Supporting Information

Enamel malformations associated with a defined *DSPP* mutation in two families

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Figure S1

Alignment of human dentin phosphoprotein (DPP) length haplotypes noting the positions of disease-causing mutations. The positions of many *DSPP* insertions-deletions (indels) are sometimes flexible. That is, they could be moved left or right in the alignment without increasing nucleotide mismatches. We elected to position the indels as annotated in Table 2 of SONG et al. (1). Indels reported by MCNIGHT et al. (2) were slid along the sequence to align with those from SONG et al., when possible, or were positioned to minimize mismatches.

The *DSPP* cDNA reference sequence (RefSeq) (NM_014208.3) encodes a DPP region having 839 amino acids. Insertions and deletions of this sequence that maintain the reading frame do not appear to cause a phenotype. The RefSeq nucleotide sequence is numbered starting with the A in the *DSPP* translation initiation codon (ATG), which is nucleotide 120 in NM_014208.3. The amino acids are numbered from the start of the DSPP protein at Met1 and refer to the number in the reference (not merged) sequence.

MCKNIGHT et al. reported 38 different haplotypes derived from 188 normal human chromosomes. Twenty indels among the 38 haplotypes were arranged in 22 different patterns. The 38 McKnight haplotypes were grouped according to their indel patterns based upon the MCKNIGHT et al. Supplementary Table S4B. One haplotype from each group was used in the alignment. The grouped haplotypes are in brackets with the aligned haplotype sequence in bold: [1, 7, 16, **35** (791 aa)], [2, 23, **28** (788 aa)], **3** (845 aa), **4** (782 aa), **5** (785 aa), **6** (785 aa), **8** (791 aa), [9, **14** (same as RefSeq; 839 aa)], [10, 12, 15, 17, 18, **34** (788 aa)], [11, **20** (788 aa)], **13** (779 aa), [19, 21, **22** (797 aa)], [23, **28** (788 aa)], **24** (902 aa), **25** (842 aa), **26** (779 aa), [27, **31**] (788 aa), **29** (785 aa), [30, **36** (794 aa)], **32** (788 aa), **33** (758 aa), **37** (770 aa), **38** (782 aa).

The McNight haplotype sequences were imported from GenBank and provided the haplotype sequence starting with *DSPP* nucleotide 2572 encoding Arg^{396}. For the purpose of this analysis, the reference sequence (NM_014208.3) was used to complete the McKnight DPP sequences 5' to this position, with the exception of Clone 37, where the short sequence 5' to nucleotide 2572 was obtained from Hap37A to ensure correct positioning of indel 1.

The SONG et al. haplotype sequences were not available in GenBank. These sequences were obtained by using the *DSPP* cDNA reference sequence (NM_014208.3) and changing it according to SONG et al., Tables 2 and 3. SONG et al. reported 15 haplotypes that showed 13 different indel patterns. One Song haplotype from each of the

13 groups was used in the alignment: **1** (803 aa), **2** (839 aa), **3** (788 aa), **4** (788 aa), **5** (790 aa), [**6**, **6T**, (797 aa)], [**6(2)**, **6(2)T** (796 aa)], **7** (782 aa), **102** (791 aa), **130** (794 aa), **106** (806 aa), **72** (785 aa), **110** (782 aa). Song haplotype 2 has the same indel pattern as McKnight haplotypes [9, 14] and the reference sequence. Song haplotype 3 has the same indel pattern as McKnight haplotypes [19, 21, 22]. There are 33 unique indel patterns reported by these two studies, with the DPP protein ranging from 770 to 902 amino acids.

In this alignment, the positions of single nucleotide polymorphisms listed in SONG et al. are noted. The five frameshift mutations listed in Table 1 of SONG et al. (1), the four frameshifts shown in Table 1 of MCKNIGHT et al. (2), the single frameshift mutation reported in MCKNIGHT et al. (3), the nine frameshifts reported by NIEMINEN et al. (4) and the two frameshifts reported by LEE et al. (5)

References

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2. MCKNIGHT DA, SUZANNE HART P, HART TC, HARTSFIELD JK, WILSON A, WRIGHT JT, FISHER LW. A comprehensive analysis of normal variation and disease-causing mutations in the human dspp gene. *Hum Mutat* 2008; **29**: 1392-1404.
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4. NIEMINEN P, PAPAGIANNOULIS-LASCARIDES L, WALTIMO-SIREN J, OLLILA P, KARJALAINEN S, ARTE S, VEERKAMP J, WALTON VT, KUSTNER EC, SILTANEN T, HOLAPPA H, LUKINMAA PL, ALALUUSUA S. Frameshift mutations in dentin phosphoprotein and dependence of dentin disease phenotype on mutation location. *J Bone Miner Res* 2011; **2010**: 14.
5. LEE KE, KANG HY, LEE SK, YOO SH, LEE JC, HWANG YH, NAM KH, KIM JS, PARK JC, KIM JW. Novel dentin phosphoprotein frameshift mutations in dentinogenesis imperfecta type ii. *Clin Genet* 2010: 8.
6. DONG J, GU T, JEFFORDS L, MACDOUGALL M. Dentin phosphoprotein compound mutation in dentin sialophosphoprotein causes dentinogenesis imperfecta type iii. *Am J Med Genet* 2005; **132**: 305-309.

```
RefSeq          gatg atcccaatag cagtgatgaa tctaatggca atgatgatgc taattcagaa 1440
Merged          gatg atcccaatag cagtgatgaa tctaatggca atgatgatgc taattcagaa
RefSeq   (463)   D   D  P  N  S   S  D  E   S  N  G   N  D  D  A   N  S  E   480

RefSeq    agtgacaata acagcagtag ccgaggagat gcttcttata actctgatga atcaaaagat 1500
Merged    agtgacaata acagcagtag ccgaggagat gcttcttata actctgatga atcaaaagat
RefSeq     S  D  N   N  S  S  S   R  G  D   A  S  Y   N  S  D  E   S  K  D   500

RefSeq    aatggcaatg gcagtgactc aaaaggagca gaagatgatg acagtgatag cacatcagac 1560
Merged    aatggcaatg gcagtgactc aaaaggagca gaagatgatg acagtgatag cacatcagac
RefSeq     N  G  N   G  S  D  S   K  G  A   E  D  D   D  S  D  S   T  S  D   520

RefSeq    actaataata gtgacagtaa tggcaatggt aacaatggga atgatgacaa tgacaaatca 1620
Merged    actaataata gtgacagtaa tggcaatggt aacaatggga atgatgacaa tgacaaatca
RefSeq     T  N  N   S  D  S  N   G  N  G   N  N  G   N  D  D  N   D  K  S   540

RefSeq    gacagtggca aaggtaaatc agatagcagt gacagtgata gtagtgatag cagcaatagc 1680
Merged    gacagtggca aaggtaaatc agatagcagt gacagtgata gtagtgatag cagcaatagc
RefSeq     D  S  G   K  G  K  S   D  S  S   D  S  D   S  S  D  S   S  N  S   560

RefSeq    agtgatagta gtgacagcag tgacagtgac agcagtgata gcaacagtag cagtgatagt 1740
Merged    agtgatagta gtgacagcag tgacagtgac agcagtgata gcaacagtag cagtgatagt
RefSeq     S  D  S   S  D  S  S   D  S  D   S  S  D   S  N  S  S   S  D  S   580
```

Note: Dentin dysplasia II mutation c.1686delT; p.D562EfsX1313 (Nieminen, kindred 1).

```
RefSeq    gacagcagtg acagtgacag cagtgatagc agtgacagtg atagtagtga tagcagcaat 1800
Merged    gacagcagtg acagtgacag cagtgatagc agtgacagtg atagtagtga tagcagcaat
RefSeq     D  S  S   D  S  D  S   S  D  S   S  D  S   D  S  S  D   S  S  N   600

RefSeq    agcagtgaca gtagtgacag cagtgatagc agtgacagta gtgatagtag tgacagcagt 1860
Merged    agcagtgaca gtagtgacag cagtgatagc agtgacagta gtgatagtag tgacagcagt
RefSeq     S  S  D   S  S  D  S   S  D  S   S  D  S   S  D  S  S   D  S  S   620
```

Note: Dentin dysplasia II mutation c.1830delC; p.S610RfsX1313 (Nieminen, kindred 2).

```
RefSeq    gacagcaagt cagacagcag caaatcagag agcgacagca gtgatagtga cagtaagtca 1920
Merged    gacagcaagt cagacagcag caaatcagag agcgacagca gtgatagtga cagtaagtca
RefSeq     D  S  K   S  D  S  S   K  S  E   S  D  S   S  D  S  D   S  K  S   640
```

Notes: Dentin dysplasia II mutation c.1870_1873delTCAG; p.K623fsX1312 (McKnight).
Dentin dysplasia II mutation c.1918_1921delTCAG; p.K639fsX1312 (McKnight).
Dentin dysplasia II mutation c.1918_1921delTCAG; p.K639fsX1312 (Nieminen, kindreds 3-5).

```
RefSeq    gacagcagtg acagcaacag cagtgacagt agtgacaaca gtgatagcag cgacagcagc 1980
Merged    gacagcagtg acagcaacag cagtgacagt agtgacaaca gtgatagcag cgacagcagc
RefSeq     D  S  S   D  S  N  S   S  D  S   S  D  N   S  D  S  S   D  S  S   660
```

Notes: Dentin dysplasia II mutation c.1922-1925delACAG; p.D641AfsX1312 (Nieminen, kindred 6).
Snp1: c1971C>T; p.S557.

```
RefSeq    aatagcagta acagcagtga tagtagtgac agcagtgata gcagtgacag cagcagtagc 2040
Merged    aatagcagta acagcagtga tagtagtgac agcagtgata gcagtgacag cagcagtagc
RefSeq     N  S  S   N  S  S  D   S  S  D   S  S  D   S  S  D  S   S  S  S   680
```

Note: Dentin dysplasia II mutation c.2040delC; p.S680fsX1313 (Song)

```
RefSeq  agtgacagca gca------------------acagcag tgatagtagt gacagtagtg acagcagcaa
SHap1:  agtgacagca gcagtagcagtgacagcagcaacagcag tgatagtagt gacagtagtg acagcagcaa
SHap2:  agtgacagca gca------------------acagcag tgatagtagt gacagtagtg acagcagcaa
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SHap3:   agtgacagca gca------------------acagcag tgatagtagt gacagtagtg acagcagcaa
SHap4:   agtgacagca gca------------------acagcag tgatagtagt gacagtagtg acagcagcaa
SHap5:   agtgacagca gcagtagcagtgacagcagcaacagcag tgatagtagt gacagtagtg acagcagcaa
SHap6:   agtgacagca gcagtagcagtgacagcagcaacagcag tgatagtagt gacagtagtg acagcagcaa
SHap6(2) agtgacagca gcagtagcagtgacagcagcaacagcag tgatagtagt gacagtagtg acagcagcaa
SHap7    agtgacagca gca------------------acagcag tgatagtagt gacagtagtg acagcagcaa
SHap102  agtgacagca gca------------------acagcag tgatagtagt gacagtagtg acagcagcaa
SHap130  agtgacagca gcagtagcagtgacagcagcaacagcag tgatagtagt gacagtagtg acagcagcaa
SHap106  agtgacagca gcagtagcagtgacagcagcaacagcag tgatagtagt gacagtagtg acagcagcaa
SHap72   agtgacagca gca------------------acagcag tgatagtagt gacagtagtg acagcagcaa
SHap110  agtgacagca gca------------------acagcag tgatagtagt gacagtagtg acagcagcaa
Merged   agtgacagca gcagtagcagtgacagcagcaacagcag tgatagtagt gacagtagtg acagcagcaa
Merged    S  D  S   S  S  S  S  D  S  S  N  S  S   D  S  S   D  S  S   D  S  S  N
```

Notes: Id1: c.2053_2054insGTAGCAGTGACAGCAGCA; p.684_N685insSSSDSS 697
Dentin dysplasia II mutation c.2063delA; p.D688VfsX1313 (Nieminen, kindreds 7 & 8).

```
RefSeq   tagcagtgag agcagtgata gtagtgacag cagtgatagt gacagcagtg atagtagtga 2150
Merged   tagcagtgag agcagtgata gtagtgacag cagtgatagt gacagcagtg atagtagtga
RefSeq     S  S  E   S  S  D   S  S  D  S   S  D  S   D  S  S   D  S  S  D  717

RefSeq   cagcagtaat agtaacagca gcgatagtga cagcagcaac agcagcgata gcagtgacag 2210
Merged   cagcagtaat agtaacagca gcgatagtga cagcagcaac agcagcgata gcagtgacag
RefSeq     S  S  N   S  N  S   S  D  S  D   S  S  N   S  S  D   S  S  D  S  737

RefSeq   cagcaacagc agtgacagca gtgatagcag tgacagcagc aacagcagtg acagtagcga 2270
Merged   cagcaacagc agtgacagca gtgatagcag tgacagcagc aacagcagtg acagtagcga
RefSeq     S  N  S   S  D  S   S  D  S  S   D  S  S   N  S  S   D  S  S  D  757

RefSeq   tagcagtgac agcagcaaca gcagtgacag cagtgatagc agtgacagca gtgatagtag 2330
Merged   tagcagtgac agcagcaaca gcagtgacag cagtgatagc agtgacagca gtgatagtag
RefSeq     S  S  D   S  S  N   S  S  D  S   S  D  S   S  D  S   S  D  S  S  777
```

Note: Dentinogenesis imperfecta type II mutation c.2272delA; p.S758fsX1313 (McKnight)

```
RefSeq   tgacagcagc aacagcagtg atagcaacga cagcagcaat agcagtgaca gcagtgatag 2390
Merged   tgacagcagc aacagcagtg atagcaacga cagcagcaat agcagtgaca gcagtgatag
RefSeq     D  S  S   N  S  S   D  S  N  D   S  S  N   S  S  D   S  S  D  S  797
```

Note: Dentinogenesis imperfecta type II mutation c.2349delT; p.S783RfsX1313 (Nieminen, kindred 9).

```
RefSeq   cagcaacagc agtgatagca gcaacagcag tgatagcagt gatagcagtg acagcagtga 2450
Merged   cagcaacagc agtgatagca gcaacagcag tgatagcagt gatagcagtg acagcagtga
RefSeq     S  N  S   S  D  S   S  N  S  S   D  S  S   D  S  S   D  S  S  D  817

RefSeq   tagcgacagc agcaatagca gtgacagcag taatagtagt gacagcagcg atagcagcaa 2510
SHap1:   tagcgacagc agcaatagca gtgacagcag taatagtagt gacagcagcg atagcagcaa
SHap2:   tagcgacagc agcaatagca gtgacagcag taatagtagt gacagcagcg atagcagcaa
SHap3:   tagcgacagc agcaatagca gtgacagcag taatagtagt gacagcagcg atagcagcaa
SHap4:   tagcgacagc agcaatagca gtgacagcag taatagtagt gacagcagcg atagcagcaa
SHap5:   tagcgacagc agcaatagca gtgacagcag taatagtagt gacagcagcg atagca---a
SHap6:   tagcgacagc agcaatagca gtgacagcag taatagtagt gacagcagcg atagcagcaa
SHap6(2) tagcgacagc agcaatagca gtgacagcag taatagtagt gacagcagcg atagca---a
SHap7    tagcgacagc agcaatagca gtgacagcag taatagtagt gacagcagcg atagcagcaa
SHap102  tagcgacagc agcaatagca gtgacagcag taatagtagt gacagcagcg atagcagcaa
SHap130  tagcgacagc agcaatagca gtgacagcag taatagtagt gacagcagcg atagcagcaa
SHap106  tagcgacagc agcaatagca gtgacagcag taatagtagt gacagcagcg atagcagcaa
SHap72   tagcgacagc agcaatagca gtgacagcag taatagtagt gacagcagcg atagcagcaa
SHap110  tagcgacagc agcaatagca gtgacagcag taatagtagt gacagcagcg atagcagcaa
Merged   tagcgacagc agcaatagca gtgacagcag taatagtagt gacagcagcg atagcagcaa
Merged     S  D  S   S  N  S   S  D  S  S   N  S  S   D  S  S   D  S  S  N  837
```

Note: Id2 is c.2507_2509delGCA; pS836fsX1313

```
RefSeq    cagcagtgat agca**g**cgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa 2570
Hap3      cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap4      cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap5      cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap6      cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap8      cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap13     cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap14     cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap20     cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap22     cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap24     cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap25     cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap26     cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap28     cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap29     cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap31     cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap32     cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap33     cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap34     cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap35     cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap36     cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Hap37     cagcagtgat agcagcgaca gcagcgatag cagtgac**a**gc agtgatagc- ----------
Hap38     cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
SHap1:    cagcagtgat agcagcgaca gcagcgatag cagtgac**a**gc agtgatagcg acagcagcaa
SHap2:    cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
SHap3:    cagcagtgat agcagcgaca gcagcgatag cagtgac**a**gc agtgatagcg acagcagcaa
SHap4:    cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
SHap5:    cagcagtgat agcagcgaca gcagcgatag cagtgac**a**gc agtgatagcg acagcagcaa
SHap6:    cagcagtgat agcagcgaca gcagcgatag cagtgac**a**gc agtgatagcg acagcagcaa
SHap6(2)  cagcagtgat agcagcgaca gcagcgatag cagtgac**a**gc agtgatagcg acagcagcaa
SHap7     cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
SHap102   cagcagtgat agcagcgaca gcagcgatag cagtgac**a**gc agtgatagcg acagcagcaa
SHap130   cagcagtgat agcagcgaca gcagcgatag cagtgac**a**gc agtgatagcg acagcagcaa
SHap106   cagcagtgat agcagcgaca gcagcgatag cagtgac**a**gc agtgatagcg acagcagcaa
SHap72    cagcagtgat agcagcgaca gcagcgatag cagtgac**a**gc agtgatagcg acagcagcaa
SHap110   cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagcg acagcagcaa
Merged    cagcagtgat agcagcgaca gcagcgatag cagtgacggc agtgatagc**g acagcagcaa**
Merged      S  S  D   S  S  D   S  S  D  S  S  D  G/**S** S  D  S   D  S  S  N   857
```

Notes: Dentinogenesis imperfecta type II mutation c.2525delG; p.S842fsX1313 (McKnight)
Snp2 is c.2548G>A p.G850S

```
RefSeq    tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag 2630
Hap3      tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap4      tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap5      tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap6      tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap8      tagaaggac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap13     tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap14     tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap20     tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap22     tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap24     tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap25     tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap26     tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap28     tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap29     tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap31     tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap32     tagaaggac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap33     tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap34     tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap35     tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap36     tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Hap37     ---------- ---------- -----gacag cagcgatagc agtgacagca gcaacagcag
Hap38     tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
SHap1:    tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
SHap2:    tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
SHap3:    tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
SHap4:    tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
SHap5:    tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
SHap6:    tagaagggac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
SHap6(2)  tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
SHap7     tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
SHap102   tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
SHap130   tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
SHap106   tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
SHap72    tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
SHap110   tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Merged    tagaagtgac agtagtaata gtagtgacag cagcgatagc agtgacagca gcaacagcag
Merged      R  S  D   S  S  N   S  S  D  S   S  D  S   S  D  S   S  N  S  S  877
```

Notes: This is the beginning of the Hap3 to Hap38 sequences.
Indel 1 (36 Nt) is only in McKnight haplotype 37.
Dentinogenesis imperfecta type II mutation c.2593delA; p.S865fsX1313 (Song).

```
RefSeq    tgacagcagt gatag---------cagtg acagcaacga aagcagcaat agcagtgaca 2680
Hap3      tgacagcagt gatag---------cagtg acagcaacga aagcagcaat agcagtgaca
Hap4      tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Hap5      tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Hap6      tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Hap8      tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Hap13     tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Hap14     tgacagcagt gatag---------cagtg acagcaacga aagcagcaat agcagtgaca
Hap20     tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Hap22     tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Hap24     tgacagcagt gatag---------cagtg acagcaacga aagcagcaat agcagtgaca
Hap25     tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Hap26     tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Hap28     tgacagcagt gatag---------cagtg acagcaacga aagcagcaat agcagtgaca
Hap29     tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Hap31     tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Hap32     tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Hap33     tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Hap34     tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Hap35     tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Hap36     tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Hap37     tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Hap38     tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
SHap1:    tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
SHap2:    tgacagcagt gatag---------cagtg acagcaacga aagcagcaat agcagtgaca
SHap3:    tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
SHap4:    tgacagcagt gatag---------cagtg acagcaacga aagcagcaat agcagtgaca
SHap5:    tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
SHap6:    tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
SHap6(2)  tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
SHap7     tgacagcagt gatag---------cagtg acagcaacga aagcagcaat agcagtgaca
SHap102   tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
SHap130   tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
SHap106   tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
SHap72    tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
SHap110   tgacagcagt gatag---------cagtg acagcaacga aagcagcaat agcagtgaca
Merged    tgacagcagt gatagtagtgacagcagtg acagcaacga aagcagcaat agcagtgaca
Merged      D  S  S   D  S  S  D  S  S   D  S  N  E   S  S  N   S  S  D  893
```

Notes: Indel 2 (9 Nt) is in 4 unique McKnight haplotypes: 3; RefSeq/9/14; 2/23/28; and 24.
Id3: c.2645_2646insTAGTGACAG; p.S82_883insSDS.
Indel 2 (McKnight) & Id3 (Song) are the same.
Dentinogenesis imperfecta type II mutation c.2666delG; p.S889TfsX1313 (Nieminen, kindred 10).

```
RefSeq    gca**g**tga**t**ag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca 2740
Hap3      gcagtgatag cagcaacagc agtga**c**agtg acagcagtga tagcagcaac agcagtgaca
Hap4      gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagt----
Hap5      gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagt----
Hap6      gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagt----
Hap8      gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
Hap13     gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagcgaca
Hap14     gcagtgatag cagcaacagc agtga**c**agtg acagcagtga tagcagcaac agcagtgaca
Hap20     gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagt----
Hap22     gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
Hap24     gcagtgatag cagcaacagc agtga**c**agtg acagcagtga tagcagcaac agcagtgaca
Hap25     gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
Hap26     gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
Hap28     gcagtgatag cagcaacagc agtga**c**agtg acagcagtga tagcagcaac agcagtgaca
Hap29     gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
Hap31     gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagt----
Hap32     gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagt----
Hap33     gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
Hap34     gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagcgaca
Hap35     gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
Hap36     gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagt----
Hap37     gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
Hap38     gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagt----
SHap1:    gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
SHap2:    gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
SHap3:    gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
SHap4:    gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
SHap5:    gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
SHap6:    gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
SHap6(2)  gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
SHap7     gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
SHap102   gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
SHap130   gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
SHap106   gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
SHap72    gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
SHap110   gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagtgaca
Merged    gcagtgatag cagcaacagc agtgatagtg acagcagtga tagcagcaac agcagt**gaca**
Merged    S  S  D  S   S  N  S   S  D  S   D  S  S  D   S  S  N   S  S  D   913
```

Notes: Dentinogenesis imperfecta type II mutation c.2684delG; p.S895fsX1313 (Song).
Indel 3 (9 Nt) is in 8 unique McKnight haplotypes: 4, 5, 6, 11/20, 27/31, 32, 30/36, & 38.
Dentinogenesis imperfecta type II mutation c.2688delT; p.D896fsX1276 (Lee:ClinGenet).

```
RefSeq    gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca 2800
Hap3      gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap4      -----gatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap5      -----gatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap6      -----gatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap8      gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap13     gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap14     gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap20     -----gatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap22     gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap24     gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap25     gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap26     gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap28     gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap29     gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap31     -----gatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap32     -----gatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap33     gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap34     gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap35     gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap36     -----gatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap37     gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Hap38     -----gatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
SHap1:    gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
SHap2:    gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
SHap3:    gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
SHap4:    gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
SHap5:    gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
SHap6:    gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
SHap6(2)  gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
SHap7     gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
SHap102   gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
SHap130   gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
SHap106   gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
SHap72    gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
SHap110   gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Merged    gcagtgatag cagcaacagc agtgatagca gtgaaagcag taatagtagt gacaacagca
Merged    S  S  D  S   S  N  S   S  D  S   S  E  S  S   N  S  S   D  N  S    933
```

```
RefSeq    atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta 2860
Hap3      atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap4      atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap5      atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap6      atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap8      atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap13     atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap14     atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap20     atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap22     atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap24     atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap25     atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap26     atagcagtga cagcagcaac agcagtgaca gcagt----- ----gacagc agtaatagta
Hap28     atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap29     atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap31     atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap32     atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap33     atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap34     atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap35     atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap36     atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Hap37     atagcagtga cagcagcaac agcagtgaca gcagt----- ----gacagc agtaatagta
Hap38     atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
SHap1:    atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
SHap2:    atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
SHap3:    atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
SHap4:    atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
SHap5:    atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
SHap6:    atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
SHap6(2)  atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
SHap7     atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
SHap102   atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
SHap130   atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
SHap106   atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
SHap72    atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
SHap110   atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Merged    atagcagtga cagcagcaac agcagtgaca gcagtgatag cagtgacagc agtaatagta
Merged    N  S  S  D   S  S  N   S  S  D   S  S  D  S   S  D  S   S  N  S    953
```

```
RefSeq    gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca 2920
Hap3      gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap4      gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap5      gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap6      gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap8      gtgacagcag caatagcggt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap13     gtgacagcag caatagcggt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap14     gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap20     gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap22     gtgacagcag caatagcggt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap24     gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap25     gtgacagcag caatagcggt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap26     gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap28     gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap29     gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap31     gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap32     gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap33     gtgacagcag caatagcggt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap34     gtgacagcag caatagcggt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap35     gtgacagcag caatagcggt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap36     gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap37     gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
Hap38     gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
SHap1:    gtgacagcag caatagcggt gacagcagca acagcagtga cagcagtgat agcaatagca
SHap2:    gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
SHap3:    gtgacagcag caatagcggt gacagcagca acagcagtga cagcagtgat agcaatagca
SHap4:    gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
SHap5:    gtgacagcag caatagcggt gacagcagca acagcagtga cagcagtgat agcaatagca
SHap6:    gtgacagcag caatagcggt gacagcagca acagcagtga cagcagtgat agcaatagca
SHap6(2)  gtgacagcag caatagcggt gacagcagca acagcagtga cagcagtgat agcaatagca
SHap7     gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
SHap102   gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
SHap130   gtgacagcag caatagcggt gacagcagca acagcagtga cagcagtgat agcaatagca
SHap106   gtgacagcag caatagcggt gacagcagca acagcagtga cagcagtgat agcaatagca
SHap72    gtgacagcag caatagcggt gacagcagca acagcagtga cagcagtgat agcaatagca
SHap110   gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
Merged    gtgacagcag caatagcagt gacagcagca acagcagtga cagcagtgat agcaatagca
Merged    S  D  S  S   N  S S/G  D  S  S   N  S  S  D   S  S  D   S  N  S   973
```

Notes: Indel 4 (9 Nt) is in 2 unique McKnight haplotypes: 26 & 37.
Snp3: c.2878A>G; p.S960G

```
RefSeq    gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca 2980
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Hap4      gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Hap5      gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Hap6      gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Hap8      gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Hap13     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Hap14     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Hap20     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Hap22     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Hap24     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Hap25     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagcaat agcagtgaca
Hap26     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagt--- ----------
Hap28     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Hap29     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagt--- ----------
Hap31     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Hap32     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Hap33     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Hap34     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Hap35     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Hap36     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Hap37     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagt--- ----------
Hap38     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
SHap1:    gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
SHap2:    gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
SHap3:    gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
SHap4:    gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
SHap5:    gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
SHap6:    gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
SHap6(2)  gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
SHap7     gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
SHap102   gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
SHap130   gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
SHap106   gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
SHap72    gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
SHap110   gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Merged    gcgacagcag tgacagcagc aacagcagcg atagcagtga cagcagtgat agcagtgaca
Merged    S  D  S   S  D  S  S   N  S  S   D  S  S   D  S  S  D   S  S  D   993
```

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RefSeq    gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg 3040
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Hap4      gcagtgacag cagtgatagc agcgacagca gtgatagcag tgacagcagt gacagcagtg
Hap5      gcagtgacag cagtgatagc agcgacagca gtgatagcag tgacagcagt gacagcagtg
Hap6      gcagtgacag cagtgatagc agcgacagca gtgatagcag tgacagcagt gacagcagtg
Hap8      gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
Hap13     gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
Hap14     gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
Hap20     gcagtgacag cagtgatagc agcgacagca gtgatagcag tgacagcagt gacagcagtg
Hap22     gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
Hap24     gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
Hap25     gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
Hap26     -----gacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
Hap28     gcagtgacag cagtgatagc agcgacagca gtgatagcag tgacagcagt gacagcagtg
Hap29     -----gacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
Hap31     gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
Hap32     gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
Hap33     gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
Hap34     gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
Hap35     gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
Hap36     gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
Hap37     -----gacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
Hap38     gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
SHap1:    gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
SHap2:    gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
SHap3:    gcagtgacag cagtgatagc agcgacagca gtgatagcag tgacagcagt gacagcagtg
SHap4:    gcagtgacag cagtgatagc agcgacagca gtgatagcag tgacagcagt gacagcagtg
SHap5:    gcagtgacag cagtgatagc agcgacagca gtgatagcag tgacagcagt gacagcagtg
SHap6:    gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
SHap6(2)  gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
SHap7     gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
SHap102   gcagtgacag cagtgatagc agcgacagca gtgatagcag tgacagcagt gacagcagtg
SHap130   gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
SHap106   gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
SHap72    gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
SHap110   gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
Merged    gcagtgacag cagtgatagc agcaacagca gtgatagcag tgacagcagt gacagcagtg
Merged    S  S  D  S   S  D  S   S  N  S   S  D  S  S   D  S  S   D  S  S   1013
```

Notes: Indel 5 (18 Nt) is in 3 unique McKnight haplotypes: 26, 29, & 37.
Snp3004A>G (not listed in Song et al.)

```
                                                                 3110
RefSeq    atagcagtaa tagtagtgacagcagcaatagcagtgacag cagcaacagcagtgacagcagcgatagcag
Hap3      atagcagtaa tagtagtgacagcagcaatagcagtgacag cagcaacagcagtgacagcagcgatagcag
Hap4      atagcagtaa tagt------------------agtgacag cagcaacagcagtgacagcagcgatagcag
Hap5      atagcagtaa tagt------------------agtgacag cagcaacagcagtgacagcagcgatagcag
Hap6      atagcagtaa tagt------------------agtgacag cagcaacagcagtgacagcagcgatagcag
Hap8      atagcagtaa tagt------------------agtgacag cagcaacagcagtgacagcagcgatagcag
Hap13     atagcagtaa tagt------------------agtgacag cagcaacagcagtgacagcagcgatagcag
Hap14     atagcagtaa tagtagtgacagcagcaatagcagtgacag cagcaacagcagtgacagcagcgatagcag
Hap20     atagcagtaa tagt------------------agtgacag cagcaacagcagtgacagcagcgatagcag
Hap22     atagcagtaa tagt------------------agtgacag cagcaacagcagtgacagcagcgatagcag
Hap24     atagcagtaa tagtagtgacagcagcaatagcagtgacag cagcaacagcagtgacagcagcgatagcag
Hap25     atagcagtaa tagtagtgacagcagcaatagcagtgacag cagcagcagcagtgacagcagcgatagcag
Hap26     atagcagtaa tagt------------------agtgacag cagcaac------------------agcag
Hap28     atagcagtaa tagt------------------agtgacag cagcaacagcagtgacagcagcgatagcag
Hap29     atagcagtaa tagt------------------agtgacag cagcaac------------------agcag
Hap31     atagcagtaa tagt------------------agtgacag cagcaacagcagtgacagcagcgatagcag
Hap32     atagcagtaa tagtagtgacagcagcaatagcagtgacag cagcaac------------------agcag
Hap33     atagcagtaa tagt------------------agtgacag cagcaac------------------agcag
Hap34     atagcagtaa tagt------------------agtgacag cagcaacagcagtgacagcagcgatagcag
Hap35     atagcagtaa tagt------------------agtgacag cagcaac------------------agcag
Hap36     atagcagtaa tagtagtgacagcagcaacagcagtgacag cagcaacagcagtgacagcagcgatagcag
Hap37     atagcagtaa tagt------------------agtgacag cagcaac------------------agcag
Hap38     atagcagtaa tagtagtgacagcagcaatagcagtgacag cagcaac------------------agcag
SHap1:    atagcagtaa tagt------------------agtgacag cagcaacagcagtgacagcagcgatagcag
SHap2:    atagcagtaa tagtagtgacagcagcaatagcagtgacag cagcaacagcagtgacagcagcgatagcag
SHap3:    atagcagtaa tagt------------------agtgacag cagcaacagcagtgacagcagcgatagcag
SHap4:    atagcagtaa tagt------------------agtgacag cagcaacagcagtgacagcagcgatagcag
SHap5:    atagcagtaa tagt------------------agtgacag cagcaac------------------agcag
SHap6:    atagcagtaa tagt------------------agtgacag cagcaac------------------agcag
SHap6(2)  atagcagtaa tagt------------------agtgacag cagcaac------------------agcag
SHap7     atagcagtaa tagt------------------agtgacag cagcaac------------------agcag
SHap102   atagcagtaa tagt------------------agtgacag cagcaacagcagtgacagcagcgatagcag
SHap130   atagcagtaa tagt------------------agtgacag cagcaac------------------agcag
SHap106   atagcagtaa tagt------------------agtgacag cagcaac------------------agcag
SHap72    atagcagtaa tagt------------------agtgacag cagcaacagcagtgacagcagcgatagcag
SHap110   atagcagtaa tagt------------------agtgacag cagcaacagcagtgacagcagcgatagcag
Merged    atagcagtaa tagtagtgacagcagcaatagcagtgacag cagcaacagcagtgacagcagcgatagcag
Merged    D  S  S  N   S  S  D  S  S  N  S  S  D  S   S  N  S  S  D  S  S  D  S  S
```

Notes: Indel 6 (18 Nt) is in 15 unique McKnight haplotypes. 1037

Id4: c.3055_3072delAGTGACAGCAGCAATAGC; p.S1024delSDSSNS

Id4 (Song) is the same as Indel 6 (McKnight).

```
RefSeq    tgacagcagc gatagcagtg acagcagcga tagcagtgac ag**c**agtgaca gcagcaat 3168
Hap3      tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap4      tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap5      tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap6      tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap8      tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap13     tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap14     tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap20     tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap22     tgacagcagc gatagcagtg acagcagcg**g** tagcagtgac agcagtgaca gcagcaat
Hap24     tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap25     tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap26     tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap28     tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap29     tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap31     tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap32     tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap33     tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap34     tgacagcag**t** gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap35     tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap36     tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap37     tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Hap38     tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
SHap1:    tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
SHap2:    tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
SHap3:    tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
SHap4:    tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
SHap5:    tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
SHap6:    tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
SHap6(2)  tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
SHap7     tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
SHap102   tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
SHap130   tgacagcagc gatagcagtg acagcagcga t--------- agcagtgaca gcagcaat
SHap106   tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
SHap72    tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
SHap110   tgacagcagc gatagcagtg acagcagcga tagcagtgac agcagtgaca gcagcaat
Merged    tgacagcagc gatagcagtg acagcagcga t**agcagtgac** agcagtgaca gcagcaat
Merged      D  S  S   D  S  S   D  S  S  D   S  S  D   S  S  D   S  S  N  1056
```

Notes: Indel 7 (18 Nt) is in 7 unique McKnight haplotypes 26; 29; 32; 33; 1/7/16/35; and 37/38
Id5: c.3088_3105delAGCAGTGACAGCAGCGAT; p.S1030_D1035DELSSDSSD
Id5 (Song) is the same as Indel 7 (McKnight).
Id6: c.3142_3150delAGCAGTGAC; p.1048_1050delSSD
Id7: c.3168_3169insAGCAGTGACAGCAGTGACAGCAGCGAC; p.N056_S1057insSSDSSDSSD
c.3141delC or p.S1047fsX223; c.3141delC, dentin dysplasia phenotype (3); possibly this mutation was described improperly as this nucleotide is not a C. It should be c.3153delC and p.S1051fsX223.

```
RefSeq    ----------------------------ag cagtgacagc agtgacagca gcgacagcag 3200
Hap3      ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap4      ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap5      ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap6      ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap8      ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap13     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap14     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap20     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap22     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap24     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap25     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap26     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap28     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap29     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap31     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap32     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap33     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap34     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap35     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap36     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap37     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Hap38     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
SHap1:    ----------------------------ag cagtgacagc agtgacagca gcgacagcag
SHap2:    ----------------------------ag cagtgacagc agtgacagca gcgacagcag
SHap3:    ----------------------------ag cagtgacagc agtgacagca gcgacagcag
SHap4:    ----------------------------ag cagtgacagc agtgacagca gcgacagcag
SHap5:    ----------------------------ag cagtgacagc agtgacagca gcgacagcag
SHap6:    ----------------------------ag cagtgacagc agtgacagca gcgacagcag
SHap6(2)  ----------------------------ag cagtgacagc agtgacagca gcgacagcag
SHap7     ----------------------------ag cagtgacagc agtgacagca gcgacagcag
SHap102   ----------------------------ag cagtgacagc agtgacagca gcgacagcag
SHap130   ----------------------------ag cagtgacagc agtgacagca gcgacagcag
SHap106   agcagtgacagcagtgacagcagcgacag cagtgacagc agtgacagca gcgacagcag
SHap72    ----------------------------ag cagtgacagc agtgacagca gcgacagcag
SHap110   ----------------------------ag cagtgacagc agtgacagca gcgacagcag
Merged    agcagtgacagcagtgacagcagcgacag cagtgacagc agtgacagca gcgacagcag
Merged     S  S  D  S  S  D  S  S  D  S   S  D  S   S  D  S   S  D  S  S 1067
```

Notes: Id7: c.3168_3169insAGCAGTGACAGCAGTGACAGCAGCGAC; p.N056_S1057insSSDSSDSSD
Snp4: c.3221A>G; p.D1074G

```
RefSeq    tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag 3260
Hap3      tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
Hap4      tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
Hap5      tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
Hap6      tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
Hap8      tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaca gcagtgatag
Hap13     tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaca gcagtgatag
Hap14     tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
Hap20     tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
Hap22     tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaca gcagtgatag
Hap24     tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
Hap25     tgacagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
Hap26     tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaca gcagtgatag
Hap28     tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
Hap29     tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaca gcagtgatag
Hap31     tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
Hap32     tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
Hap33     tgatagcagt gacagcagtg gcagcagcga cagcagtgat agcagtgaca gcagtgatag
Hap34     tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaca gcagtgatag
Hap35     tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaca gcagtgatag
Hap36     tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
Hap37     tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaca gcagtgatag
Hap38     tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
SHap1:    tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaca gcagtgatag
SHap2:    tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
SHap3:    tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaca gcagtgatag
SHap4:    tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
SHap5:    tgatagcagt gacagcagtg gcagcagcga cagcagtgat agcagtgaca gcagtgatag
SHap6:    tgatagcagt gacagcagtg gcagcagcga cagcagtgat agcagtgaca gcagtgatag
SHap6(2)  tgatagcagt gacagcagtg gcagcagcga cagcagtgat agcagtgaca gcagtgatag
SHap7     tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
SHap102   tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
SHap130   tgatagcagt gacagcagtg gcagcagcga cagcagtgat agcagtgaca gcagtgatag
SHap106   tgatagcagt gacagcagtg gcagcagcga cagcagtgat agcagtgaca gcagtgatag
SHap72    tgacagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaca gcagtgatag
SHap110   tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
Merged    tgatagcagt gacagcagtg acagcagcga cagcagtgat agcagtgaaa gcagtgatag
Merged      D  S  S   D  S  S  D/G S  S  D   S  S  D  S  S  E/D  S  S  D  S 1087
```

Notes: Snp5: c.3249A>C; p.E1083D
Snp6: c.3264T>C; p.S1088

```
RefSeq    cagtg[---------]acagc agcaatagca gtgacagcag cgatagcagc gacagcagcg 3310
Hap3      cagtg[---------]acagc agcaatagca gtgacagcag cgatagcagc gacagcagcg
Hap4      cagcg[---------]----- ---------- ---------- ---------- ----------
Hap5      cagcg[---------]----- ---------- ---------- ---------- ----------
Hap6      cagcg[---------]----- ---------- ---------- ---------- ----------
Hap8      cagcg[---------]----- ---------- ---------- ---------- ----------
Hap13     cagcg[---------]----- ---------- ---------- ---------- ----------
Hap14     cagtg[---------]acagc agcaatagca gtgacagcag cgatagcagc gacagcagcg
Hap20     cagcg[---------]----- ---------- ---------- ---------- ----------
Hap22     cagcg[---------]----- ---------- ---------- ---------- ----------
Hap24     cagtg[In 189 nt]acagc agcaatagca gtgacagcag cgatagcagc gacagcagcg
Hap25     cagtg[---------]acagc agcaatagca gtgacagcag cgatagcagc gacagcagcg
Hap26     cagcg[---------]----- ---------- ---------- ---------- ----------
Hap28     cagcg[---------]----- ---------- ---------- ---------- ----------
Hap29     cagcg[---------]----- ---------- ---------- ---------- ----------
Hap31     cagcg[---------]----- ---------- ---------- ---------- ----------
Hap32     cagcg[---------]----- ---------- ---------- ---------- ----------
Hap33     cagcg[---------]----- ---------- ---------- ---------- ----------
Hap34     cagcg[---------]----- ---------- ---------- ---------- ----------
Hap35     cagcg[---------]----- ---------- ---------- ---------- ----------
Hap36     cagcg[---------]----- ---------- ---------- ---------- ----------
Hap37     cagcg[---------]----- ---------- ---------- ---------- ----------
Hap38     cagcg[---------]----- ---------- ---------- ---------- ----------
SHap1:    cagcg[---------]----- ---------- ---------- ---------- ----------
SHap2:    cagtg[---------]acagc agcaatagca gtgacagcag cgatagcagc gacagcagcg
SHap3:    cagcg[---------]----- ---------- ---------- ---------- ----------
SHap4:    cagcg[---------]----- ---------- ---------- ---------- ----------
SHap5:    cagcg[---------]----- ---------- ---------- ---------- ----------
SHap6:    cagcg[---------]----- ---------- ---------- ---------- ----------
SHap6(2)  cagcg[---------]----- ---------- ---------- ---------- ----------
SHap7     cagcg[---------]----- ---------- ---------- ---------- ----------
SHap102   cagcg[---------]----- ---------- ---------- ---------- ----------
SHap130   cagcg[---------]----- ---------- ---------- ---------- ----------
SHap106   cagcg[---------]----- ---------- ---------- ---------- ----------
SHap72    cagcg[---------]----- ---------- ---------- ---------- ----------
SHap110   cagcg[---------]----- ---------- ---------- ---------- ----------
Merged    cagtg[In 189 nt]acagc agcaatagca gtgacagcag cgatagcagc gacagcagcg
Merged      S  [  63 aa  ]D  S   S  N  S   S  D  S  S   D  S  S   D  S  S   1103
```

Notes: Indel 8 (189 Nt): The insertion is only in Hap24. The sequence is shown below.
Indel 9 (54 Nt). The insertion is only in 4 unique haplotypes: 3; 14/RefSeq; 24; & 25.
Id8: c.3266_3400del135bp; p.D1089_S1133delDSSNSS(DSS)$_9$NSS(DSS)$_3$

```
Hap24 [189 nt insert]:
Hap24   acagcagcaatagcagtgacagcagtgacagcagcgacagcagtgatagcagtgacagcagcg
        D  S  S  N  S  S  D  S  S  D  S  S  D  S  S  D  S  S  D  S  S
Hap24   atagcagtgacagcagtgacagcagcaatagcagtgacagcagtgacagcagcgacagcagtg
        D  S  S  D  S  S  D  S  S  N  S  S  D  S  S  D  S  S  D  S  S
Hap24   atagcagtgacagcagtgacagcagcgacagcagtgatagcagtgaaagcagtgatagcagtg
        D  S  S  D  S  S  D  S  S  D  S  S  D  S  S  E  S  S  D  S  S

Merged  acagcagcaatagcagtgacagcagtgacagcagcgacagcagtgatagcagtgacagcagcg
Merged  D  S  S  N  S  S  D  S  S  D  S  S  D  S  S  D  S  S  D  S  S
Merged  atagcagtgacagcagtgacagcagcaatagcagtgacagcagtgacagcagcgacagcagtg
Merged  D  S  S  D  S  S  D  S  S  N  S  S  D  S  S  D  S  S  D  S  S
Merged  atagcagtgacagcagtgacagcagcgacagcagtgatagcagtgaaagcagtgatagcagtg
Merged  D  S  S  D  S  S  D  S  S  D  S  S  D  S  S  E  S  S  D  S  S

RefSeq    acagcagcga tagcagtgac agcagcgata gcagtgacag cagtgacagc agcaatagca 3370
Hap3      acagcagcga tagcagtgac agcagcgata gcagtgacag cagtgacagc agcaatagca
Hap4      ---------- ---------- ---------- ---------- ---------- ----------
Hap5      ---------- ---------- ---------- ---------- ---------- ----------
Hap6      ---------- ---------- ---------- ---------- ---------- ----------
Hap8      ---------- ---------- ---------- ---------- ---------- ----------
Hap13     ---------- ---------- ---------- ---------- ---------- ----------
Hap14     acagcagcga tagcagtgac agcagcgata gcagtgacag cagtgacagc agcaatagca
Hap20     ---------- ---------- ---------- ---------- ---------- ----------
Hap22     ---------- ---------- ---------- ---------- ---------- ----------
Hap24     acagcagcga tagcagtgac agcagcgata gcagtgacag cagtgacagc agcaatagca
Hap25     acagcagcga tagcagtgac agcagcgata gcagtgacag cagtgacagc agcaatagca
Hap26     ---------- ---------- ---------- ---------- ---------- ----------
Hap28     ---------- ---------- ---------- ---------- ---------- ----------
Hap29     ---------- ---------- ---------- ---------- ---------- ----------
Hap31     ---------- ---------- ---------- ---------- ---------- ----------
Hap32     ---------- ---------- ---------- ---------- ---------- ----------
Hap33     ---------- ---------- ---------- ---------- ---------- ----------
Hap34     ---------- ---------- ---------- ---------- ---------- ----------
Hap35     ---------- ---------- ---------- ---------- ---------- ----------
Hap36     ---------- ---------- ---------- ---------- ---------- ----------
Hap37     ---------- ---------- ---------- ---------- ---------- ----------
Hap38     ---------- ---------- ---------- ---------- ---------- ----------
SHap1:    ---------- ---------- ---------- ---------- ---------- ----------
SHap2:    acagcagcga tagcagtgac agcagcgata gcagtgacag cagtgacagc agcaatagca
SHap3:    ---------- ---------- ---------- ---------- ---------- ----------
SHap4:    ---------- ---------- ---------- ---------- ---------- ----------
SHap5:    ---------- ---------- ---------- ---------- ---------- ----------
SHap6:    ---------- ---------- ---------- ---------- ---------- ----------
SHap6(2)  ---------- ---------- ---------- ---------- ---------- ----------
SHap7     ---------- ---------- ---------- ---------- ---------- ----------
SHap102   ---------- ---------- ---------- ---------- ---------- ----------
SHap130   ---------- ---------- ---------- ---------- ---------- ----------
SHap106   ---------- ---------- ---------- ---------- ---------- ----------
SHap72    ---------- ---------- ---------- ---------- ---------- ----------
SHap110   ---------- ---------- ---------- ---------- ---------- ----------
Merged    acagcagcga tagcagtgac agcagcgata gcagtgacag cagtgacagc agcaatagca
Merged    D  S  S  D  S  S  D  S  S  D  S  S  D  S  S  D  S  S  N  S  1123
```

Note: Indel 10 (81 Nt)

```
RefSeq    gtgacagcag tgacagcagc gacagcagtg atagcagtga cagcagcaac agcagtgaca 3430
Hap3      gtgacagcag tgacagcagc gacagcagtg atagcagtga cagcagcaac agcagtgaca
Hap4      ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Hap5      ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Hap6      ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Hap8      ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Hap13     ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Hap14     gtgacagcag tgacagcagc gacagcagtg atagcagtga cagcagcaac agcagtgaca
Hap20     ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Hap22     ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Hap24     gtgacagcag tgacagcagc gacagcagtg atagcagtga cagcagcaac agcagtgaca
Hap25     gtgacagcag tgacagcagc gacagcagtg atagcagtga cagcagcaac agcagtgaca
Hap26     ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Hap28     ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Hap29     ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Hap31     ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Hap32     ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Hap33     ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Hap34     ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Hap35     ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Hap36     ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Hap37     ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Hap38     ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
SHap1:    ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
SHap2:    gtgacagcag tgacagcagc gacagcagtg atagcagtga cagcagcaac agcagtgaca
SHap3:    ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
SHap4:    ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
SHap5:    ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
SHap6:    ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
SHap6(2)  ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
SHap7     ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
SHap102   ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
SHap130   ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
SHap106   ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
SHap72    ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
SHap110   ---------- ---------- ---------- atagcagtga cagcagcgac agcagtgaca
Merged    gtgacagcag tgacagcagc gacagcagtg atagcagtga cagcagcaac agcagtgaca
Merged    S  D  S  S  D  S  S  D  S  S  D  S  S  D  S  S  N  S  S  D  1143
```

Note: Snp7 is c.3418A>G; p.N1140D.

```
RefSeq    gcagtga**c**ag cagtgaa------------------agc agcgacagca gtgacagcag 3470
Hap3      gcagtgacag cagtgaa------------------agc agcgacagca gtgacagcag
Hap4      gcagtgacag cagtgaa------------------agc agcgacagca gtgacagcag
Hap5      gcagtgacag cagtgaa------------------agc agcgacagca gtgacagcag
Hap6      gcagtgacag cagtgaa------------------agc agcgacagca gtgacagcag
Hap8      gcagtgacag cagtgaaagcagcgacagcagcgatagc agcgacagca gtgacagcag
Hap13     gcagcgacag ca------------------gcgatagc agcgacagca gtgac-----
Hap14     gcagtgacag cagtgaa------------------agc agcgacagca gtgacagcag
Hap20     gcagtgacag cagtgaa------------------agc agcgacagca gtgacagcag
Hap22     gcagtgacag cagtgaaagcagcgacagcagcgatagc agcgacagca gtgacagcag
Hap24     gcagtgacag cagtgaa------------------agc agcgacagca gtgacagcag
Hap25     gcagtgacag cagtgaa------------------agc agcgacagca gtgacagcag
Hap26     gcagtgacag cagtgaaagcagcgacagcagcgatagc agcgacagca gtgacagcag
Hap28     gcagtgacag cagtgaa------------------agc agcgacagca gtgacagcag
Hap29     gcagtgacag cagtgaaagcagcgacagcagcgatagc agcgacagca gtgacagcag
Hap31     gcagtgacag cagtgaa------------------agc agcgacagca gtgacagcag
Hap32     gcagtgacag cagtgaa------------------agc agcgacagca gtgacagcag
Hap33     gcagtgacag cagtgaaagcagcgacagcagcgatagc agcgacagca gtgacagcag
Hap34     gcagtgacag cagtgaaagcagcgacagcagcgatagc agcgacagca gtgac-----
Hap35     gcagtgacag cagtgaaagcagcgacagcagcgatagc agcgacagca gtgacagcag
Hap36     gcagtgacag cagtgaa------------------agc agcgacagca gtgacagcag
Hap37     gcagtgacag cagtgaaagcagcgacagcagcgatagc agcgacagca gtgacagcag
Hap38     gcagtgacag cagtgaa------------------agc agcgacagca gtgacagcag
SHap1:    gcagtgacag cagtgaaagcagcgacagcagcgatagc agcgacagca gtgacagcag
SHap2:    gcagtgacag cagtgaa------------------agc agcgacagca gtgacagcag
SHap3:    gcagtgacag cagtgaaagcagcgacagcagcgatagc agcgacagca gtgacagcag
SHap4:    gcagtgacag cagtgaaagcagcgacagcagcgatagc agc**a**acagca gtgacagcag
SHap5:    gcagtgacag cagtgaaagcagcgacagcagcgatagc agcgacagca gtgacagcag
SHap6:    gcagtgacag cagtgaaagcagcgacagcagcgatagc agcgacagca gtgacagcag
SHap6(2)  gcagtgacag cagtgaaagcagcgacagcagcgatagc agcgacagca gtgacagcag
SHap7     gcagtgacag cagtgaaagcagcgacagcagcgatagc agc**a**acagca gtgacagcag
SHap102   gcagtgacag cagtgaaagcagcgacagcagcgatagc agcgacagca gtgacagcag
SHap130   gcagtgacag cagtgaaagcagcgacagcagcgatagc agcgacagca gtgacagcag
SHap106   gcagtgacag cagtgaaagcagcgacagcagcgatagc agcgacagca gtgacagcag
SHap72    gcagtgacag cagtgaa------------------agc agcgacagca gtgac-----
SHap110   gcagtgacag cagtgaaagcagcgacagcagcgatagc agc**a**acagca gtgacagcag
Merged    gcagtgacag cagtgaa**agcagcgacagcagcgat**agc agcgacagca gtgac**agcag**
Merged    S  S  D  S   S  E  **S  S  D  S  S  D**  S   S D/**N** S   S  D  S  S 1157
```

Notes: Dentinogenesis imperfecta type II mutation c.3438delC; p.D1146fsX1313 (Song).
Indel 10 (18 Nt) and is in a slightly different place in Hap13.
Id9: c.3447_3448insAGCAGCGACAGCAGCGAT; p. E1149_S1150insSSDSSD.
Id9 (Song) is the same as Indel 10 (McKnight).
Snp8: c.3454G>A; p.D1152N

```
RefSeq    cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag 3530
Hap3      cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap4      cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap5      cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap6      cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap8      cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap13     -------------------- --agcagcga cagcagcgac agcagcgata gcagtgacag
Hap14     cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap20     cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap22     cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap24     cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap25     cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap26     cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap28     cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap29     cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap31     cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap32     cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap33     cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap34     -------------------- --agcagcga cagcagcgac agcagcgata gcagtgacag
Hap35     cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap36     cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap37     cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Hap38     cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
SHap1:    cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
SHap2:    cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
SHap3:    cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
SHap4:    cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
SHap5:    cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
SHap6:    cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
SHap6(2)  cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
SHap7     cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
SHap102   cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
SHap130   cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
SHap106   cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
SHap72    -------------------- --agcagcga cagcagcgac agcagcgata gcagtgacag
SHap110   cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Merged    cgacagcagtgacagcagcg atagcagcga cagcagcgac agcagcgata gcagtgacag
Merged      D  S  S  D  S  S   D  S  S  D   S  S  D   S  S  D   S  S  D  S 1177
```

Notes: Indel 11 (27 Nt) is only in 2 unique McKnight haplotypes: 13 and 10/12/15/17/18/34.
Id10: c.3466_3492delAGCAGCGACAGCAGTGACAGCAGCGAT
Id10 (Song) is the same as Indel 11 (McKnight).

```
RefSeq    cagcaatagc agtga**tagca** gcgacagca**g** tgatagcagt gacagcagcg a**cagcagcga** 3590
Hap3      cagcaatagc agtgatagca gcgacagcag tgatagcag**c** gacagcagtg acagcagcga
Hap4      cagcaatagc agtgatagca gcgacagcag tga**c**------ ---agcagtg acagcagcga
Hap5      cagcaatagc agtgatagca gcgacagcag tga**c**------ ---agcagtg acagcagcga
Hap6      cagcaatagc agtgatagca gcgacagcag tga**c**------ ---agcagtg acagcagcga
Hap8      cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
Hap13     cagcaatagc agtgatagca gcgacagcag tgatagcag**c** gacagcagcg acagcagcga
Hap14     cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
Hap20     cagcaatagc agtgatagca gcgacagcag tga**c**agcagt gacagcagcg acagcagcga
Hap22     cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
Hap24     cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
Hap25     cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
Hap26     cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
Hap28     cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
Hap29     cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
Hap31     cagcaatagc agtgatagca gcgacagcag tga**c**agcagt gacagcagcg acagcagcga
Hap32     cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
Hap33     cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
Hap34     cagcaatagc agtgatagca gcgacagcag tgatagcag**c** gacagcagcg acagcagcga
Hap35     cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
Hap36     cagc**g**a**c**agc agtgatagca gcgacagcag tga**c**agcagt gacagcagcg acagcagcga
Hap37     cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
Hap38     cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
SHap1:    cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
SHap2:    cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
SHap3:    cagcaatagc agtgatagca gcgacagcag tgatagcag**c** gacagcagcg acagcagcga
SHap4:    cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
SHap5:    cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
SHap6:    cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
SHap6(2)  cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
SHap7     cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
SHap102   cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
SHap130   cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
SHap106   cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
SHap72    cagcaatagc agtgatagca gcgacagcag tgat------ ---agcagtg acagcagcga
SHap110   cagcaatagc agtgatagca gcgacagcag tgatagcagt gacagcagcg acagcagcga
Dong:                                                 agcagcga tagcagcgac
Merged    cagcaatagc agtgatagca gcgacagcag tgat**agcagt** **gac**agcagcg acagcagcga
Merged      S  N  S   S  D  S   S  D  S  S   D  S  S   D  S  S   D  S  S  D 1197
```

Notes: Dentinogenesis imperfecta type II mutation c.3546delTAGCAinsG; p.D1182fsX1312 (Song).
Indel 12 (9 Nt) is in haplotypes 4, 5, and 8
Id11: c.3565_3573delAGCAGTGAC; p.S1189_D1191delSSD
Id11 (Song) is the same as Indel 12 (McKnight).
Snp9: c.3570T>C; p.S1190
Dentinogenesis imperfecta type II mutation c.3560delG; p.S1187fsX1270 (Lee:ClinGenet).
Dentin dysplasia II mutation c.3582-3591delCAGCAGCGAT; p.D1194EfsX1310 (Nieminen, kind. 11).

```
RefSeq    tagcagcgac agcagcgata gtagtgatag ca------------------gtgacagc 3630
Hap3      tagcagcgac agcagcgata gcagcgacag cagcgatagtagtgatagcagtgacagc
Hap4      tagcagcgac agcagtgaca gtagtgatag ca------------------------
Hap5      tagcagcgac agcagtgaca gtagtgatag ca------------------------
Hap6      tagcagcgac agcagtgaca gtagtgatag ca------------------------
Hap8      tagcagcgac agcagtgata gtagtgatag ca------------------------
Hap13     cagcagcgac agcagtgata gtagtgatag ca------------------------
Hap14     tagcagcgac agcagcgata gtagtgatag ca------------------gtgacagc
Hap20     tagcagcgac agcagtgaca gtagtgatag ca------------------------
Hap22     tagcagcgac agcagtgata gtagtgatag ca------------------gtgacagc
Hap24     tagcagcgac agcagcgata gtagtgatag ca------------------gtgacagc
Hap25     tagcagcgac agcagcgata gtagtgatag ca------------------gtgacagc
Hap26     tagcagcgac agcagtgata gtagtgatag ca------------------------
Hap28     tagcagcgac agcagcgata gtagtgatag ca------------------gtgacagc
Hap29     tagcagcgac agcagtgata gtagtgatag ca------------------gtgacagc
Hap31     tagcagcgac agcagtgaca gtagtgatag ca------------------------
Hap32     tagcagcgac agcagtgata gtagtgatag ca------------------------
Hap33     tagcagcgac agcagtgata gtagtgatag ca------------------gtgacagc
Hap34     cagcagcgac agcagtgata gtagtgatag ca------------------gtgacagc
Hap35     tagcagcgac agcagtgata gtagtgatag ca------------------gtgacagc
Hap36     tagcagcgac agcagtgaca gtagtgatag ca------------------------
Hap37     tagcagcgac agcagtgata gtagtgatag ca------------------gtgacagc
Hap38     tagcagcgac agcagtgata gtagtgatag ca------------------------
SHap1:    tagcagcgac agcagtgata gtagtgatag ca------------------gtgacagc
SHap2:    tagcagcgac agcagcgata gtagtgatag ca------------------gtgacagc
SHap3:    cagcagcgac agcagtgata gtagtgatag ca------------------gtgacagc
SHap4:    tagcagcgac agcagcgata gtagtgatag ca------------------gtgacagc
SHap5:    tagcagcgac agcagtgata gtagtgatag ca------------------------
SHap6:    tagcagcgac agcagtgata gtagtgatag ca------------------gtgacagc
SHap6(2)  tagcagcgac agcagtgata gtagtgatag ca------------------gtgacagc
SHap7     tagcagcgac agcagcgata gtagtgatag ca------------------gtgacagc
SHap102   tagcagcgac agcagtgata gtagtgatag ca------------------------
SHap130   tagcagcgac agcagtgata gtagtgatag ca------------------gtgacagc
SHap106   tagcagcgac agcagtgata gtagtgatag ca------------------gtgacagc
SHap72    cagcagcgac agcagtgata gtagtgatag ca------------------gtgacagc
SHap110   tagcagcgac agcagcgata gtagtgatag ca------------------------
Dong:     tagcagcgac agcagtgata gtagtgatag ca------------------------
Merged    tagcagcgac agcagcgata gtagtgatag cagcgatagtagtgatagcagtgacagc
Merged      S  S  D   S  S  D   S  S  D  S   S  D  S  S  D  S  S  D  S  1210
```

Notes: Snp10: c.3591T>C; p.D1197
Snp11: c.3606C>T; p.S1202
Overlapping Indels that start at the same position but vary in length:
--Indel 13 (18 Nt): RefSeq/14; 10/12/15/17/18/34; 19/21/22; 23/28; 24; 25; 29; 33; 1/7/16/35; 37;& 38
--Indel 14 (27 Nt): 13 & 26.
--Indel 15 (36 Nt): 4; 5; 6; 8; ll/20; 30/36; 27/31; & 32.
--Indel 16 (54 Nt): is only in haplotype 38; This is a deletion of 36 Nt relative to the wild type and is part of the polymorphism in the DGI-III family (6).
Id12: c.3623_3640delGTGACAGCAGTGACAGCA; p.S1208_S1219delSDSSDS
Id12 (Song) is the same as Indel 15 (McKnight).
Id13: c3641_3658delGCGACAGCAGTGACAGCA; p.SDSSDS
Id12 + Id13 (Song) together are the same as Indel 16 (McKnight).
Dentin dysplasia II mutation c.3625-3700del 76 bp; p.D1209AfsX1288 (Nieminen, kindred 12).

```
RefSeq  agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac 3690
Hap3    agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap4    ---------- gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap5    ---------- gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap6    ---------- gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap8    ---------- gtgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap13   -gtgacagca gtgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtggc
Hap14   agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap20   ---------- gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap22   agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap24   agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap25   agtgacagca gcgacagcag tgacagcagc aacagcagtg acagcagcga cagcagtgac
Hap26   -gtgacagca gtgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap28   agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap29   agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap31   ---------- gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap32   ---------- gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap33   agtgacagca gcgacagcag tgacagcagc ---------- ---------- ----------
Hap34   agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap35   agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap36   ---------- gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap37   agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Hap38   ---------- ---------- --------gc gacagcagtg acagcagcga cagcagtgac
SHap1:  agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
SHap2:  agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
SHap3:  agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
SHap4:  agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
SHap5:  ---------- gtgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
SHap6:  agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
SHap6(2)agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
SHap7   agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
SHap102 ---------- gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
SHap130 agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
SHap106 agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
SHap72  agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
SHap110 ---------- gtgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Dong    ---------- ---------- --------gc gacagcagtg acagcagcga cagcagtgac
Merged  agtgacagca gcgacagcag tgacagcagc gacagcagtg acagcagcga cagcagtgac
Merged   S  D  S   S  D  S  S   D  S  S   D  S  S   D  S  S  D   S  S  D  1230
```

Notes: Dentin dysplasia II mutation c.3625-3700del76 bp; p.D1209AfsX1288 (Nieminen, kindred 12).
Id13: c3641_3658delGCGACAGCAGTGACAGCA; p.SDSSDS
Indel 17 (117 Nt) is only in McKnight haplotype 33.

```
                                                                                                                3740
RefSeq    agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
Hap3      agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
Hap4      agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagcagcgatagcagtgacagcag
Hap5      agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagcagcgatagcagtgacagcag
Hap6      agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagcagcgatagcagtgacagcag
Hap8      agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
Hap13     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
Hap14     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
Hap20     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagcagcgatagcagtgacagcag
Hap22     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
Hap24     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
Hap25     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
Hap26     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
Hap28     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
Hap29     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
Hap31     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagcagcgatagcagtgacagcag
Hap32     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagcagcgatagcagtgacagcag
Hap33     ---------- ---------- ---------- ---------- --------------------------
Hap34     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
Hap35     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
Hap36     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagcagcgatagcagtgacagcag
Hap37     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
Hap38     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagcagcgatagcagtgacagcag
SHap1:    agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
SHap2:    agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
SHap3:    agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
SHap4:    agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
SHap5:    agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
SHap6:    agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
SHap6(2)  agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
SHap7     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
SHap102   agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagcagcgatagcagtgacagcag
SHap130   agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
SHap106   agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
SHap72    agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
SHap110   agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagca-----------------g
Dong:     agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagcagcgatagcagtgacagcag
Merged    agcaatgaaa gcagcgacag cagtgacagc agcgatagca gtgacagcagcgatagcagtgacagcag
Merged     S  N  E   S  S  D  S   S  D  S   S  D  S   S  D  S  S  D  S  S  D  S  S
```

Notes: Indel 18 (18 Nt) corresponds to the insertion in DGI-III family (Dong et al.) 1247
Id14: c.3739_3740insGCGATAGCAGTGACAGCA; p.1246_S1247insSDSSDS
Id14 (Song) is the same as Indel 18 (McKnight).

```
RefSeq    caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga 3800
Hap3      caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap4      caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap5      caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap6      caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap8      caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap13     caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap14     caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap20     caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap22     caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap24     caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap25     caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap26     caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap28     caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap29     caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap31     caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap32     caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap33     ---------- ---------g acagcagtga tagcagtgac agcacatctg acagcaatga
Hap34     caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap35     caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap36     caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap37     caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Hap38     caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
SHap1:    caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
SHap2:    caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
SHap3:    caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
SHap4:    caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
SHap5:    caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
SHap6:    caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
SHap6(2)  caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
SHap7     caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
SHap102   caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
SHap130   caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
SHap106   caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
SHap72    caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
SHap110   caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Merged    caacagcagt gacagcagcg acagcagtga tagcagtgac agcacatctg acagcaatga
Merged      N  S  S   D  S  S   D  S  S  D   S  S  D   S  T  S   D  S  N  D  1267
```

```
RefSeq    tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag 3860
Hap3      tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap4      tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap5      tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap6      tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap8      tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap13     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap14     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap20     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap22     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap24     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap25     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap26     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap28     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap29     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap31     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap32     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap33     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap34     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap35     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap36     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap37     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Hap38     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
SHap1:    tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
SHap2:    tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
SHap3:    tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
SHap4:    tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
SHap5:    tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
SHap6:    tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
SHap6(2)  tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
SHap7     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
SHap102   tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
SHap130   tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
SHap106   tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
SHap72    tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
SHap110   tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Dong:     tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Merged    tgagagtgac agccagagca agtctggtaa cggtaacaac aatggaagtg acagtgacag
Merged     E  S  D   S  Q  S  K   S  G  N   G  N  N   N  G  S  D   S  D  S   1287
```

```
RefSeq    tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag 3906
Hap3      tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
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Hap13     tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
Hap14     tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
Hap20     tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
Hap22     tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
Hap24     tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
Hap25     tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
Hap26     tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
Hap28     tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
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SHap1:    tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
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SHap3:    tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
SHap4:    tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
SHap5:    tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
SHap6:    tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
SHap6(2)  tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
SHap7     tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
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SHap110   tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
Dong:     tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
Merged    tgacagtgaa ggcagtgaca gtaaccactc aaccagtgat gattag
Merged      D  S  E   G  S  D   S  N  H  S  T  S  D   D   *  1301
```

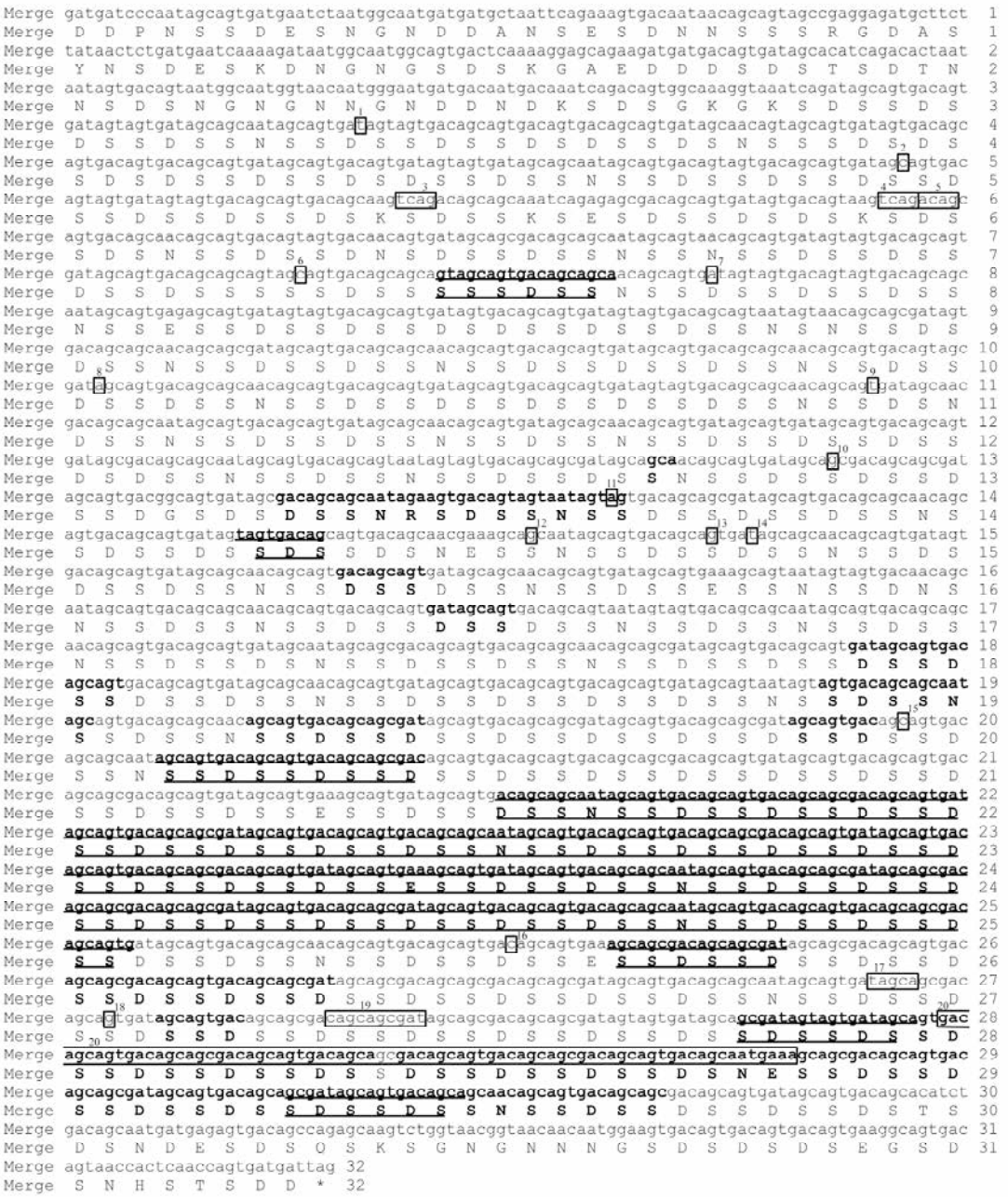

Figure S2. The merged DPP sequence and *DSPP* 5-prime disease-causing mutations. The merged human DPP sequence containing all of the indel sequences is shown with 90 nucleotides or 20 amino acids per row. The merge protein contains 938 amino acids. All indels are in bold. Segments missing from the RefSeq are underlined. Mutations are boxed and numbered according to the list in Fig. 3.

A) Translation of Human DPP reference sequence in the proper reading frame.

```
DDPNSSDESNGNDDANSESDNNSSSRGDASYNSDESKDNGNGSDSKGAEDDDSDSTSDTNNSDSNGNGNNG
NDDNDKSDSGKGKSDSSDSDSSDSSNSSDSSDSSDSDSSDSNSSSDSDSSDSDSSDSSDSDSSDSSNSSDS
SDSSDSSDSSDSSDSSDSKSDSSKSESDSSDSDSKSDSSDSNSSDSSDNSDSSDSSNSSNSSDSSDSSDSS
DSSSSSDSSNSSDSSDSSDSSNSSESSDSSDSSDSSDSSDSSNSNSSDSDSSNSSDSSDSSNSSDSSDS
SDSSNSSDSSDSSDSSNSSDSSDSSDSSDSSDSSNSSDSNDSSNSSDSSDSSNSSDSSNSSDSSDSSDSSD
SDSSNSSDSSNSSDSSDSSNSSDSSDSSDSSDGSDSDSSNRSDSSNSSDSSDSSDSSNSSDSSDSSDSNES
SNSSDSSDSSNSSDSDSSDSSNSSDSSDSSNSSDSSESSNSSDNSNSSDSSNSSDSSDSSDSSNSSDSSNS
SDSSNSSDSSDSNSSDSSDSSNSSDSSDSSDSSDSSDSSDSSNSSDSSDSSDSSDSSNSSDSSNSSDSSNS
SDSSDSSDSSDSSDSSDSSDSSDSSNSSDSSDSSDSSDSSDSSDSSDSSDSSESSDSSDSSNSSDSSDSSD
SSDSSDSSDSSDSSDSSDSSNSSDSSDSSDSSDSSDSSNSSDSSDSSESSDSSDSSDSSDSSDSSDSSDSS
DSSDSSNSSDSSDSSDSSDSSDSSDSSDSSDSSDSSDSSDSSDSSDSSDSSDSSDSSDSNESSDSSDSSDS
SDSSNSSDSSDSSDSSDSTSDSNDESDSQSKSGNGNNNGSDSDSDSEGSDSNHSTSDD*
```

B) Translation after removing the first nucleotide of DPP (-1 frameshift).

```
MIPIAVMNLMAMMMLIQKVTITAVAEEMLLITLMNQKIMAMAVTQKEQKMMTVIAHQTLIIVTVMAMVTMG
MMTMTNQTVAKVNQIAVTVIVVIAAIAVIVVTAVTVTAVIATVAVIVTAVTVTAVIAVTVIVVIAAIAVTV
VTAVIAVTVVIVVTAVTASQTAANQRATAVIVTVSQTAVTATAVTVVTTVIAATAAIAVTAVIVVTAVIAV
TAAVAVTAATAVIVVTVVTAAIAVRAVIVVTAVIVTAVIVVTAVIVTAAIVTAATAAIAVTAATAVTAVIA
VTAATAVTVAIAVTAATAVTAVIAVTAVIVVTAATAVIATTAAIAVTAVIAATAVIAATAVIAVIAVTAVI
ATAAIAVTAVIVVTAAIAATAVIAATAAIAVTAVIATAAIEVTVVIVVTAAIAVTAATAVTAVIAVTATKA
AIAVTAVIAATAVIVTAVIAATAVTAVIAATAVIAVKAVIVVTTAIAVTAATAVTAVIAVTAVIVVTAAIA
VTAATAVTAVIAIAATAVTAATAAIAVTAVIAVTAVTAVIAATAVIAVTAVTAVIAVIVVTAAIAVTAATA
VTAAIAVTAAIAVTAAIAVTAVTAAIAVTAVTAATAVIAVTAVTAATAVIAVKAVIAVTAAIAVTAAIAAT
AATAAIAVTAAIAVTAVTAAIAVTAVTAATAVIAVTAATAVTAVTAVKAATAVTAATAVTAAIAATAATAA
IAVTAAIAVIAATAVIAVTAATAAIAATAAIVVIAVTAVTAATAVTAATAVTAATAVTAMKAATAVTAAIA
VTAATAVTAATAVIAVTAHLTAMMRVTARASLVTVTTMEVTVTVTVKAVTVTTQPVMIRTKEKPVRFLL*
```

C) Translation after removing the first two nucleotides of DPP (-2 frameshift).

```
*SQ*Q**I*WQ**C*FRK*Q*QQ*PRRCFL*L**IKR*WQWQ*LKRSRR**Q**HIRH****Q*WQW*QWE
**Q*QIRQWQR*IR*Q*Q*****QQ*Q****QQ*Q*QQ**QQ*Q***QQ*Q*QQ**Q*Q*****QQ*Q*Q*
*QQ**Q*Q*****QQ*QQVRQQQIRERQQ***Q*VRQQ*QQQQ*Q**QQ**QRQQQ*Q*QQ****QQ**Q*
QQQ*Q*QQQQQ****Q**QQQ*Q*EQ****QQ***QQ****QQ***QQR**QQQQQR*Q*QQQQQ*QQ**Q
*QQQQQ*Q*R*Q*QQQQQ*QQ**Q*QQ****QQQQQ**QRQQQ*Q*QQ**QQQQ**QQQQ**Q**Q*QQ**
RQQQ*Q*QQ****QQR*QQQQ**QRQQR*Q*RQ**RQQQ*K*Q*****QQR*Q*QQQQQ*QQ**Q*QQRKQ
Q*Q*QQ**QQQQ***QQ**QQQQ*QQ**QQQQ**Q*KQ****QQQ*Q*QQQQQ*QQ**Q*QQ****QQQ*Q
*QQQQQ*QQ**Q*QRQQ*QQQQQR*Q*QQ**Q*QQ*QQ**QQQQ**Q*QQ*QQ**Q****QQQ*Q*QQQQQ
*QQR*Q*QQR*Q*QQR*Q*QQ*QQQ*Q*QQ*QQRQQ**Q*QQ*QQRQQ**Q*KQ**Q*QQQ*Q*QQR*QRQ
QRQQR*Q*QQR*Q*QQ*QQQ*Q*QQ*QQRQQ**Q*QQQQQ*QQ*QQ*KQRQQ*QQRQQ*QQR*QRQQRQQR
*Q*QQQ*Q**QRQQ**Q*QQRQQR*QRQQR****Q*QQ*QQRQQ*QQRQQ*QQRQQ*QQ*KQRQQ*QQR*Q
*QQQQQ*QQRQQ**Q*QHI*QQ**E*QPEQVW*R*QQWK*Q*Q*Q*RQ*Q*PLNQ**LEQKKNP*
```

Figure S3. Human DPP reference sequence translated in three reading frames. ***A:*** The human DPP amino acid sequence. ***B:*** Translation of the same sequence as in ***A*** after removing 1 nucleotide. Note all -1 frameshifts in DPP lead to a large missense protein. ***C:*** Translation of the same sequence as in ***A*** after removing 2 nucleotides. Note the longest possible missense sequence is 13 amino acids before hitting a stop codon (*).

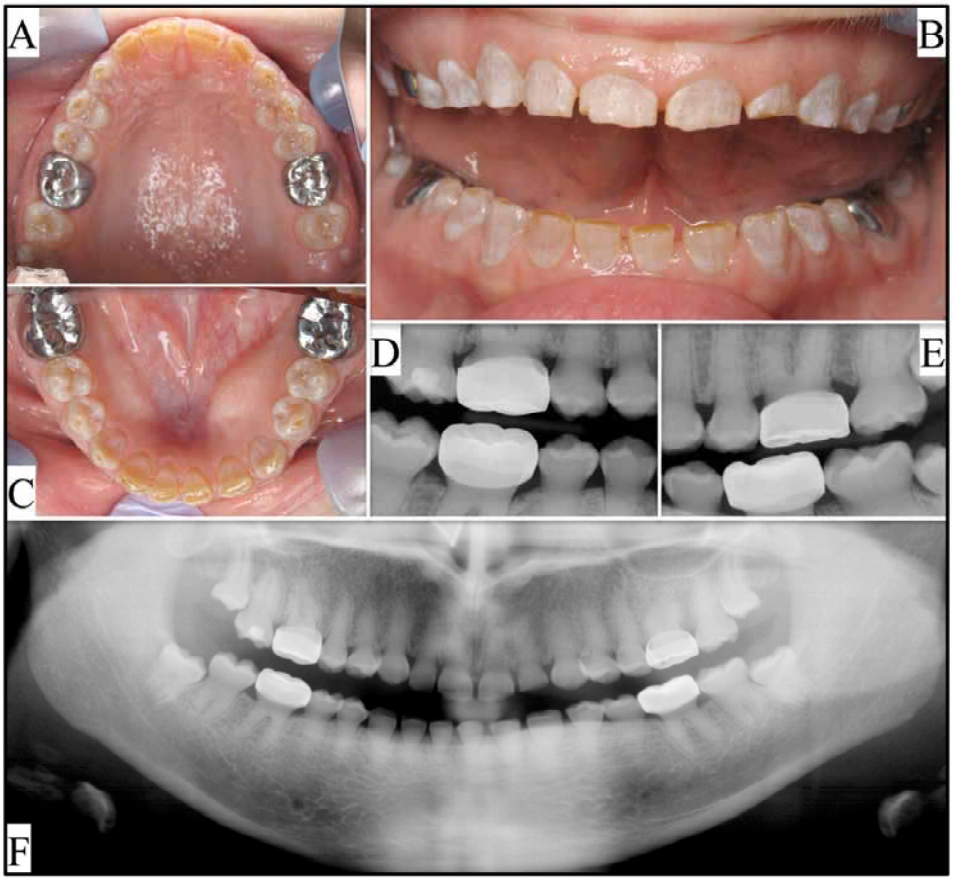

Figure S4. Oral photographs and dental radiographs of the family 1 proband (IV:1). This female subject was 18 years old at the time of documentation. ***A:*** Maxillary occlusal photograph; ***B:*** mandibular occlusal photograph; ***C:*** frontal photograph. The dental crowns are translucent with vertical grooves of hypoplastic enamel. There are spaces between the anterior teeth. ***D:*** Right bitewing radiograph; ***E:*** left bitewing radiograph; ***F:*** panoramic radiograph. The pulp cambers and root canals of all the teeth except the third molars are obliterated. The roots of many teeth appear shorter than normal, particularly the anteriors. The premolar and molar crowns appear bulbous.

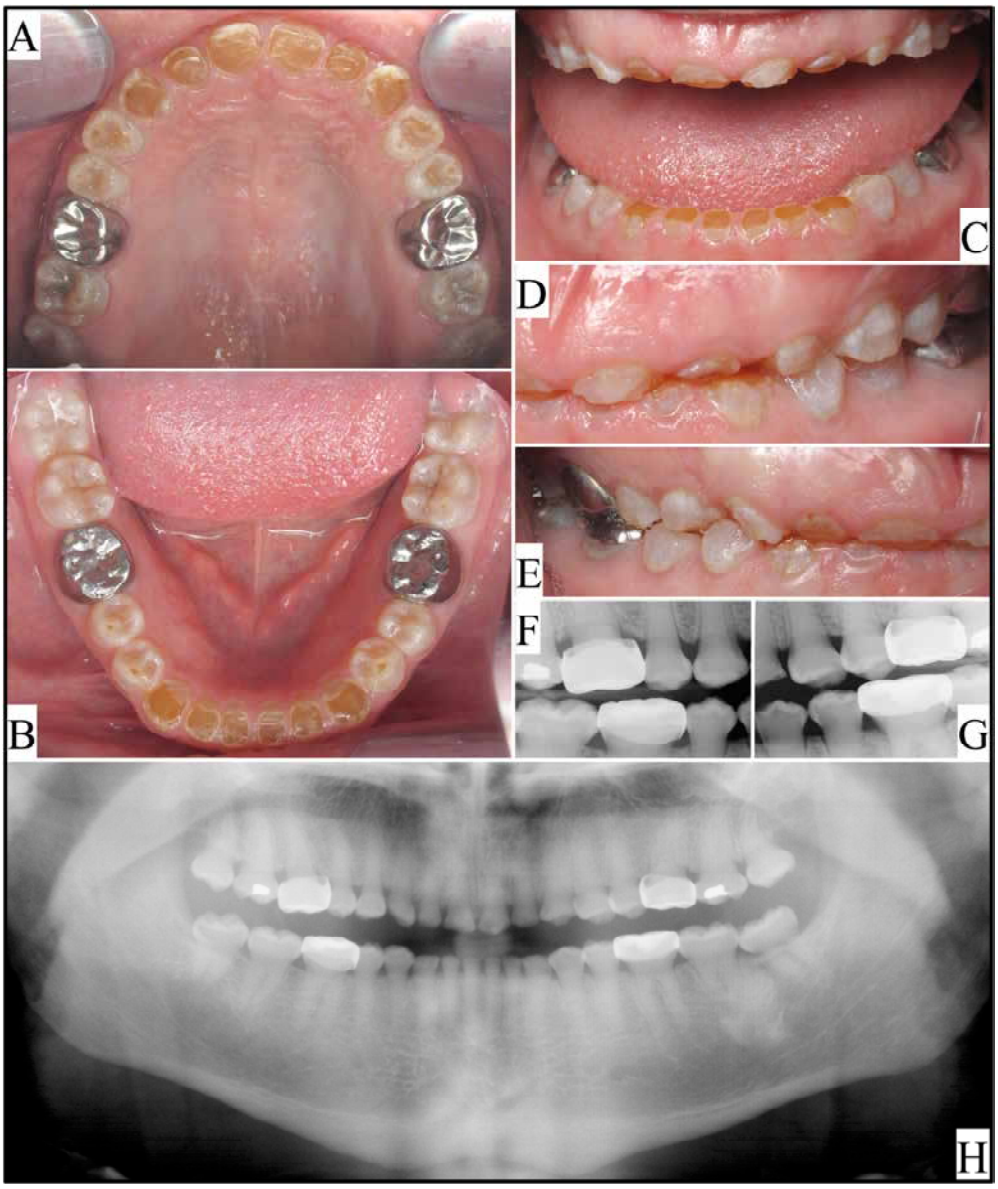

Figure S5. Oral photographs and dental radiographs of family 1 proband's affected youngest brother (IV:3). ***A:*** Maxillary occlusal; ***B:*** mandibular occlusal; ***C:*** mandibular frontal; ***D:*** left buccals; ***E:*** right buccals; ***F:*** right bitewing radiograph; ***G:*** left bitewing radiograph; ***H:*** panoramic radiograph.

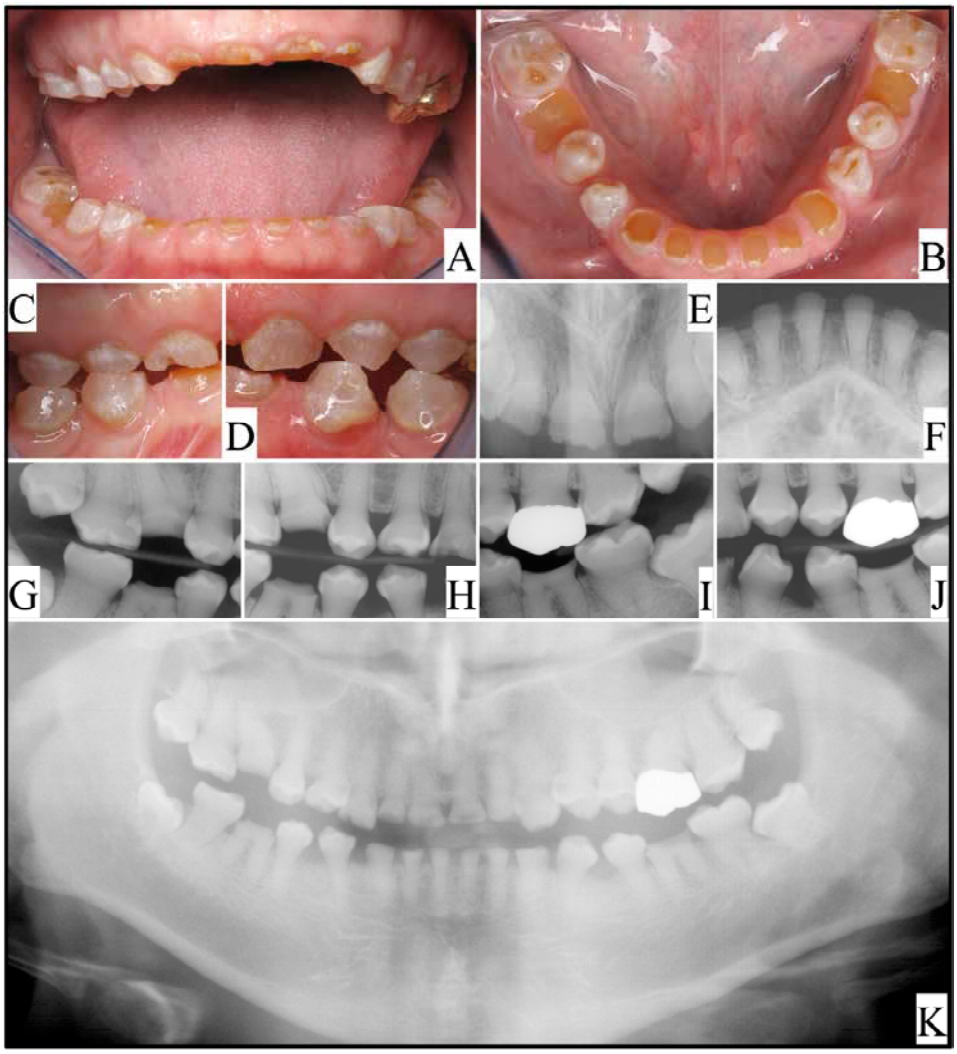

Figure S6. Oral photographs and dental radiographs of family 1 proband's other affected brother (IV:2). This male subject was 17 years old at the time of documentation. ***A:*** Frontal photograph; ***B:*** mandibular occlusal; ***C:*** right buccals; ***D:*** left buccals; ***E:*** maxillary anterior periapical radiograph; ***F:*** mandibular anterior periapical radiograph; ***G-H:*** right bitewing radiographs; ***I-J:*** left bitewing radiographs; ***K:*** panoramic radiograph. Crowns are translucent; first molars show failed restoration with crowns. Pulp cambers and root canals are obliterated of all teeth, except erupting third molars.

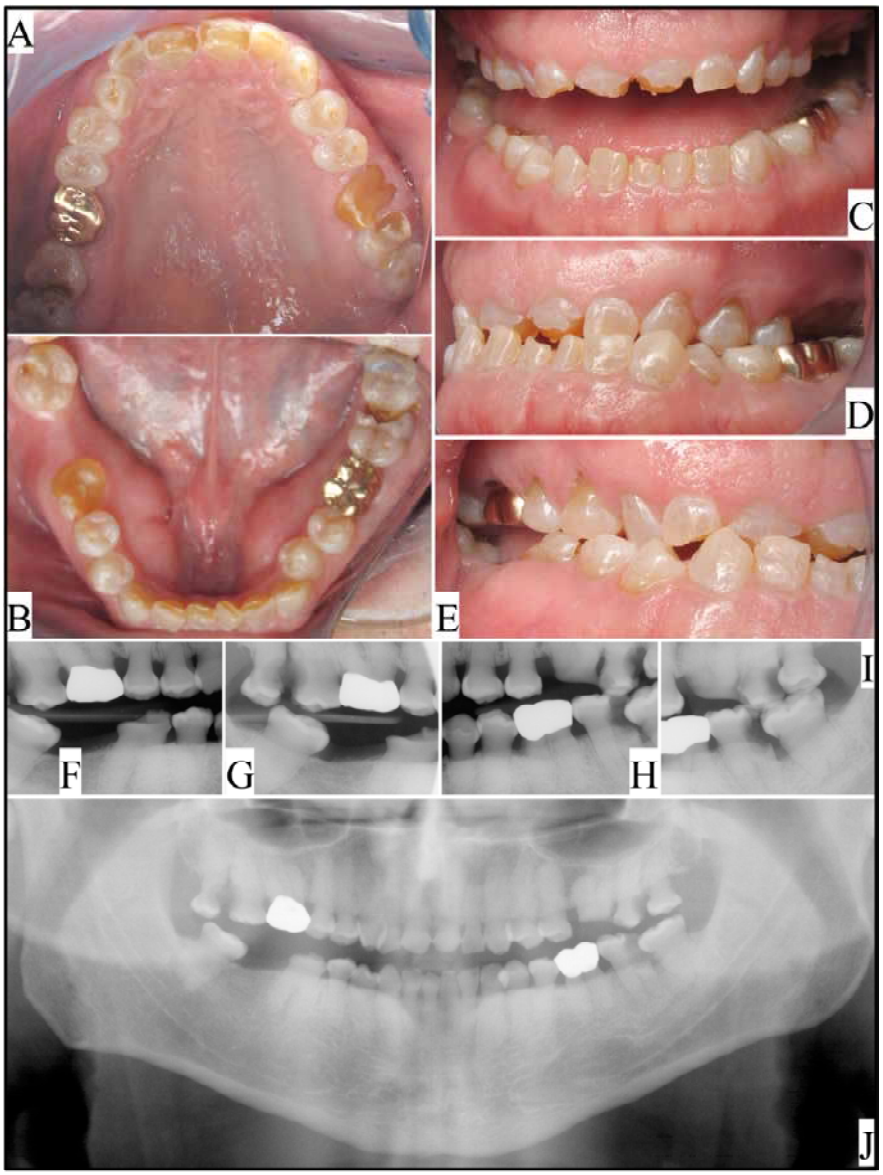

Figure S7. Oral photographs and dental radiographs of the family 1 affected father (III:7).

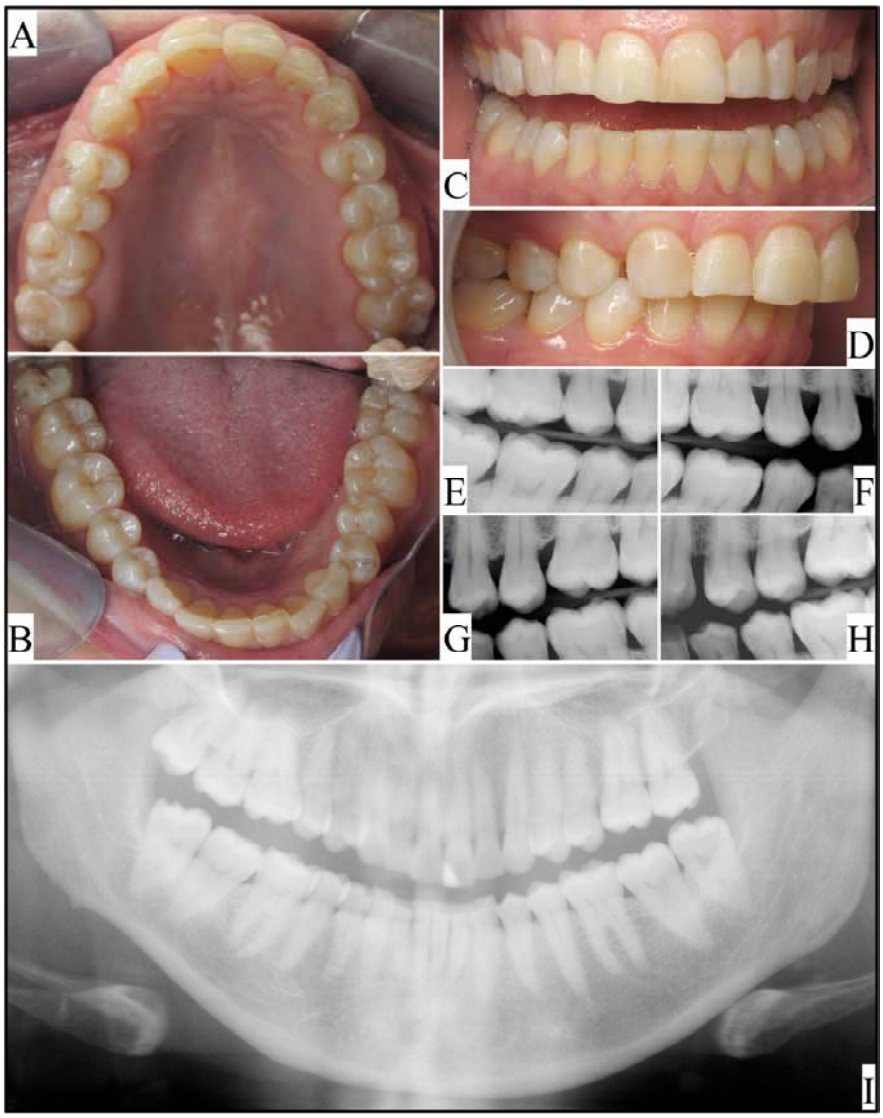

Figure S8. Oral photographs and dental radiographs of the family 1 unaffected mother (III:8).

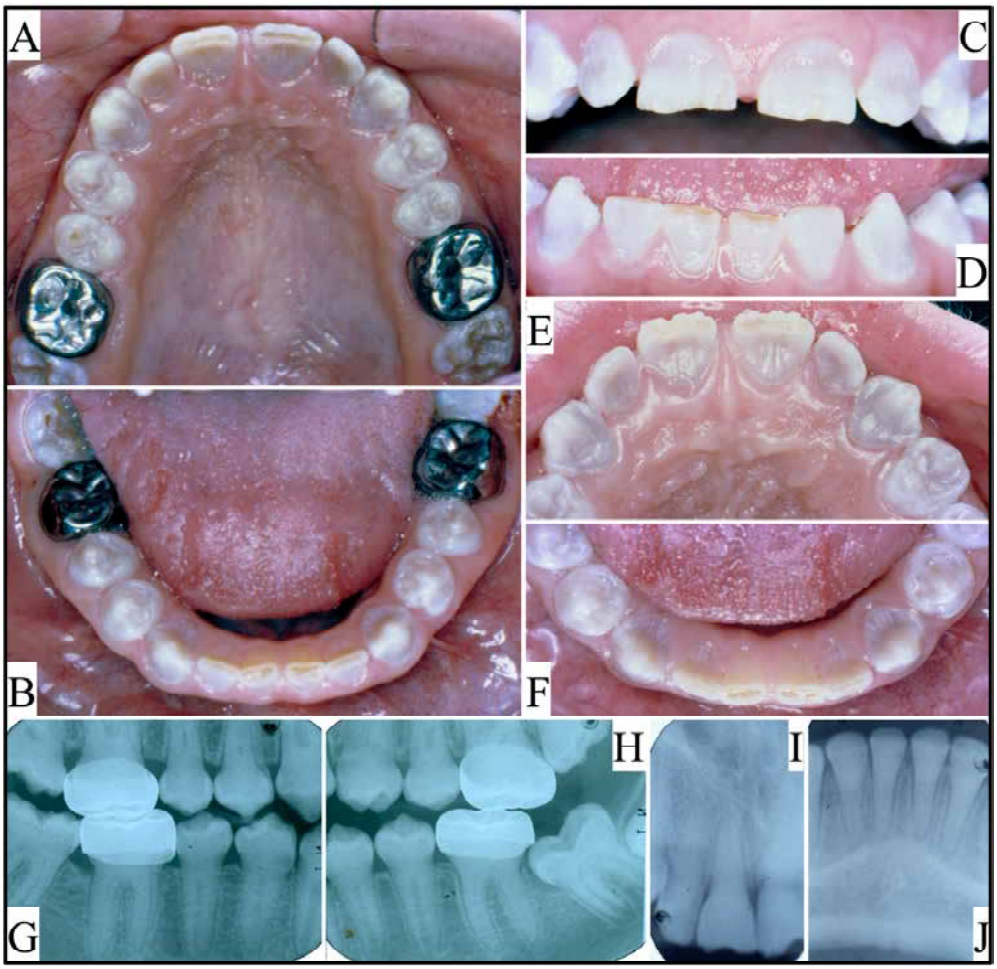

Figure S9. Oral photographs and dental radiographs of family 2 proband (IV:2). This male subject was 12 years old at the time of documentation. ***A:*** Maxillary occlusal photograph; ***B:*** mandibular occlusal photograph; ***C:*** maxillary frontal photograph; ***D:*** mandibular frontal photograph; ***E:*** maxillary anterior occlusal photograph; ***F:*** mandibular anterior occlusal photograph; ***G:*** right bitewing radiograph; ***H:*** left bitewing radiograph; ***I:*** maxillary anterior periapical; ***J:*** maxillary anterior periapical. The crowns are mostly translucent with some chalky white areas. The vertical grooves of enamel defect are evident, especially on the lingual surfaces of lower anterior teeth. The incisal edges of the anterior teeth exhibit moderate attrition. Pulp chambers and root canals are mostly obliterated in the incisors and first molars, but can be detected in other teeth, especially those that are still forming their roots.