DISEASE:
Costello syndrome

NAME:	Costello syndrome
DESCRIPTION:	A rare syndrome with intellectual disability, characterized by failure to thrive, short stature, joint laxity, soft skin, and distinctive facial features. Cardiac and neurological involvement is common and there is an increased lifetime risk of certain tumors. Costello syndrome belongs to the RASopathies, a group of conditions resulting from germline derived point mutations affecting the RAS-mitogen activated protein kinase pathway.
ORPHACODE:	3071
SYNONYMS:	FCS syndrome Faciocutaneoskeletal syndrome
XREF(S):	Orphanet MeSH MedDRA ICD-10 OMIM
ANALYTE(S):	HRAS
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RELATED CONTENT

Related Genetic Tests

- Cardiomyopathy, hereditary (gene panel)
- Costello Syndrome- Schimmelpenning syndrome
- RASopathy (gene panel)
- Short Stature (gene panel)

Related Laboratories

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

Related Analytes

- HRas proto-oncogene, GTPase

Related Gene Panels

- Cardiomyopathy, hereditary (208 genes) - VUB
- RASopathy - KUL
- Short Stature (46 genes) - IPG

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