Opis choroby *

Definicja

A rare neurologic disease characterized by spastic paraparesis presenting in late childhood and hearing loss. Additional features may include retinal anomalies, lenticular opacities, short stature, hypogonadism, sensory deficits, tremor, dysdiochokinesia, elevated cerebrospinal fluid protein, and absent or prolonged somatosensory evoked potentials. Plasma and fibroblast levels of saturated very long-chain fatty acids are normal. There have been no further descriptions in the literature since 1986.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Spastic paraparesis-hearing loss syndrome
Zespół Wellsa i Jankovica
Wells-Jankovic syndrome

Kod ORPHA
2815

Kod OMIM
312910

Kod ICD10
G11.4

Kod ICD11
LD2H.Y

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*Źródło

orphanet