

Opis choroby *

Definicja

A rare genetic disease characterized by choanal atresia and early onset of lymphedema of the lower extremities. Additional reported features include facial dysmorphism (hypertelorism, broad forehead, smooth philtrum, unilateral low-set ear, and high-arched palate), hypoplastic nipples, and pectus excavatum.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

99141

Kod OMIM

613611

Kod ICD10

Q82.0

Kod ICD11

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*Źródło

orphanet