco-endocrine pathologies (gene panel)

Related Laboratories

- Centre de Génétique Humaine - Erasme ULB
- Centre de Génétique Médicale UCL
- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- estrogen receptor 2
- neurotrophic receptor tyrosine kinase 1
- ret proto-oncogene

Related Gene Panels

- Medullary thyroid carcinoma (3 genes) - UCL

- Onco-endocine pathologies (50 genes) - UCL
