

## **Opis choroby \***

### Definicja

Microphthalmia-ankyloblepharon-intellectual disability syndrome is characterized by microphthalmia, ankyloblepharon and intellectual deficit. It has been described in seven male patients from two generations of a Northern Ireland family. The causative gene is localized to the Xq27-q28 region. The syndrome is transmitted as an X-linked recessive trait.

### Dane

#### **Klasyfikacja**

Zespół wad wrodzonych MCOPS4

MCOPS4

Syndromiczne małoocze typu 4

Syndromic microphthalmia type 4

#### **Kod ORPHA**

85275

#### **Kod OMIM**

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#### **Kod ICD10**

Q11.2

#### **Kod ICD11**

LD21.0

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#### \*Źródło

orphanet