

Opis choroby *

Definicja

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.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych DBQD	Dysplazja Desbuquoisa Desbuquois dysplasia

Kod ORPHA	Kod OMIM	Kod ICD10
1425	615777	Q78.8

Kod ICD11
LD24.E

*Źródło

orphanet