

Opis choroby *

Definicja

Genitopatellar syndrome is a rare congenital patellar anomaly syndrome characterized by patellar aplasia or hypoplasia associated with microcephaly, characteristic coarse facial features (microcephaly, bitemporal narrowing, large, broad nose with high nasal bridge, prominent cheeks and micro/retrognathia or prognathism), arthrogryposis of the hips and knees, urogenital abnormalities and intellectual deficiency.

Dane

Klasyfikacja

Zespół wad wrodzonych
Absent patellae-scrotal hypoplasia-renal anomalies-facial dysmorphism-intellectual disability syndrome
Brak rzeplki - hipoplazja moszny - wady nerek - dysmorfia twarzy - niepełnosprawność intelektualna

Kod ORPHA

85201

Kod OMIM

606170

Kod ICD10

Q87.8

Kod ICD11

LD2F.1Y

*Źródło

orphanet