

Opis choroby *

Definicja

Cleft palate-large ears-small head syndrome is a rare, genetic syndrome characterized by cleft palate, large protruding ears, microcephaly and short stature (prenatal onset). Other skeletal abnormalities (delayed bone age, distally tapering fingers, hypoplastic distal phalanges, proximally placed thumbs, fifth finger clinodactyly), Pierre Robin sequence, cystic renal dysplasia, proximal renal tubular acidosis, hypospadias, cerebral anomalies on imaging (enlargement of lateral ventricles, mild cortical atrophy), seizures, hypotonia and developmental delay are also observed.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Say-Barber-Hobbs syndrome
	Zespół Say, Barber i Hobbsa

Kod ORPHA	Kod OMIM	Kod ICD10
2013	181180	Q87.8

Kod ICD11

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*Źródło

orphanet