Opis choroby *

Definicja

A rare genetic disease characterized by the association of primary lymphedema (typically presenting in one or both lower limbs and frequently affecting the genitalia) and acute myeloid leukemia (often preceded by pancytopenia or myelodysplasia), with or without congenital deafness. Additional reported features include bilateral syndactyly of the toes, hypotelorism and epicanthic folds, long tapering fingers, and neck webbing.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Emberger syndrome
Zespół Embergera
Hearing loss-lymphedema-leukemia syndrome

Kod ORPHA
3226

Kod OMIM
614038

Kod ICD10
D46.7

Kod ICD11
BD93.0

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*Źródło

orphanet