Opis choroby *

Definicja

X-linked intellectual disability-short stature-overweight syndrome is a multiple congenital anomalies syndrome characterized by borderline to severe intellectual disability, speech delay, short stature, elevated body mass index, a pattern of truncal obesity (reported in older males), and variable neurologic features (e.g. hypotonia, tremors, gait disturbances, behavioral problems, and seizure disorders). Less common manifestations include microcephaly, microorchidism and/or microphallus. Dysmorphic features have been reported in some patients but no consitent pattern has been noted.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA 457240

Kod OMIM 300957

Kod ICD10 O87.8

Kod ICD11

LD90

*Źródło

orphanet