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