Opis choroby *

Definicja

A rare genetic bone development disorder characterized by multiple congenital fractures, slender ribs and long bones, deficient ossification of the skull, and dysmorphic facial features reminiscent of Hallermann-Streiff syndrome (such as high forehead and triangular face with small jaw, deep-set eyes, beaked, narrow nose, downturned mouth, and posteriorly angulated ears). Bilateral microphthalmia, cataracts, and pulmonary hypoplasia have also been reported. The disease is fatal in the neonatal period. There have been no further descriptions in the literature since 1995.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Dennis-Fairhurst-Moore syndrome
Ciężka postać zespołu Hallermanna, Streiffa i Françoisa
Zespół Dennisa, Fairhursta i Moore'a
Zespoł Hallermanna, Streiffa i Françoisa, postać ciężka
Zespół podobny do zespołu Hallermana i Streiffa
Hallermann-Streiff-François syndrome, severe form
Severe Hallermann-Streiff-François syndrome

Kod ORPHA
2109

Kod OMIM
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Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet