

## Opis choroby \*

### Definicja

A rare, X-linked syndromic intellectual disability disorder characterized by non-progressive ataxia, apraxia, variable intellectual disability and/or visuospatial, visuographic and visuoconstructive dysfunctions in male patients. Seizures, congenital clubfoot and macroorchidism have also been associated. Partial clinical expression was noted in obligate female carriers. There have been no further descriptions in the literature since 1992.

### Dane

### Klasyfikacja

Choroba

#### Kod ORPHA

85338

#### Kod OMIM

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#### Kod ICD10

G31.8

#### Kod ICD11

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### \*Źródło

orphanet