

Opis choroby *

Definicja

Microcephaly-deafness-intellectual disability syndrome is characterised by microcephaly, deafness, intellectual deficit and facial dysmorphism (facial asymmetry, prominent glabella, low-set and cup-shaped ears, protruding lower lip, micrognathia). It has been described in a mother and her son. The mode of inheritance is probably autosomal dominant.

Dane

Klasyfikacja

Zespół wad wrodzonych Kawashima-Tsuji syndrome

Synonimy

Zespół Kawashima i Tsuji

Microcephaly-hearing loss-intellectual disability syndrome

Kod ORPHA

2533

Kod OMIM

156620

Kod ICD10

Q87.8

Kod ICD11

LD2H.Y

*Źródło

orphanet