Opis choroby *

Definicja

Piebald trait-neurologic defects syndrome is a rare, genetic, pigmentation anomaly of the skin syndrome characterized by ventral as well as dorsal leukoderma of the trunk and a congenital white forelock, in association with cerebellar ataxia, impaired motor coordination, intellectual disability of variable severity and progressive, mild to profound, uni- or bilateral sensorineural hearing loss. There have been no further descriptions in the literature since 1971.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Telfer-Sugar-Jaeger syndrome
Zespół Telfera, Sugar i Jaegera

Kod ORPHA
2885

Kod OMIM
172850

Kod ICD10
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Kod ICD11
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*Źródło

orphanet