



**Supplementary Figure 1. Identification of mutations in *ALPL* in odontohypophosphatasia kindred.**

Sequencing data and PCR analysis for 1318\_20ACC deletion (p.N440del) in the *ALPL* gene.

Electropherogram representative of DNA sequencing analysis of exon 12 in **(A)** the mother (control sequence), and **(B)** probands, revealing a three base pair in-frame deletion (AAC) at 1318-20-nt position, corresponding to codon 440 of protein that encodes asparagine (N440). Arrow indicates the initial position of the 1318\_20ACC deletion corresponding to the point where the sequence became truncated.

**(C)** Differential amplification by PCR of native TNAP (**TNAP**) and mutant (**1318\_20delAAC**) alleles.

Products of differential amplification of native TNAP and mutant alleles from Mother (**M**), Father (**F**) and probands (**PA** and **PB**) were visualized by ethidium bromide staining after 1.5% agarose gel electrophoresis. The mother was normal homozygous, while the father and the probands were heterozygous for 1318\_20delAAC (p.N440del) genotype, exhibiting both alleles.