

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

Kod ORPHA	Kod OMIM	Kod ICD10
2815	312910	G11.4

Kod ICD11
LD2H.Y

*Źródło

orphanet