Opis choroby *

Definicja

A rare X-linked syndromic intellectual disability which in symptomatic, female carriers is characterized by a highly variable phenotype including facial dysmorphisms (prominent forehead, hypertelorism, down-slanting palpebral fissures, epicanthic folds, thick lips with everted lower vermilion, thick nasal alae, and septum), short hands with tapering fingers, short stature and skeletal findings (progressive kyphoscoliosis). Intellectual disability is mild to moderate, but intellect can also be normal. A high rate of psychiatric disorders has also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
276630

Kod OMIM
-

Kod ICD10
Q87.0

Kod ICD11
-

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*Źródło

orphanet