

Disease

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

Gene

CYP2R1

Inheritance	AR
Current Treatments	calcifediol (25-OH-D3)
Evidence for Treatment	guideline ³
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum calcium, parathyroid hormone, 25-hydroxy vitamin D levels ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Vitamin+D-Dependent+Rickets%2C+Type+1B&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/600081>
- ² <https://pubmed.ncbi.nlm.nih.gov/28548312/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/32596195/>