

## Opis choroby \*

### Definicja

Lethal occipital encephalocele-skeletal dysplasia syndrome is a rare, genetic, bone development disorder characterized by occipital and parietal bone hypoplasia leading to occipital encephalocele, calvarial mineralization defects, craniosynostosis, radiohumeral fusions, oligodactyly and other skeletal anomalies (arachnodactyly, terminal phalangeal aplasia of the thumbs, bilateral absence of the great toes, pronounced bilateral angulation of femora, shortened limbs, advanced osseous maturation). Fetal death in utero is associated.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

293925

#### Kod OMIM

614416

#### Kod ICD10

Q87.5

#### Kod ICD11

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### \*Źródło

orphanet