

Novel *KLK4* Mutations Cause Hypomaturation Amelogenesis Imperfecta

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Table S1. Statistics for exome sequencing.

Sample	Total reads	Mapping rate (%)	Median target coverage	Coverage of target region (%)	Fraction of target covered with at least		
					20X	10X	
Family 1 V:2	74,485,830	98.2	70	96.4	91.6	94.9	
Family 2	III:4	123,184,317	99.6	92	96.2	92.9	94.9
	III:5	126,406,940	99.9	95	96.1	93.3	95.0
	IV:2	89,232,516	99.6	66	96.2	90.4	94.3
Family 3	III:6	198,709,274	99.7	120	99.5	97.8	98.9
	IV:1	125,774,962	99.4	73	99.4	95.7	98.4
Family 4	III:3	99,282,916	99.4	63	99.4	94.6	98.1
	III:4	136,200,546	98.9	74	99.4	96.4	98.5
	IV:2	142,866,102	99.2	80	99.3	96.6	98.5

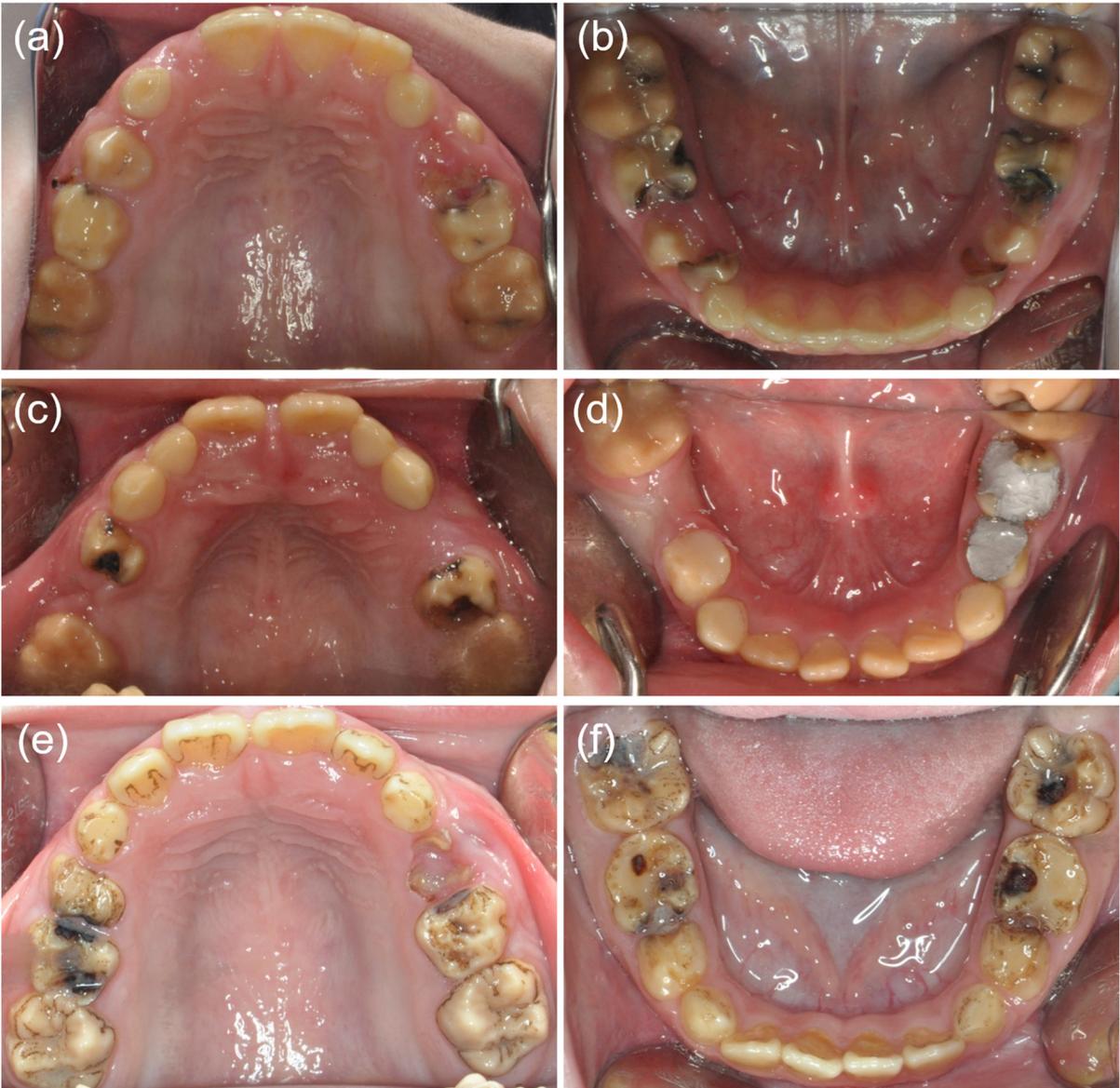


Figure S1. (a,b) Clinical photos of the proband in family 1. (c,d) Clinical photos of the proband in family 2. (e,f) Clinical photos of the proband in family 3.

KLK4:NM_004917.4:c.170C>A:p.(Ser57*)

Wt ACGAATTGTTCTGCTCGGGCGTCCTGGTG
Mt ACGAATTGTTCTGCTAGGGCGTCCTGGTG

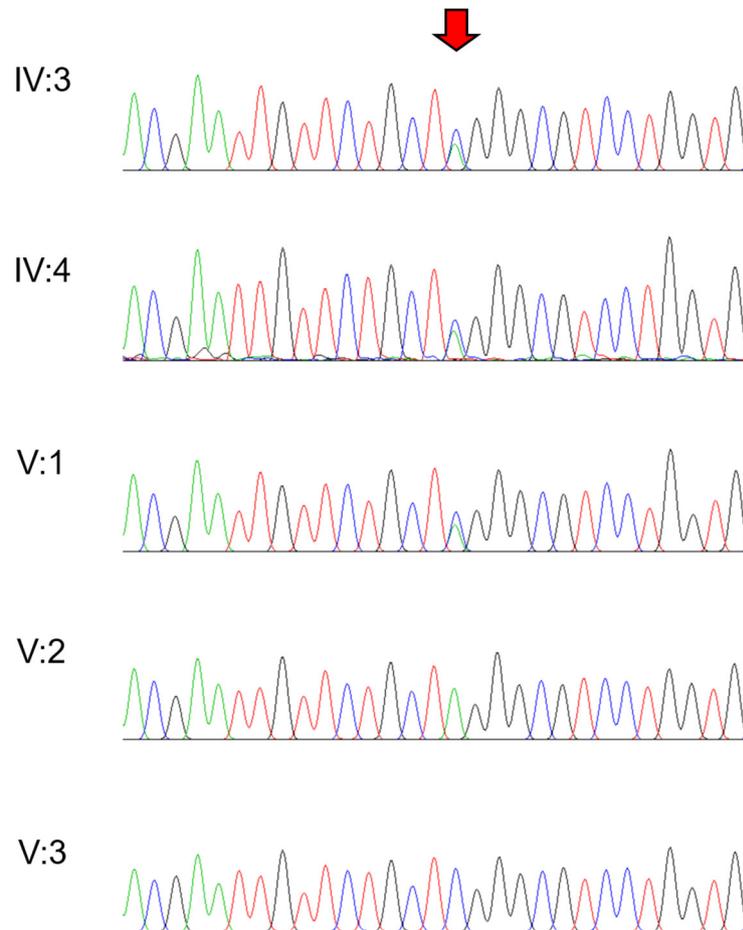


Figure S2. Sequencing chromatograms of the participating individuals of family 1. Wild type (Wt) and mutant (Mt) nucleotide sequences are shown above the chromatograms. Nucleotides affected by the mutation are underlined. The location of the mutation is indicated with a red arrow. Individual identifications are indicated on the left side of each chromatogram.

KLK4:NM_004917.4:c.170C>A:p.(Ser57*)

Wt **ACGAATTGTTCTGCTCGGGCGTCCTGGTG**
Mt **ACGAATTGTTCTGCTAGGGCGTCCTGGTG**

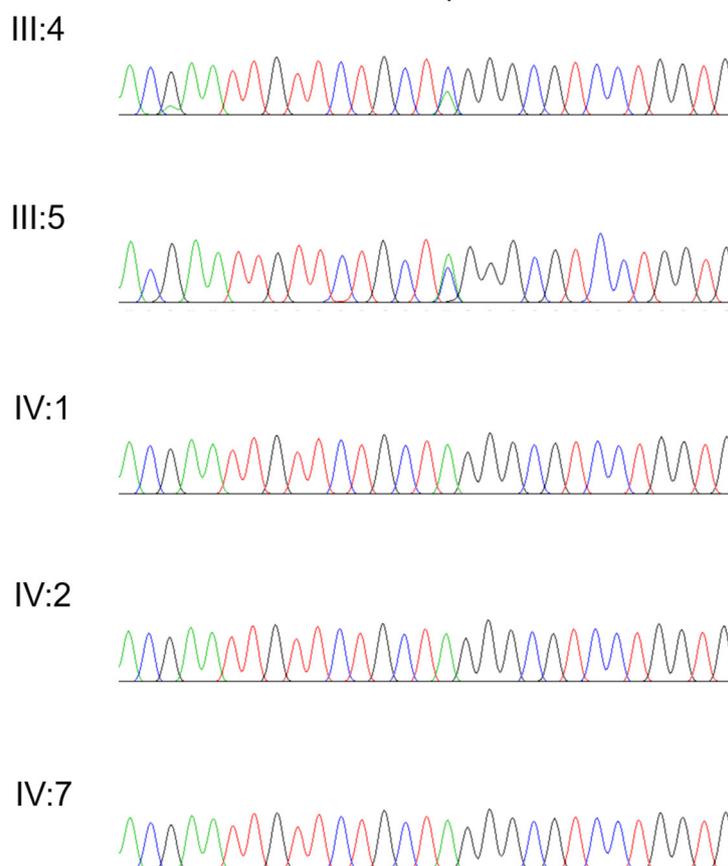


Figure S3. Sequencing chromatograms of the participating individuals of family 2. Wild type (Wt) and mutant (Mt) nucleotide sequences are shown above the chromatograms. Mutated nucleotide is underlined. The location of the mutation is indicated with a red arrow. Individual identifications are indicated on the left side of each chromatogram.

KLK4:NM_004917.4:c.170C>A:p.(Ser57*)

Wt ACGAATTGTTCTGCTCGGGCGTCCTGGTG

Mt ACGAATTGTTCTGCTAGGGCGTCCTGGTG

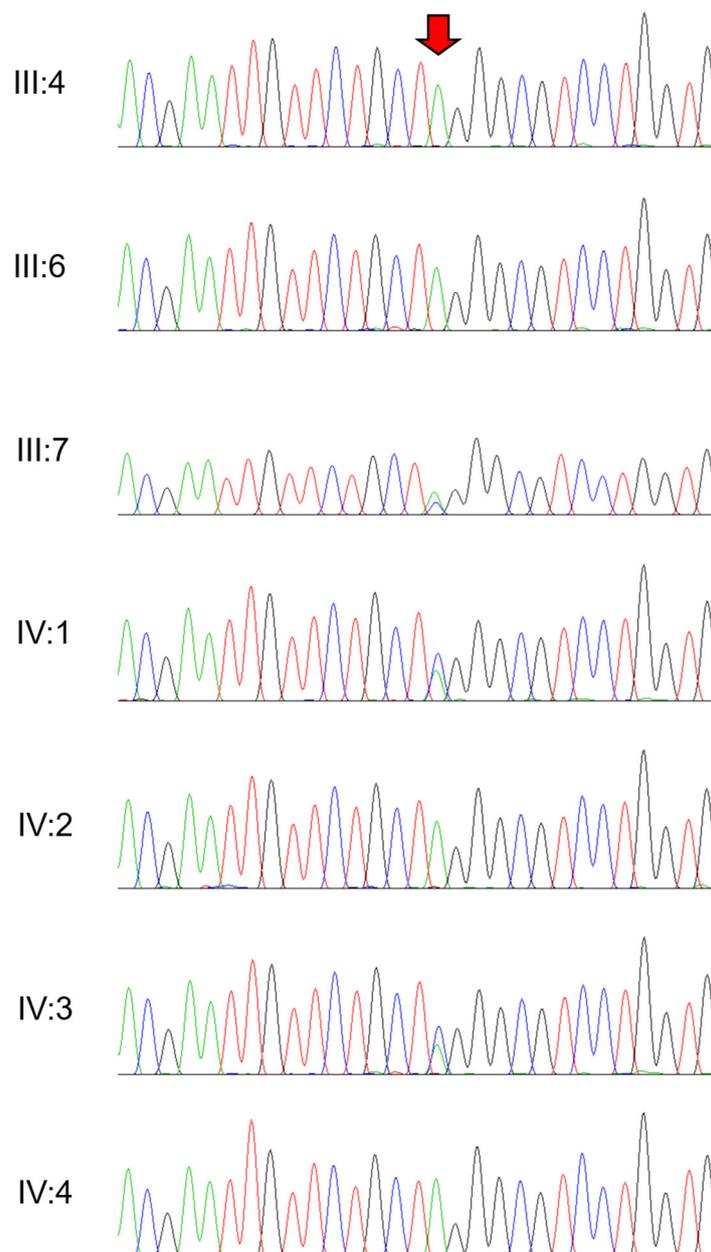


Figure S4. Sequencing chromatograms of the participating individuals of family 3. Wild type (Wt) and mutant (Mt) nucleotide sequences are shown above chromatograms. Mutated nucleotide is underlined. The location of the mutation is indicated with a red arrow. Individual identifications are indicated on the left side of each chromatogram.

KLK4:NM_004917.4:c.637T>C;p.(Cys213Arg)

Wt GGGGGGCCCCCTGATCTGCAACGGGTACTT
Mt GGGGGGCCCCCTGATCCGCAACGGGTACTT

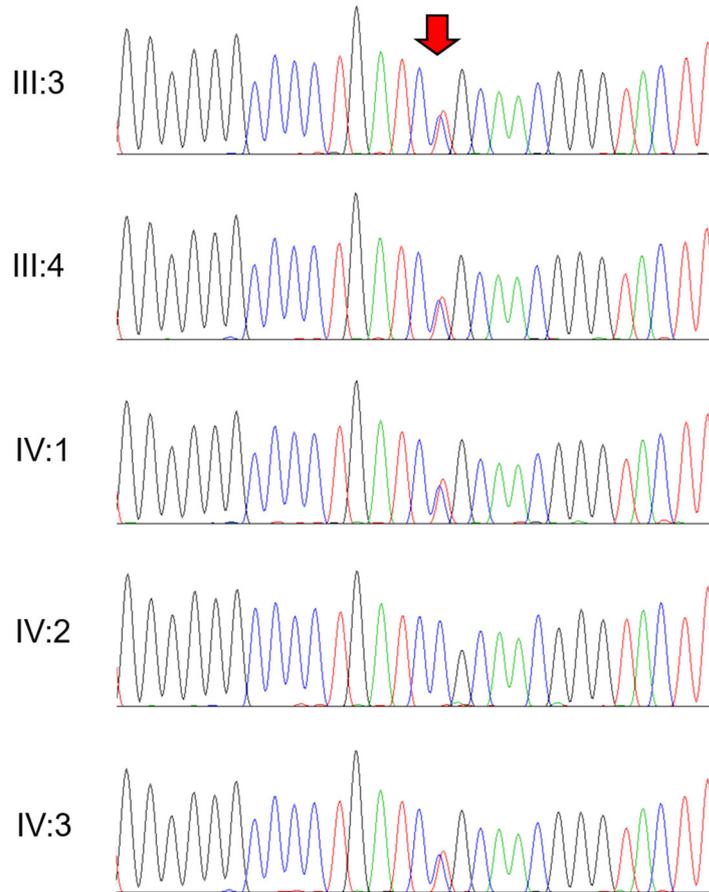


Figure S5. Sequencing chromatograms of the participating individuals of family 4. Wild type (Wt) and mutant (Mt) nucleotide sequences are shown above chromatograms. Mutated nucleotide is underlined. The location of the mutation is indicated with a red arrow. Individual identifications are indicated on the left side of each chromatogram.