

## 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

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#### \*Źródło

orphanet