Opis choroby *

Definicja

A rare, genetic, X-linked syndromic intellectual disability disorder characterized by moderate to severe intellectual disability associated with epilepsy, short stature, autistic features and behavioral problems, such as self injury and aggressive outbursts. Observed facial dysmorphism includes brachycephaly, prominent supraorbital ridges, and deep set eyes. Additional variable manifestations include malposition of feet, asthenic habitus, hyporeflexia, bowel occlusions, hydronephrosis, ren arcuatus, delayed motor development and disturbed sleep-wake cycle.

Dane

Klasyfikacja
Choroba

Kod ORPHA
364028

Kod OMIM
300699

Kod ICD10
F72

Kod ICD11
LD90

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*Źródło

orphanet