

Disease

Vitamin D-dependent rickets, type IA^{1, 2}

Gene

CYP27B1

Inheritance	AR
Current Treatments	vitamin D as calcitriol
Evidence for Treatment	guideline ³
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum calcium, parathyroid hormone, 1,25-dihydroxy vitamin D levels ⁴
Disease Frequency (per 100,000)	0.3 ⁵
Genes Included in Disease Frequency	CYP27B1
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=vitamin+d+dependent&term=&cntry=&state=&city=&dist=

References

- ¹ <https://pubmed.ncbi.nlm.nih.gov/27060335/>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/31261480>
- ³ <https://pubmed.ncbi.nlm.nih.gov/32596195/>
- ⁴ <https://omim.org/clinicalSynopsis/264700>
- ⁵ <https://www.ncbi.nlm.nih.gov/pubmed/27777911>