

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