Hyperphosphatemic Familial Tumoral Calcinosis

National Cancer Institute

Source

National Cancer Institute. Hyperphosphatemic Familial Tumoral Calcinosis. NCI Thesaurus. Code C131851.

An autosomal recessive disorder caused by loss-of-function mutation(s) in the GALNT3, FGF23, or KL gene, which encode polypeptide N-acetylgalactosaminyltransferase 3, fibroblast growth factor 23, and klotho, respectively. This condition, the biochemical hallmark of which is hyperphosphatemia caused by increased renal phosphate absorption, is characterized by the progressive deposition of calcium phosphate crystals in periarticular spaces, soft tissues, and/or bone.