Opis choroby *

Definicja

A rare, genetic, primary bone dysplasia with increased bone density disorder characterized by bone abnormalities, including metaphyseal plaques, osteopathia striata, marked cranial sclerosis, and sclerosis of the ribs and long bones, as well as macrocephaly, cleft palate, hearing loss, developmental delay, and facial dysmorphism (hypertelorism, prominent forehead, wide nasal bridge). Hypotonia, tracheo-/laryngomalacia, and astigmatic myopia are also associated.

Dane

Klasyfikacja
Choroba

Kod ORPHA
324364

Kod OMIM
-

Kod ICD10
M85.8

Kod ICD11
-

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*Źródło

orphanet