

Disease

Hypophosphatasia^{1, 2}

Gene

ALPL

Inheritance	AR and AD
Current Treatments	tissue-nonspecific alkaline phosphatase (TNSALP) enzyme replacement therapy - asfotase alfa, avoid bisphosphonates
Evidence for Treatment	guideline ^{3, 4}
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum pyridoxal 5'-phosphate (PLP), serum unfractionated alkaline phosphatase (ALP) activity ¹
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC004
Disease Frequency (per 100,000)	1 ⁵
Genes Included in Disease Frequency	ALPL
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=ALPL&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1150/>
- ² <https://pubmed.ncbi.nlm.nih.gov/34884378/>
- ³ <https://www.ncbi.nlm.nih.gov/pubmed/31413732>
- ⁴ <https://www.ncbi.nlm.nih.gov/pubmed/30811537>
- ⁵ <https://ghr.nlm.nih.gov/condition/hypophosphatasia#statistics>