

DISEASE:
Multiple endocrine neoplasia type 2B

NAME:	Multiple endocrine neoplasia type 2B
DESCRIPTION:	A rare form of multiple endocrine neoplasia type 2 (MEN2) syndrome characterized by aggressive medullary thyroid carcinoma in association with other endocrine tumors, notably pheochromocytoma (one or both adrenal glands can be affected). Onset is typically in infancy or childhood and patients often have a typical facies (mucosal neuromas of the lips and tongue, and bumpy lips), ophthalmologic abnormalities (alacrima in infancy, thickened and everted eyelids, mild ptosis, and prominent corneal nerves), skeletal anomalies (marfanoid body habitus, narrow long facies, pes cavus, pectus excavatum, high-arched palate, scoliosis, hyperextensible joints and slipped capital femoral epiphyses), and a generalized ganglioneuromatosis throughout the aerodigestive tract. Chronic constipation, abdominal distension, diarrhea, or megacolon at birth are often the initial manifestations.
ORPHACODE:	247709
SYNONYMS:	MEN2B Multiple endocrine neoplasia type 3 Wagenmann-Froboese syndrome

XREF(S):	Orphanet MeSH MedDRA ICD-10 OMIM
ANALYTE(S):	RET
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- Paraganglioma-pheochromocytoma (6 genes) - ULG

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