Opis choroby *

Definicja

A rare genetic syndromic intellectual disability characterized by microcephaly, global developmental delay, mild to severe intellectual disability, impairment of speech, feeding problems, behavior problems (often autism spectrum disorder) and dysmorphic facial features (such as prominent ears, deep-set eyes, a short nose with a broad nasal tip, and retrognathia with a broad chin). Other, more variable manifestations include seizures, short stature, ocular anomalies, cardiac anomalies, urogenital anomalies and musculoskeletal defects.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
DYRK1A syndrome
DYRK1A syndrome

Kod ORPHA
464306

Kod OMIM
614104

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet