DISEASE:
Combined immunodeficiency with facio-oculo-skeletal anomalies

NAME:	Combined immunodeficiency with facio-oculo-skeletal anomalies
DESCRIPTION:	A rare combined immunodeficiency disorder characterized by primary immunodeficiency manifesting with repeated bacterial, viral and fungal infections, in association with neurological manifestations (hypotonia, cerebellar ataxia, myoclonic seizures), developmental delay, optic atrophy, facial dysmorphism (high forehead, hypoplastic supraorbital ridges, palpebral edema, hypertelorism, flat nasal bridge, broad nasal root and tip, anteverted nares, thin lower lip overlapped by upper lip, square chin) and skeletal anomalies (short metacarpals/metatarsals with cone-shaped epiphyses, osteopenia).
ORPHACODE:	221139
SYNONYMS:	Roifman-Chitayat syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	KNSTRN PIK3CD
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RELATED CONTENT

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- Primary immune deficiencies (gene panel)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- kinetochore localized astrin (SPAG5) binding protein
- phosphatidylinositol-4,5-bisphosphate 3-kinase catalytic subunit delta

Related Gene Panels

- Immunogenetics (21 genes)
- Primary immune deficiencies (444 genes) - KUL