Opis choroby *

Definicja

A rare primary lymphedema characterized by bilateral, painless lower limb swelling present at birth. Prominent veins around the ankles and on the dorsa of the feet, dysplastic and upslanting toenails due to edema of the nailbed, and subtle dysmorphic facial features (such as high forehead, hypertelorism, depressed nasal bridge, mild bilateral ear dysplasia, and short neck) have also been described. The degree of lymphatic impairment is milder than in the otherwise clinically similar Milroy disease, as evidenced by slightly less severe lymphedema and significantly more uptake of tracers on lymphoscintigraphy.

Dane

Klasyfikacja
Choroba

Synonimy
VEGFC-related congenital primary lymphedema
VEGFC-related congenital primary lymphedema

Kod ORPHA
569821

Kod OMIM
-

Kod ICD10
Q82.0

Kod ICD11
BD93.0

---

*Źródło

orphanet