Opis choroby *

Definicja

A rare primary lymphedema characterized by a highly variable lymphatic phenotype ranging from severe lymphatic-related hydrops fetalis, which may cause perinatal demise or fully resolve to become completely asymptomatic, to a mild presentation in older patients with persistent varicose veins, peripheral edema, and impaired lymph drainage in the lower limbs. Atrial septal defect has been described in association and may be the only anomaly in some patients.

Dane

Klasyfikacja
Choroba

Synonimy
EPHB4-related LRHF/GLD
Uogólniona dysplazja limfatyczna z ubytkiem w przegrodzie międzyprzedsionkowej związana z EPHB4
Uogólniona dysplazja limfatyczna z nie-immunologicznym obrzękiem płodu związana z EPHB4
EPHB4-related generalized lymphatic dysplasia with atrial septal defect
EPHB4-related generalized lymphatic dysplasia with non-immune hydrops fetalis

Kod ORPHA
568065

Kod OMIM
617300

Kod ICD10
P83.2

Kod ICD11
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*Źródło

orphanet