

Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by the association of congenital hypoparathyroidism, nephropathy, congenital lymphedema, mitral valve prolapse and brachytelephalangy. Additional features include mild facial dysmorphism, hypertrichoses, and nail abnormalities. There have been no further descriptions in the literature since 1993.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Dahlberg syndrome

Zespół Dahlberga

Zespół obrzęk limfatyczny - niedoczynność
przyszczyk

Lymphedema-hypoparathyroidism syndrome

Kod ORPHA

1563

Kod OMIM

247410

Kod ICD10

Q87.8

Kod ICD11

LD27.0Y

*Źródło

orphanet