

Opis choroby *

Definicja

McDonough syndrome is a rare, multiple congenital anomalies/dysmorphic syndrome characterized by facial dysmorphism (prominent superciliary arcs, synophrys, strabismus, large, anteverted ears, large nose, malocclusion of teeth), delayed psychomotor development, intellectual disability and congenital heart defects (e.g. pulmonic stenosis, patent ductus arteriosus, atrial septal defect). Additional features include thorax deformation (pectus excavatum/carinatum), kyphoscoliosis, diastasis recti and cryptorchidism. There have been no further descriptions in the literature since 1984.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

2471

Kod OMIM

248950

Kod ICD10

Q87.8

Kod ICD11

LD2F.1Y

*Źródło

orphanet