

Disease

Hyperphosphatemic familial tumoral calcinosis 2^{1, 2, 3}

Gene

FGF23

Inheritance	AR
Current Treatments	dietary restriction, antacids, phosphate binders, acetazolamide, hemodialysis
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum phosphate, calcium, PTH, alkaline phosphatase, vitamin D serum levels, urine calcium, phosphate levels, plasma levels of the C-terminal portion of the phosphate-regulating hormone, fibroblast growth factor 23 ⁴
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=familial+tumoral+calcinosis&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/617993>
- ² <https://pubmed.ncbi.nlm.nih.gov/15340747/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/29548779/>
- ⁴ <https://www.ncbi.nlm.nih.gov/books/NBK476672/>