

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

Kod ORPHA	Kod OMIM	Kod ICD10
3255	272440	Q87.8

Kod ICD11
LD2F.1Y

*Źródło

orphanet