Opis choroby *

Definicja

46,XY gonadal dysgenesis-motor and sensory neuropathy syndrome is a rare, genetic, developmental defect during embryogenesis disorder characterized by partial (unilateral testis, persistence of Müllerian duct structures) or complete (streak gonads only) gonadal dysgenesis, usually manifesting with primary amenorrhea in individuals with female phenotype but 46,XY karyotype, and sensorimotor dysmyelinating minifascicular polyneuropathy, which presents with numbness, weakness, exercise-induced muscle cramps, sensory disturbances and reduced/absent deep tendon reflexes. Germ cell tumors (seminoma, dysgerminoma, gonadoblastoma) may develop from the gonadal tissue.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
168563

Kod OMIM
607080

Kod ICD10
Q56.1

Kod ICD11
LD2A.1

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*Źródło

orphanet