Opis choroby *

Definicja

A rare disorder characterised by the absence of the upper limbs and severe underdevelopment of the lower limbs. Minor facial abnormalities (depressed nasal root, upturned nose, infra-orbital creases, prominent cheeks and micrognathia) were also reported. The syndrome has been described in three foetuses born to non consanguineous parents.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
1027

Kod OMIM
601360

Kod ICD10
Q73.0

Kod ICD11
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*Źródło

orphanet