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