

Opis choroby *

Definicja

Richieri Costa-Pereira syndrome is characterized by short stature, Robin sequence, cleft mandible, pre/postaxial hand anomalies (including hypoplastic thumbs), and clubfoot. It has been described in 14 Brazilian families and in one unrelated French patient. Prominent low set ears and a highly arched palate were also observed. Transmission is autosomal recessive.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Short stature-Pierre Robin sequence-cleft mandible-hand anomalies clubfoot syndrome

Niski wzrost - sekwencja Pierre'a Robina -

rozszczep żuchwy - wady rąk - stopa końsko-szpotawa

Niski wzrost - zespół Pierre'a Robina - rozszczep żuchwy - wady rąk - stopa końsko-szpotawa

Short stature-Pierre Robin syndrome-cleft

mandible-hand anomalies clubfoot syndrome

Kod ORPHA

3102

Kod OMIM

268305

Kod ICD10

Q87.8

Kod ICD11

LD2F.1Y

*Źródło

orphanet