Zespół Ruvalcaba
Kod Orpha: 3121 Kod OMIM: 180870

Opis choroby *

Definicja
Ruvalcaba syndrome is an extremely rare malformation syndrome, described in less than 10 patients to date, characterized by microcephaly with characteristic facies (downslanting parpebral fissures, microstomia, beaked nose, narrow maxilla), very short stature, narrow thoracic cage with pectus carinatum, hypoplastic genitalia and skeletal anomalies (i.e. characteristic brachydactyly and osteochondritis of the spine) as well as intellectual and developmental delay.
Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
3121

Kod OMIM
180870

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.