Opis choroby *

Definicja

A rare severe, X-linked, neurodevelopmental disorder characterized by rapid developmental regression in infancy, partial or complete loss of purposeful hand movements, loss of speech, gait abnormalities, and stereotypic hand movements, commonly associated with deceleration of head growth, severe intellectual disability, seizures, and breathing abnormalities. The disorder has a progressive clinical course and may associate various comorbidities including gastrointestinal diseases, scoliosis, and behavioral disorders.

Dane

Klasyfikacja
Choroba

Kod ORPHA
778

Kod OMIM
312750

Kod ICD10
F84.2

Kod ICD11
LD90.4

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*Źródło

orphanet