Choroba tkanki łącznej z powodu niedoboru hydroksylazy-3 lizylowej
Kod Orpha: 300284 Kod OMIM: 612394

Opis choroby *

Definicja

Connective tissue disorder due to lysyl hydroxylase-3 deficiency is a rare, genetic disease, caused by lack of lysyl hydrohylase 3 (LH3) activity, characterized by multiple tissue and organ involvement, including skeletal abnormalities (club foot, progressive scoliosis, osteopenia, pathologic fractures), ocular involvement (flat retinae, myopia, cataracts) and hair, nail and skin anomalies (coarse, abnormally distributed hair, skin blistering, reduced palmar creases, hypoplastic nails). Patients also present intrauterine growth retardation, facial dysmorphism (flat facial profile, low-set ears, shallow orbits, short and upturned nose, downturned corners of mouth) and joint flexion contractures. Growth and developmental delay, bilateral sensorineural deafness, friable diaphragm and later-onset spontaneous vascular ruptures are additional reported features.

Dane

Klasyfikacja
Choroba

Synonimy
Bone fragility-contractures-arterial rupture-deafness syndrome
Choroba tkanki łącznej z powodu niedoboru LH3
Zespół kruchość kości-przykurcze-pęknięcie tętnicy-głuchota
Bone fragility-contractures-arterial rupture-hearing loss syndrome
Connective tissue disorder due to LH3 deficiency

Kod ORPHA
300284

Kod OMIM
612394

Kod ICD10
M35.8

Kod ICD11
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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.