

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

Zespół wad wrodzonych DBQD

Synonimy

Dysplazja Desbuquoisa

Desbuquois dysplasia

Kod ORPHA

1425

Kod OMIM

615777

Kod ICD10

Q78.8

Kod ICD11

LD24.E

[*Źródło](#)

orphanet