

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](#)

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