



## Supplementary Material

# A Recurrent Mutation in *CTSK* Gene is Responsible for Autosomal Recessive Pycnodysostosis in Consanguineous Pakistani Families

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Supplementary Table I.- Mutations in the *CTSK* gene known for causing pycnodysostosis among Pakistani population.

Genotype	Amino acid change	Type of mutation	Location in DNA sequence of <i>CTSK</i>	Reported by
c.136C > T	p.Arg46Trp	Missense	Exon 3	Schilling et al., 2007
c.282dupA	p.Val95SerfsX9	Frameshift	Exon 4	Donnarumma et al., 2007
c.728G > A	p.Gly243Glu	Missense	Exon 6	Khan et al., 2010
c.749A > G	p.Asp250Gly	Missense	Exon 7	Donnarumma et al., 2007
c.830C > T	p.Gly262AlafsX70	Frameshift	Exon 7	Donnarumma et al., 2007; Naeem et al., 2009
c.955G > T	p.Gly319Cys	Missense	Exon 8	Donnarumma et al., 2007

**Supplementary Table II.- Primer sequences used for the amplification of *CTSK* gene.**

Gene	Primer sequence
CTSK_E1F	GTCTTGTTATTTGCCACCAAAG
CTSK_E1R	GGCTAAAAGTTTATTGTGATCCTC
CTSK_E2F	CTCTGTTCCCTGCCAAATG
CTSK_E2R	CTCAGGTCTCAGCCTTCCTG
CTSK_E3+4F	CCTTTATTCTCCTAAAACCCTTCA
CTSK_E3+4R	AGCAGAAAAAGGAAAAGGTCA
CTSK_E5F	GAATTTCTTCAGGCAGGTAGAG
CTSK_E5R	TCCAGTACAAAACATCATGCTG
CTSK_E6F	TCACTGACTCATTGCCTATTGC
CTSK_E6R	CCCAAGGTCCTTCGAGAAAC
CTSK_E7F	TGGGAGATCCCACAGGG
CTSK_E7R	AAGCTGGAGGTGAGGTTGAG
CTSK_E8F	GTGTACCATCAGTACCTCGCAC
CTSK_E8R	CTCCAAAGTGCATCGTTACAC