Opis choroby *

Definicja

A rare, genetic, syndromic rod-cone dystrophy disorder characterized by psychomotor developmental delay from early childhood, intellectual disability, short stature, mild facial dysmorphism (e.g. upslanted palpebral fissures, hypoplastic alae nasi, malar hypoplasia, attached earlobes), excessive dental spacing and malocclusion, juvenile cataract and ophthalmologic findings of atypical retinitis pigmentosa (i.e. salt-and-pepper retinopathy, attenuated retinal arterioles, generalized rod-cone dysfunction, mottled macula, peripapillary sparing of retinal pigment epithelium).

Dane

Klasyfikacja
Choroba

Synonimy
Retinal dystrophy-juvenile cataract-short stature syndrome
Zespół dystrofii siatkówki, zaćmy młodzieńczej i niskiego wzrostu

Kod ORPHA
436245

Kod OMIM
616108

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet