Opis choroby *

Definicja

Microcephaly-facio-cardio-skeletal syndrome, Hadziselimovic type is a rare syndrome with cardiac malformations (see this term), characterized by prenatal-onset growth retardation (low birth weight and short stature), hypotonia, developmental delay and intellectual disability associated with microcephaly and craniofacial (low anterior hairline, hypotelorism, thick lips with carp-shaped mouth, high-arched palate, low-set ears), cardiac (conotruncal heart malformations such as tetralogy of Fallot; see these terms) and skeletal (hypoplastic thumbs and first metacarpals) abnormalities.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Hadziselimovic syndrome
Zespół Hadziselimovica
Microcephaly-faciocardioskeletal syndrome

Kod ORPHA
217026

Kod OMIM
612946

Kod ICD10
Q87.8

Kod ICD11
LD2F.1Y

---

*Źródło

orphanet