DISEASE:
Noonan syndrome with multiple lentigines

NAME:	Noonan syndrome with multiple lentigines
DESCRIPTION:	A rare multisystem genetic disorder characterized by cutaneous lentigines, hypertrophic cardiomyopathy, short stature, pectus deformity, and dysmorphic facial features.
ORPHACODE:	500
SYNONYMS:	Cardiomyopathic lentiginosis Familial multiple lentigines syndrome LEOPARD syndrome
XREF(S):	Orphanet MedDRA ICD-10 MeSH OMIM OMIM OMIM MeSH
ANALYTE(S):	PTPN11 BRAF RAF1

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RELATED CONTENT

Related Genetic Tests

- Cardiomyopathy, hereditary (gene panel)
- Noonan syndrome (Screening PTPN11)
- RASopathy (gene panel)
- Short Stature (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- B-Raf proto-oncogene, serine/threonine kinase
- protein tyrosine phosphatase non-receptor type 11
- Raf-1 proto-oncogene, serine/threonine kinase

Related Gene Panels

- Cardiomyopathy, hereditary (208 genes) - VUB
- Congenital heart disease (29 genes) - VUB
- RASopathy - KUL

- Short Stature (46 genes) - IPG

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