Opis choroby *

Definicja

Microcephaly-cerebellar hypoplasia-cardiac conduction defect syndrome is a rare, genetic congenital anomalies/dysmorphic syndrome characterized by growth failure, global developmental delay, profound intellectual disability, autistic behaviors, acquired second-degree heart block with bradycardia and vasomotor instability. Hands and feet present with long fusiform fingers, campto-clinodactyly and crowded toes while craniofacial dysmorphism includes microcephaly, broad forehead, thin eyebrows, upslanting palpebral fissures, large ears with prominent antihelix, prominent nose, long philtrum, thin upper lip vermillion and prominent lower lip. Neurological signs include hypotonia, brisk reflexes, dystonic-like movements and truncal ataxia and imaging shows cerebellar hypoplasia and simplified gyral pattern.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Microcephaly-cerebellar hypoplasia-congenital heart conduction defect syndrome
Zespół mikrocefalia-hipoplazja móżdżku-wrodzone zaburzenia przewodzenia w sercu

Kod ORPHA
329332

Kod OMIM
614407

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet