Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by vertebral segmentation defects associated with cardiac (patent ductus arteriosus, atrial septal defect, hypoplastic left heart) and renal (hypoplastic kidneys, chronic kidney disease) anomalies. Additional reported features include limb defects, short stature, global developmental delay, intellectual disability, and sensorineural hearing loss, among others.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Congenital NAD deficiency disorder
Wrodzony niedobór NAD

Kod ORPHA
521438

Kod OMIM
617661

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet