

Opis choroby *

Definicja

Female restricted epilepsy with intellectual disability is a rare X-linked epilepsy syndrome characterized by febrile or afebrile seizures (mainly tonic-clonic, but also absence, myoclonic, and atonic) starting in the first years of life and, in most cases, developmental delay and intellectual disability of variable severity. Behavioral disturbances (e.g. autistic features, hyperactivity, and aggressiveness) are also frequently associated. This disease affects exclusively females, with male carriers being unaffected, despite an X-linked inheritance.

Dane

Klasyfikacja

Choroba

Synonimy

EFMR

EFMR

Zespół Juberga i Hellmana

Juberg-Hellman syndrome

Kod ORPHA

101039

Kod OMIM

300088

Kod ICD10

G40.8

Kod ICD11

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*Źródło

orphanet