

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	Synonimy
Zespół wad wrodzonych	Congenital NAD deficiency disorder
	Wrodzony niedobór NAD

Kod ORPHA	Kod OMIM	Kod ICD10
521438	617661	Q87.8

Kod ICD11

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*Źródło

orphanet