

Disease

Hyperphosphatemic familial tumoral calcinosis 1^{1, 2}

Gene

GALNT3

Inheritance	AR
Current Treatments	dietary restriction, phosphate binders, acetazolamide
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 ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=familial+tumoral+calcinosis&term=&cntry=&state=&city=&dist=

References

¹ <https://omim.org/entry/211900>

² <https://www.ncbi.nlm.nih.gov/books/NBK476672/>