Opis choroby *

Definicja

A rare genetic disease characterized by childhood onset of bilateral progressive sensorineural hearing loss, ocular anomalies (myopia, cataract, retinitis pigmentosa), central and peripheral nervous system features (dementia, epilepsy, ataxia, peripheral neuropathy), ectodermal features (skin atrophy, alopecia, dental caries), and skeletal anomalies (bone cysts, joint stiffness, scoliosis, kyphosis). Laboratory examination may reveal elevated cerebrospinal fluid protein.

Dane

Klasyfikacja
Choroba

Kod ORPHA
2047

Kod OMIM
136300

Kod ICD10
Q87.8

Kod ICD11
-

---

*Źródło

orphanet