

Zespół Flynna i Airda

Kod Orpha: 2047 Kod OMIM: 136300

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

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[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.