Opis choroby *

Definicja

A rare multiple congenital anomalies syndrome characterized by upper limb defects (hypoplastic thumb with hypoplasia of the metacarpal bone and phalanges and delayed bone maturation), developmental delay, central hearing loss, unilateral poorly developed antihelix, bilateral choroid coloboma and growth retardation.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2489

Kod OMIM
274205

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet