Opis choroby *

Definicja

A rare developmental defect during embryogenesis characterized by moderate to severe prenatal and postnatal growth retardation, microcephaly, a distinctive facial appearance, profound psychomotor delay, hip and knee contractures and rockerbottom feet.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Bowen syndrome, Hutterite type
Zespół Bowena, typ Hutterite

Kod ORPHA
1270

Kod OMIM
211180

Kod ICD10
Q87.8

Kod ICD11
LD20.2

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*Źródło

orphanet