DISEASE:
Ellis Van Creveld syndrome

NAME:	Ellis Van Creveld syndrome
DESCRIPTION:	A rare chondral and ectodermal dysplasia characterized by short ribs, polydactyly, growth retardation, and ectodermal and heart defects.
ORPHACODE:	289
SYNONYMS:	Chondroectodermal dysplasia Mesodermic dysplasia
XREF(S):	Orphanet OMIM OMIM MeSH OMIM MedDRA ICD-10

ANALYTE(S):	DYNC2LI1 GLI1 EVC EVC2 PRKACA PRKACB
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RELATED CONTENT

Related Genetic Tests

- Ciliopathy (gene panel)
- Ellis-van Creveld syndrome (2 genes)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Gent

Related Analytes

- dynein cytoplasmic 2 light intermediate chain 1
- EvC ciliary complex subunit 1
- EvC ciliary complex subunit 2
- GLI family zinc finger 1
- protein kinase cAMP-activated catalytic subunit alpha
- protein kinase cAMP-activated catalytic subunit beta

Related Gene Panels

- Ciliopathy (120 genes) - UGent
- Ellis-van Creveld (2 genes) - IPG

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