DISEASE:
Noonan syndrome-like disorder with loose anagen hair

NAME:	Noonan syndrome-like disorder with loose anagen hair
DESCRIPTION:	A Noonan-related syndrome, characterized by facial anomalies suggestive of Noonan syndrome, loose anagen hair, frequent congenital heart defects, distinctive skin features (darkly pigmented skin, keratosis pilaris, eczema or ichtyosis), and short stature that is often associated with a growth hormone deficiency. Psychomotor delay with attention deficit/hyperactivity disorder (ADHD) is frequently observed.
ORPHACODE:	2701
SYNONYMS:	Mazzanti syndrome NS/LAH
XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	PPP1CB SHOC2
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RELATED CONTENT

Related Genetic Tests

- RASopathy (gene panel)
- Short Stature (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- protein phosphatase 1 catalytic subunit beta
- SHOC2 leucine rich repeat scaffold protein

Related Gene Panels

- RASopathy - KUL
- Short Stature (46 genes) - IPG