

## 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

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych	Question mark ear syndrome
	Question mark ear syndrome

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
137888	615706	Q75.8

**Kod ICD11**  
LD2F.16

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### \*Źródło

orphanet