Opis choroby *

Definicja

McDonough syndrome is a rare, multiple congenital anomalies/dysmorphic syndrome characterized by facial dysmorphsim (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

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*Źródło

orphanet