

Opis choroby *

Definicja

Lymphedema-atrial septal defects-facial changes syndrome is characterised by congenital lymphoedema of the lower limbs, atrial septal defect and a characteristic facies (a round face with a prominent forehead, a flat nasal bridge with a broad nasal tip, epicanthal folds, a thin upper lip and a cleft chin). It has been described in two brothers and a sister. Transmission appears to be autosomal recessive.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Irons-Bhan syndrome

Zespół Ironsa i Bianchi

Irons-Bianchi syndrome

Kod ORPHA

86915

Kod OMIM

601927

Kod ICD10

Q87.8

Kod ICD11

BD93.0

*Źródło

orphanet