Familial hyperphosphatemic tumoral calcinosis/Hyperphosphatemic hyperostosis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Familial hyperphosphatemic tumoral calcinosis/Hyperphosphatemic hyperostosis syndrome. ORPHA:306661

Familial tumoral calcinosis (FTC) refers to 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. FTC can occur in the setting of hyperphosphatemia or normophosphatemia, depending on the type of gene mutation involved.