DISEASE:
Coffin-Lowry syndrome

NAME:	Coffin-Lowry syndrome
DESCRIPTION:	A rare X-linked syndromic intellectual disability characterized by global development delay, postnatal growth retardation leading to short stature, facial dysmorphism, short hands with tapering fingers and progressive skeletal abnormalities including kyphoscoliosis and pectus carinatum/excavatum. Intellectual disability ranges from mild to severe.
ORPHACODE:	192
SYNONYMS:	CLS
XREF(S):	Orphanet OMIM MeSH ICD-10
ANALYTE(S):	RPS6KA3
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RELATED CONTENT

Related Genetic Tests

- Short Stature (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

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- ribosomal protein S6 kinase A3

Related Gene Panels

- Short Stature (46 genes) - IPG