## 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 Synonimy

Zespół wad wrodzonych DYRK1A syndrome

DYRK1A syndrome

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 464306
 614104
 Q87.8

**Kod ICD11** 

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## \*Źródło

orphanet