Opis choroby *

Definicja

X-linked intellectual disability-limb spasticity-retinal dystrophy-diabetes insipidus syndrome is a rare genetic neurometabolic disease characterized by severe intellectual disability, spastic quadraparesis, Leber´s congenital amaurosis and diabetes insipidus. Additional manifestations include facial dysmorphy (dolichocephalic skull, hypertelorism, deep-set eyes, hypoplastic nares, low-set ears), short stature, truncal hypotonia and axial hypertonia. Brain anomalies (e.g. thin corpus callosum with lack of isthmus and tapered splenium, hypoplasia or atrophy of the optic chiasm, prominent lateral ventricles, diminished white matter), described on magnetic resonance imaging, have been reported. High prenatal α-fetoprotein and intrauterine growth restriction is observed in routine pregnancy examination.

Dane

Klasyfikacja
Choroba

Kod ORPHA
423479

Kod OMIM
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Kod ICD10
E79.8

Kod ICD11
LD90

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*Źródło

orphanet