

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** ALPL Gene Deletion/Duplication Analysis
- **Test Code:** 5649

Related Diseases & Genes:

- “Hypophosphatasia, adult” (OMIM #: 146300) || Gene: *ALPL* (OMIM #: 171760)
- “Hypophosphatasia, childhood” (OMIM #: 241510)
- “Hypophosphatasia, infantile” (OMIM #: 241500)
- Odontohypophosphatasia (OMIM #: 146300)

Synonyms:

- “ALKALINE PHOSPHATASE, LIVER/BONE/KIDNEY TYPE”
- “ALKALINE PHOSPHATASE, TISSUE-NONSPECIFIC”
- TNSALP
- TNAP

Test Details:

- **Methodology:** MLPA
- **Performed:** Everyday
- **Reported:** 4 weeks

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 3 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Inborn Errors of Metabolism

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