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
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*Źródło

orphanet