

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 Dennis-Fairhurst-Moore syndrome

Synonimy

Ciężka postać zespołu Hallermanna, Streiffa i François

Zespół Dennisa, Fairhursta i Moore'a

Zespół Hallermanna, Streiffa i François, 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