Opis choroby *

Definicja

A rare X-linked syndromic intellectual disability characterized by intellectual impairment of variable severity, progressive lower limb spasticity, and diffuse palmoplantar hyperkeratosis. Additional manifestations include pes cavus, extensor plantar responses, hand tremor, and mild dysmorphic facial features.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Fitzsimmons-McLachlan-Gilbert syndrome
Zespół Fitzsimmonsa, McLachlan i Gilberta

Kod ORPHA
2824

Kod OMIM
309560

Kod ICD10
G82.1

Kod ICD11
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*Źródło

orphanet