

## Opis choroby \*

### Definicja

A rare chromosomal anomaly syndrome characterized by complete or partial loss of an X chromosome in phenotypic females, clinically manifesting with short stature, primary ovarian insufficiency as well as cardiovascular, renal, liver, autoimmune diseases, hearing loss and neurocognitive abnormalities.

### Dane

#### Klasyfikacja

Zespół wad wrodzonych 45,X syndrome

#### Synonimy

Zespół 45,X

Zespół 45,X/46,XX

45,X/46,XX syndrome

#### Kod ORPHA

881

#### Kod OMIM

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#### Kod ICD10

Q96.4

#### Kod ICD11

LD50.0

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#### \*Źródło

orphanet