Opis choroby *

Definicja

A rare genetic neurological disorder characterized by congenital microcephaly, severe intellectual disability, hypertonia at birth lessening with age, ataxia, and specific dysmorphic facial features including hirsutism, low anterior hairline and bitemporal narrowing, arched, thick, and medially sparse eyebrows, long eyelashes, lateral upper eyelids swelling and a skin fold partially covering the inferior eyelids, low-set posteriorly rotated protruding ears, anteverted nares, and a full lower lip. Brain imaging shows partial to almost complete agenesis of the corpus callosum and variable degrees of cerebellar hypoplasia.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
466688

Kod OMIM
616819

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet