

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
Zespół Wellsa i Jankovica
Wells-Jankovic syndrome

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

*Źródło

orphanet