DISEASE:
Cardiofaciocutaneous syndrome

NAME:	Cardiofaciocutaneous syndrome
DESCRIPTION:	A rare, multiple congenital anomalies syndrome characterized by craniofacial dysmorphology, congenital heart disease, dermatological abnormalities (most commonly hyperkeratotic skin and sparse, curly hair), neurological manifestations (hypotonia, seizures), failure to thrive and intellectual disability.
ORPHACODE:	1340
SYNONYMS:	CFC syndrome
XREF(S):	Orphanet ICD-10 MeSH OMIM OMIM OMIM OMIM
ANALYTE(S):	BRAF KRAS MAP2K1 MAP2K2
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RELATED CONTENT

Related Genetic Tests

- Cardiofaciocutaneous syndrome (5 genes)
- Primary lymphedema / fetal hydrops (gene panel)
- RASopathy (gene panel)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique Médicale UCL
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- B-Raf proto-oncogene, serine/threonine kinase
- KRAS proto-oncogene, GTPase
- mitogen-activated protein kinase kinase 1
- mitogen-activated protein kinase kinase 2

Related Gene Panels

- Cardiofaciocutaneous syndrome (5 genes))
- Congenital heart disease (29 genes) - VUB
- RASopathy - KUL
