

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 vermilion 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 mózdzku-wrodzone zaburzenia przewodzenia w sercu

Kod ORPHA

329332

Kod OMIM

614407

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet