

## Opis choroby \*

### Definicja

FRAXF syndrome was originally identified in a family with developmental delay and an expanded CCG repeat at the folate-sensitive FRAXF fragile site. Since this initial description, FRAXF has been associated with a range of manifestations but no clear phenotype has been established.

### Dane

### Klasyfikacja

Choroba

#### Kod ORPHA

100974

#### Kod OMIM

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#### Kod ICD10

Q99.2

#### Kod ICD11

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### \*Źródło

orphanet