

## 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

#### Synonimy

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

#### Kod ORPHA

85201

#### Kod OMIM

606170

#### Kod ICD10

Q87.8

#### Kod ICD11

LD2F.1Y

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#### \*Źródło

orphanet