DISEASE:
Kostmann syndrome

NAME:	Kostmann syndrome
DESCRIPTION:	Kostmann syndrome is a rare, severe, congenital neutropenia disorder characterized by a lack of mature neutrophils (absolute neutrophil counts less than 500 cells/mm3) associated with frequent, recurrent bacterial infections (e.g. otitis media, pneumonia, sinusitis, urinary tract infections, abscesses of skin and/or liver) and increased promyelocytes in the bone marrow. Periodontal disease, as well as neurological symptoms, such as cognitive impairment, severe neurodegeneration and epilepsy, have been reported in some patients.
ORPHACODE:	99749
SYNONYMS:	Infantile agranulocytosis Severe congenital neutropenia type 3
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	HAX1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest-acc.healthdata.be/disease/3541

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