Opis choroby *

Definicja

A rare genetic bone development disorder characterized by pre- and postnatal growth retardation, skeletal anomalies such as arachnodactyly and bilateral talipes equinovarus, joint contractures with camptodactyly, dysmorphic facial features (including midface hypoplasia or micrognathia), and abnormalities of the anterior segment of the eye. Skeletal imaging may show diffuse osteopenia and multiple fractures. The syndrome is lethal within the first year of life.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Al Gazali-Al Talabani syndrome
Zespół Al Gazali i Al Talabani
Zespół Al Gazali i Lytle
Al Gazali-Lytle syndrome

Kod ORPHA
2725

Kod OMIM
609465

Kod ICD10
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Kod ICD11
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*Źródło

orphanet