## 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