DISEASE:
Familial hyperphosphatemic tumoral calcinosis/Hyperphosphatemic hyperostosis syndrome

NAME:	Familial hyperphosphatemic tumoral calcinosis/Hyperphosphatemic hyperostosis syndrome
DESCRIPTION:	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.
ORPHACODE:	306661
SYNONYMS:	Hypercalcemic tumoral calcinosis
XREF(S):	Orphanet OMIM ICD-10 OMIM OMIM
ANALYTE(S):	FGF23 GALNT3 KL
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Analytes

- fibroblast growth factor 23
- polypeptide N-acetylgalactosaminyltransferase 3
- klotho