

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 fetuses 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