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
 Kod OMIM
 Kod ICD10

 168563
 607080
 Q56.1

Kod ICD11 LD2A.1

*Źródło

orphanet