

## Opis choroby \*

### Definicja

A rare autosomal recessive disorder characterized by the occurrence of cutaneous and subcutaneous calcified masses, usually adjacent to large joints, such as hips, shoulders and elbows. It can occur in the setting of hyperphosphatemia or normophosphatemia, depending on the type of gene mutation involved.

### Dane

#### Klasyfikacja

Podtyp kliniczny

#### Synonimy

Hypercalcemic tumoral calcinosis

GALNT3-CDG

Hiperkalcemiczna kalcynoza guzowata

#### Kod ORPHA

306661

#### Kod OMIM

617994

#### Kod ICD10

M11.2

#### Kod ICD11

-

---

#### \*Źródło

orphanet