DISEASE:
Noonan syndrome-like disorder with juvenile myelomonocytic leukemia

NAME:	Noonan syndrome-like disorder with juvenile myelomonocytic leukemia
DESCRIPTION:	A rare, genetic, polymalformative syndrome characterized by a Noonan-like phenotype associated with increased risk of developing juvenile myelomonocytic leukemia (JMML). The Noonan-like (NS) phenotype includes dysmorphic facial features (i.e. high forehead, hypertelorism, downslanting palpebral fissures, ptosis, low-set ears, prominent philtrum and short neck with or without pterygium colli), developmental delay, hypotonia and small head circumference. It can be associated with congenital heart defects or cardiomyopathy, ectodermal anomalies, and short stature. The NS phenotype is subtle or even inapparent in a large proportion of subjects, but may occasionally be severe. Leukemia can be the only clinical manifestation of the syndrome.
ORPHACODE:	363972
SYNONYMS:	CBL syndrome Noonan syndrome-like disorder with JMML
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	CBL
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