Opis choroby *

Definicja

A rare primary bone dysplasia characterized by micromelia with rhizomelic shortening, metaphyseal widening of the long bones, brachydactyly, small scapulae, micrognathia and thoracic insufficiency requiring tracheostomy and ventilation, and severe myopia and sensorineural hearing loss. Further dysmorphic craniofacial features include frontal bossing, proptosis, epicanthal folds, short nose, flat nasal bridge, anteverted nares, midfacial retrusion, and cleft palate.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Autosomal dominant myopia-midfacial retrusion-sensorineural deafness-rhizomelic dysplasia syndrome
Autosomal dominant myopia-midfacial retrusion-sensorineural deafness-rhizomelic dysplasia syndrome

Kod ORPHA
440354

Kod OMIM
-

Kod ICD10
Q87.5

Kod ICD11
-

---

*Źródło

orphanet