Opis choroby *

Definicja

A rare genetic primary lymphedema characterized by uniform, widespread lymphedema, often with systemic involvement such as intestinal and pulmonary lymphangiectasia, pleural and pericardial effusions, and chylothorax. There is a high incidence of non-immune hydrops fetalis, which may result in fetal demise or fully resolve after birth. Severe, recurrent facial cellulitis is observed in some patients. Presence of epicanthic folds or micrognathia has occasionally been reported, while intelligence is normal, and seizures are absent.

Dane

Klasyfikacja
Choroba

Synonimy
Generalized lymphatic dysplasia of Fotiou
Uogólniona dysplazja limfatyczna z zajęciem wielu układów związana z PIEZO1
PIEZO1-related LRHF/GLD
PIEZO1-related generalized lymphatic dysplasia with systemic involvement
PIEZO1-related lymphatic-related hydrops fetalis

Kod ORPHA
568062

Kod OMIM
-

Kod ICD10
Q82.0

Kod ICD11
-

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*Źródło

orphanet