DISEASE:
Autosomal recessive Robinow syndrome

NAME:	Autosomal recessive Robinow syndrome
DESCRIPTION:	Autosomal recessive Robinow syndrome (RRS) is the less common type of Robinow syndrome (RS, see this term) characterized by short-limb dwarfism, costovertebral segmentation defects and abnormalities of the head, face and external genitalia.
ORPHACODE:	1507
SYNONYMS:	COVESDEM syndrome Costovertebral segmentation defect-mesomelia syndrome RRS
XREF(S):	Orphanet OMIM OMIM MeSH ICD-10
ANALYTE(S):	ROR2 NXN
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Short Stature (gene panel)
- Short stature/ Growth retardation/ (gene panel)
- cleft lip with/whitout cleft palate (virtual gene panel)

Related Laboratories

- Centre de Génétique Médicale UCL
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- nucleoredoxin
- receptor tyrosine kinase like orphan receptor 2

Related Gene Panels

- Cleft lip and palate / dysmorphic facial features / craniofacial anomalies (255 genes)) - UCL
- Growth retardation/short stature (genepanel) - UZA
- Short Stature (46 genes) - IPG