Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by developmental delay with mild intellectual disability, short stature, facial dysmorphism (such as sparse hair, high forehead, deep-set eyes, short and upslanting palpebral fissures, short nose, anteverted nares, wide nasal base with broad nasal tip and broad columella, long philtrum, thin upper lip, and low-set, posteriorly rotated ears), and variable onset of sensorineural hearing loss and retinitis pigmentosa. Additional features are other ocular anomalies, abnormalities of the fingers, hypothyroidism, and signs of premature aging. Brain imaging shows cerebellar atrophy and dysmyelination.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Retinitis pigmentosa-deafness-premature aging-short stature-facial dysmorphism syndrome
Retinitis pigmentosa-deafness-premature aging-short stature-facial dysmorphism syndrome

Kod ORPHA
494439

Kod OMIM
617763

Kod ICD10
Q87.8

Kod ICD11
-

---

*Źródło

orphanet