Opis choroby *

Definicja

A rare, genetic dysostosis with predominant craniofacial involvement characterized by bilateral external ear malformations, mandibular condyle hypoplasia, microstomia, micrognathia, microglossia and facial asymmetry. Additional manifestations include hypotonia, ptosis, cleft palate, full cheeks, developmental delay, hearing impairment and respiratory distress. Significant intra- and interfamilial phenotypic variation has been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Question mark ear syndrome
Question mark ear syndrome

Kod ORPHA
137888

Kod OMIM
615706

Kod ICD10
Q75.8

Kod ICD11
LD2F.16

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*Źródło

orphanet