DISEASE:
Desbuquois syndrome

NAME:	Desbuquois syndrome
DESCRIPTION:	Desbuquois syndrome (DBQD) is an osteochondrodysplasia characterized by severe micromelic dwarfism, facial dysmorphism, joint laxity with multiple dislocations, vertebral and metaphyseal abnormalities and advanced carpotarsal ossification. Two forms have been distinguished on the basis of the presence (type 1) or the absence (type 2) of characteristic hand anomalies. A variant form of DBQD, Kim variant (see these terms), has also been described and is characterized by short stature and articular, minor facial and significant hand anomalies.
ORPHACODE:	1425
SYNONYMS:	DBQD Desbuquois dysplasia
XREF(S):	Orphanet ICD-10 MeSH OMIM OMIM
ANALYTE(S):	CSGALNACT1 CANT1 XYLT1

CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Ehlers-Danlos syndroom, EDS (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Gent

Related Analytes

- calcium activated nucleotidase 1
- chondroitin sulfate N-acetylgalactosaminyltransferase 1
- xylosyltransferase 1

Related Gene Panels

- Ehlers-Danlos syndrome -UGent