Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay, intellectual disability, absent scrotum or labia majora, absent or underdeveloped nipples and a tuft of hair extruding from the lactiferous ducts, bilateral corneal opacities, and dysmorphic craniofacial features (microcephaly, short forehead, and ear abnormalities, among others). Patients also show horizontal nystagmus and ataxic gait. Brain MRI reveals small cerebellar hemispheres and vermis and a small pons.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Congenital agenesis of labia majora or scrotum-cerebellar malformation-corneal dystrophy-facial dysmorphism syndrome
Congenital agenesis of labia majora or scrotum-cerebellar malformation-corneal dystrophy-facial dysmorphism syndrome

Kod ORPHA
495875

Kod OMIM
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Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet