Opis choroby *

Definicja

A rare genetic neurodevelopmental disorder characterized by global developmental delay (DD) and variable degrees of intellectual disability (ID) with delayed or limited/absent speech development associated with neonatal hypotonia, feeding difficulties, cardiac anomalies and dysmorphic facial features, predominantly broad nasal tip and thin, tented upper lip. Microcephaly, frequent infections, gastrointestinal and/or ocular anomalies have also been described.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Arboleda-Tham syndrome
KAT6A syndrome
Autosomalny dominujący zespół niepełnosprawności intelektualnej - anomalii twarzoczaszki - wad serca

Kod ORPHA
457193

Kod OMIM
616268

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet