## Opis choroby *

Definicja

Filippi syndrome is characterised by microcephaly, cutaneous syndactyly of the fingers and toes, intellectual deficit, growth retardation and a characteristic facies (high and broad nasal bridge, thin alae nasi, micrognathia and a high frontal hairline). So far, less than 25 cases have been reported. Cryptorchidism, polydactyly, and teeth and hair anomalies may also be present. Transmission is autosomal recessive.

Dane

**Klasyfikacja**
Zespół wad wrodzonych

Synonimy
Type 1 syndactyly-microcephaly-intellectual disability syndrome
Syndaktylia, typu 1 - małogłowie - niepełnosprawność intelektualna

**Kod ORPHA**
3255

**Kod OMIM**
272440

**Kod ICD10**
Q87.8

**Kod ICD11**
LD2F.1Y

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*Źródło

orphanet