Opis choroby *

Definicja

Intellectual disability-severe speech delay-mild dysmorphism syndrome is a rare, genetic, syndromic intellectual disability disorder, with highly variable phenotype, typically characterized by mild to severe global development delay, severe speech and language impairment, mild to severe intellectual disability, dysphagia, hypotonia, relative to true macrocephaly, and behavioral problems that may include autistic features, hyperactivity, and mood lability. Facial gestalt typically features a broad, prominent forehead, hypertelorism, downslanting palpebral fissures, ptosis, a short bulbous nose with broad tip, thick vermilion border, wide, and open mouth with downturned corners. Brain, cardiac, urogenital and ocular malformations may be associated.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
FOXP1 syndrome
FOXP1 syndrome

Kod ORPHA
391372

Kod OMIM
613670

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet