Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by congenital cataract, sensorineural hearing loss, developmental delay with variable degrees of intellectual disability, seizures, short stature, brachycephaly, and dysmorphic facial features (such as flat facial appearance, ptosis, short nasal tip, long philtrum, low-set and posteriorly rotated ears, and small mouth). Additional reported manifestations are skeletal abnormalities, nail dystrophy, mammary gland hypoplasia, and autism spectrum disorder.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Brachycephaly-deafness-cataract-intellectual disability syndrome
Krótkogłowie - głuchota - zaćma - niepełnosprawność intelektualna
Brachycephaly-hearing loss-cataract-intellectual disability syndrome
Fine-Lubinsky syndrome

Kod ORPHA
1272

Kod OMIM
601353

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet