Opis choroby *

Definicja

A rare genetic syndrome characterized by skeletal anomalies, including short stature, ridging of the metopic suture, a fusion of cervical vertebrae, thoracic hemivertebrae, scoliosis, sacral hypoplasia, short middle phalanges. Patients also had a moderate intellectual disability and abducens palsies. Glucose intolerance and imperforate anus were also described.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Christian syndrome
Zespół Christiana

Kod ORPHA
1436

Kod OMIM
309620

Kod ICD10
Q87.5

Kod ICD11
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*Źródło

orphanet