

Opis choroby *

Definicja

Connective tissue disorder due to lysyl hydroxylase-3 deficiency is a rare, genetic disease, caused by lack of lysyl hydroxylase 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