DISEASE:
Juvenile myelomonocytic leukemia

NAME:	Juvenile myelomonocytic leukemia
DESCRIPTION:	A rare myelodysplastic/myeloproliferative neoplasm characterized by a proliferation primarily of granulocytic and monocytic lineages with infiltration of the liver and spleen, among other organs. Blasts and promonocytes account for less than 20% of white blood cells in peripheral blood and bone marrow. Erythroid and megakaryocytic abnormalities are often present. BCR-ABL1 fusion is absent, while somatic mutations in genes of the RAS pathway or monosomy 7 may be found. The condition may also occur in the context of neurofibromatosis type 1 or Noonan syndrome-like disorder. Children of less than three years are predominantly affected, with a clear male preponderance. Most patients present with constitutional symptoms, signs of infection, and hepatosplenomegaly.
ORPHACODE:	86834
SYNONYMS:	JMML Juvenile chronic myelomonocytic leukemia
XREF(S):	Orphanet OMIM MeSH MedDRA ICD-10

ANALYTE(S):	RRAS PTPN11 KRAS NF1 NRAS CBL
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Primary immune deficiencies (gene panel)
- Trombosis - Hemostasis (gene panel)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- Cbl proto-oncogene
- KRAS proto-oncogene, GTPase
- neurofibromin 1
- NRAS proto-oncogene, GTPase
- protein tyrosine phosphatase non-receptor type 11
- RAS related

Related Gene Panels

- Primary immune deficiencies (444 genes) - KUL
- Trombosis - Hemostasis (107 genes) - KUL