

## Opis choroby \*

### Definicja

A rare congenital disorder of copper metabolism that is principally characterized by bony exostoses (including the pathognomonic occipital horns), and connective tissue manifestations with cutis laxa and bladder diverticula. Central nervous system involvement is variable.

### Dane

### Klasyfikacja

Choroba

#### Kod ORPHA

198

#### Kod OMIM

304150

#### Kod ICD10

E83.0

#### Kod ICD11

LD28.2

---

### \*Źródło

orphanet