

## 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

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#### \*Źródło

orphanet