Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by postnatal tall stature with long hands and feet, scoliosis, distinctive dysmorphic facial features (prominent forehead, proptosis, downslanting palpebral fissures, broad nasal bridge, thin upper lip, and pointed chin), hyperelastic, thin, and fragile skin, lipodystrophy, and variable intellectual disability and neurological deterioration. Additional reported manifestations include craniosynostosis, camptodactyly, progressive flexion contractures, joint dislocation, and cerebrovascular complications, among others. Brain MRI may show extensive periventricular white matter lesions and other anomalies.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Skeletal overgrowth-craniofacial dysmorphism-hyperelastic skin-white matter lesions syndrome
Zespół nadmiernego wzrastania Kosakiego
Zespół przerostu Kosaki

Kod ORPHA
477831

Kod OMIM
616592

Kod ICD10
Q87.3

Kod ICD11
LD2C

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*Źródło

orphanet