**Heterozygous *COL17A1* variants are a frequent cause of Amelogenesis Imperfecta**

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**Supplementary Files**

1. **Table S1:** *COL17A1* variants associated with JEB (Database search) and their distribution in different protein domains.

The *COL17A1* gene encodes a transmembrane protein collagen XVII consisting of 1497 amino acid residues. The amino acid residues in different domains are as defined below [1, 2].

| Domains | Exons | Amino Acid residues | Total Amino Acid |
|---|---|---|---|
| Endodomain | exon 2-exon 17 | 1-466 | 466 |
| Transmembrane | exon 17 | 466-489 | 23 |
| Ectodomain | exon 18-exon 56 | 490-1498 | 1008 |
| | | | 1497 |

By combining a literature search on the NCBI database (https://pubmed.ncbi.nlm.nih.gov/) with access to the HGMD professional database, we identified 232 *COL17A1* variants reported to cause JEB [3]. 172 of these were located in the ectodomain, 3 in the transmembrane domain and 57 in the endodomain.

| Variant | Exonic Function | Domain |
|---|---|---|
| c.-11-2A>G | Splice | Endodomain |
| c.2T>A;p.0? | Missense | Endodomain |
| c.2T>C;p.0? | Missense | Endodomain |
| c.25C>T;p.(Arg9*) | Premature termination codon | Endodomain |
| c.51+1G>A | Splice | Endodomain |
| c.56delT;p.(Val19Alafs*5) | Frameshift small indels | Endodomain |
| c.82dupA;p.(Thr28Asnfs*15) | Frameshift small indels | Endodomain |
| c.158del; p.(Glu53fs) | Frameshift small indels | Endodomain |
| c.202delA;p.(Thr68Leufs*106) | Frameshift small indels | Endodomain |
| c.209-210insCA | Frameshift small indels | Endodomain |
| c.213-214dupAC;p.(Arg72Hisfs*103) | Frameshift small indels | Endodomain |
| c.214C>T:p.(Arg72*) | Premature termination codon | Endodomain |
| c.366dup;p.(Arg123fs) | Frameshift small indels | Endodomain |
| c.372_373insA;p.(Glu125Argfs*17) | Frameshift small indels | Endodomain |
| c.380-1G>A | Splice | Endodomain |
| c.412C>T;p.(Arg138*) | Premature termination codon | Endodomain |
| c.418_419delAG;p.(Ser140*) | Frameshift small indels | Endodomain |
| c.426delT p.(Ile142fs) | Frameshift small indels | Endodomain |
| c.427C>T;p.(Arg143*) | Premature termination codon | Endodomain |
| c.433C>T;p.(Arg145*) | Premature termination codon | Endodomain |
| c.460C>T;p.(Arg154*) | Premature termination codon | Endodomain |
| c.464-2A>G | Splice | Endodomain |
| c.505C>T;p.(Arg169*) | Premature termination codon | Endodomain |
| c.520_521del;p.(Ser174fs) | Frameshift small indels | Endodomain |
| c.558_567del;p.(Lys187Leufs*7) | Frameshift small indels | Endodomain |
| c.569-2A>G | Splice | Endodomain |

| | | |
|---|---|---|
| c.621C>G;p.(Tyr207*) | Premature termination codon | Endodomain |
| c.655dupC;p.(His219Profs*118) | Frameshift small indels | Endodomain |
| c.675_688dup14;p.(Ser230Cysfs*14) | Frameshift small indels | Endodomain |
| c.718delA;p.(Thr240Profs*52) | Frameshift small indels | Endodomain |
| c.772G>A;p.(Gly258Arg) | Missense | Endodomain |
| c.772G>T;p.(Gly258*) | Premature termination codon | Endodomain |
| c.779delC;p.(Pro260Glnfs*32) | Frameshift small indels | Endodomain |
| c.783delC;p.(Asn261Lysfs*31) | Frameshift small indels | Endodomain |
| c.794C>G;p.(Ser265Cys) | Missense | Endodomain |
| c.823delC;p.(Ser275fs) | Frameshift small indels | Endodomain |
| c.884delC;p.(His296fs) | Frameshift small indels | Endodomain |
| c.980-1G>C | Splice | Endodomain |
| c.997C>T;p.(Glu333*) | Premature termination codon | Endodomain |
| c.1139C>T;p.(Ala380Val) | Missense | Endodomain |
| c.1141+2T>C | Splice | Endodomain |
| c.1141+5G>A | Splice | Endodomain |
| c.1179delA;p.(Ala394Leufs*9) | Frameshift small indels | Endodomain |
| c.1239_1240delGT;p.(Ser414Hisfs*87) | Frameshift small indels | Endodomain |
| c.1260delC;p.(Thr421Leufs*72) | Frameshift small indels | Endodomain |
| c.1267+1G>T | Splice | Endodomain |
| c.1267+2T>C | Splice | Endodomain |
| c.1268-2A>G | Splice | Endodomain |
| p.Ile18del389 | Large deletion (exon2-15) | Endodomain |
| c.1268-267_1465+369del834;p.(Asp423Aladel66) | Large Deletion | Endodomain+Transmembrane |
| c.1284delA;p.(Ser429fs) | Frameshift small indels | Endodomain |
| c.1285delA;p.(Ser429fs) | Frameshift small indels | Endodomain |
| c.1336G>A;p.(Gly446Ser) | Missense | Endodomain |
| c.1365delC;p.(Trp455fs) | Frameshift small indels | Endodomain |
| c.1372+1G>T | Splice | Endodomain |
| c.1374C>A;p.(Cys458*) | Premature termination codon | Endodomain |
| c.1392G>A;p.(Trp464*) | Premature termination codon | Endodomain |
| c.1395G>A;p.(Trp465*) | Premature termination codon | Endodomain |
| c.1445T>C;p.(Leu482Pro) | Missense | Transmembrane |
| c.1465+2T>C | Splice | Transmembrane |
| c.1480_1482dupAAG;p.(Lys494dup) | In frame small indel | Ectodomain |
| c.1490_1491delCGinsT;p.(Ala497Valfs*23) | Frameshift small indels | Ectodomain |
| c.1507delG;p.(Glu503Argfs*17) | Frameshift small indels | Ectodomain |
| c.1601delA;p.(Asp534Alafs*19) | Frameshift small indels | Ectodomain |
| c.1601_1602insG;p.(Asp534Glufs*10) | Frameshift small indels | Ectodomain |
| c.1612delA;p.(Ile538Leufs*15) | Frameshift small indels | Ectodomain |
| c.1616G>A;p.(Gly539Glu) | Missense | Ectodomain |
| c.1696C>T;p.(Arg566*) | Premature termination codon | Ectodomain |
| c.1706delA;p.(Pro569fs) | Frameshift small indels | Ectodomain |
| c.1745-2A>C | Splice | Ectodomain |
| c.1745-2A>G | Splice | Ectodomain |
| c.1750C>T;p.(Arg584*) | Premature termination codon | Ectodomain |

| | | |
|---|---|---|
| c.1772-2A>C | Splice | Ectodomain |
| c.1817G>A;p.(Gly606Asp) | Missense | Ectodomain |
| c.1826G>A;p.(Gly609Asp) | Missense | Ectodomain |
| c.1834G>A;p.(Gly612Arg) | Missense | Ectodomain |
| c.1834G>C;p.(Gly612Arg) | Missense | Ectodomain |
| c.1852G>A;p.(Gly618Ser) | Missense | Ectodomain |
| c.1861G>A;p.(Gly621Ser) | Missense | Ectodomain |
| c.1877-2A>C | Splice | Ectodomain |
| c.1880G>T;p.(Gly627Val) | Missense | Ectodomain |
| c.1880delG;p.(Gly627Alafs*56) | Frameshift small indels | Ectodomain |
| c.1898G>A;p.(Gly633Asp) | Missense | Ectodomain |
| c.1992_1995delGGGT;p.(Gly665Profs*17) | Frameshift small indels | Ectodomain |
| c.2002+2T>G | Splice | Ectodomain |
| c.2003-1G>C | Splice | Ectodomain |
| c.2062delC;p.(Arg688Glufs*4) | Frameshift small indels | Ectodomain |
| c.2062C>T;p.(Arg688*) | Premature termination codon | Ectodomain |
| c.2227+153_2336-318del | Large Deletion | Ectodomain |
| c.2228-101_2263+70delins15 | Frameshift small indels | Ectodomain |
| c.2237delG;p.(Gly746Alafs*53) | Frameshift small indels | Ectodomain |
| c.2240delC;p.(Pro747Glnfs*52) | Frameshift small indels | Ectodomain |
| c.2251C>T;p.(Gln751*) | Premature termination codon | Ectodomain |
| c.2282_2283delGG;p.(Gly761Aspfs*40) | Frameshift small indels | Ectodomain |
| c.2336-1G>T | Splice | Ectodomain |
| c.2336-2A>G | Splice | Ectodomain |
| c.2342delG;p.(Thr781fs) | Frameshift small indels | Ectodomain |
| c.2350C>T;p.(Gln784*) | Premature termination codon | Ectodomain |
| c.2363-2A>G | Splice | Ectodomain |
| c.2363dup;p.(Leu789Thrfs*13) | Frameshift small indels | Ectodomain |
| c.2383C>T;p.(Arg795*42) | Premature termination codon | Ectodomain |
| c.2407G>T;p.(Gly803*) | Premature termination codon | Ectodomain |
| c.2434+1G>A | Splice | Ectodomain |
| c.2441-2A>G | Splice | Ectodomain |
| c.2441-1G>T | Splice | Ectodomain |
| c.2468-2A>G | Splice | Ectodomain |
| c.2468C>A;p.(Pro823Gln) | Missense | Ectodomain |
| c.2488G>A;p.(Gly830Arg) | Missense | Ectodomain |
| c.2496dupT;p.(Gly833Argfs*22) | Frameshift small indels | Ectodomain |
| c.2518del10 | Frameshift small indels | Ectodomain |
| c.2520dupT;p.(Ala841Cysfs*14) | Frameshift small indels | Ectodomain |
| c.2544delA;p.(His849Ilefs*217) | Frameshift small indels | Ectodomain |
| c.2551+1G>A | Splice | Ectodomain |
| c.2561_2565delATTTA;p.(Asn854Thrfs*109) | Frameshift small indels | Ectodomain |
| c.2563_2564delTT;p.(Leu855Thrfs*109) | Frameshift small indels | Ectodomain |
| c.2564T>G;p.(Leu855*) | Premature termination codon | Ectodomain |
| c.2564T>A;p.(Leu855*) | Premature termination codon | Ectodomain |
| c.2566C>T;p.(Gln856*) | Premature termination codon | Ectodomain |

| | | |
|---|---|---|
| c.2585_2586delCC;p.(Pro862Argfs*102) | Frameshift small indels | Ectodomain |
| c.2635C>T;p.(Arg879*) | Premature termination codon | Ectodomain |
| c.2666delTT | Frameshift small indels | Ectodomain |
| c.2690insT;p.(Ser898fs) | Frameshift small indels | Ectodomain |
| c.2706dup;p.(Phe903Leufs*62) | Frameshift small indels | Ectodomain |
| c.2716G>A;p.(Gly906Ser) | Missense | Ectodomain |
| c.2723dupC;p.(Gly909Argfs*56) | Frameshift small indels | Ectodomain |
| c.2725G>C;p.(Gly909Arg) | Missense | Ectodomain |
| c.2776delA;p.(Arg926Glufs*140) | Frameshift small indels | Ectodomain |
| c.2840_2844delTCAAC;p.(Leu947Profs*16) | Frameshift small indels | Ectodomain |
| c.2861delG;p.(Gly954Alafs*112) | Frameshift small indels | Ectodomain |
| c.2875delC;p.(Gln959Argfs*107) | Frameshift small indels | Ectodomain |
| c.2881delA | Frameshift small indels | Ectodomain |
| c.2897-9G>A | Splice | Ectodomain |
| c.2897-2A>C | Splice | Ectodomain |
| c.2944del;p.(Glu982Lysfs84*) | Frameshift small indels | Ectodomain |
| c.2948-1G>C | Splice | Ectodomain |
| c.2965delG;p.(Met989fs) | Frameshift small indels | Ectodomain |
| c.2971G>A;p.(Val991Met) | Missense | Ectodomain |
| c.2972delT;p.(Val991Glyfs*75) | Frameshift small indels | Ectodomain |
| c.2975C>A;p.(Ser992*) | Premature termination codon | Ectodomain |
| c.2993dupC;p.(Gly999Trpfs*22) | Frameshift small indels | Ectodomain |
| c.3000-3008del;p.(Pro1003_Gly1005del) | In frame small indel | Ectodomain |
| c.3002-2A>C | Splice | Ectodomain |
| c.3046C>T;p.(Gln1016*) | Premature termination codon | Ectodomain |
| c.3053-1G>C | Splice | Ectodomain |
| c.3067C>T;p.(Gln1023*) | Premature termination codon | Ectodomain |
| c.3071-6C>A | Splice | Ectodomain |
| c.3131delC;p.(Pro1044Glnfs*22) | Frameshift small indels | Ectodomain |
| c.3164delT;p.(Phe1055Serfs*11) | Frameshift small indels | Ectodomain |
| c.3171_3173delCTC;p.(Tyr1057*) | Frameshift small indels | Ectodomain |
| c.3175delG;p.(Glu1059Serfs*7) | Frameshift small indels | Ectodomain |
| c.3193_3208del16;p.(Val1065Leufs*35) | Frameshift small indels | Ectodomain |
| c.3198C>T;p.(Ser1066Ser | Splice | Ectodomain |
| c.3205C>T;p.(Arg1069Trp) | Missense | Ectodomain |
| c.3236delC;p.(Ser1079Cysfs*26) | Frameshift small indels | Ectodomain |
| c.3269dupT;p.(Leu1091Alafs*5) | Frameshift small indels | Ectodomain |
| c.3275A>C;(Gln1092Pro) | Missense | Ectodomain |
| c.3288_3295del8;p.(Arg1097Profs*33) | Frameshift small indels | Ectodomain |
| c.3292C>T;p.(Gln1098*) | Premature termination codon | Ectodomain |
| c.3301C>T;p.(Arg1101Cys) | Missense | Ectodomain |
| c.3327delT;p.(Pro1110Argfs*21) | Frameshift small indels | Ectodomain |
| c.3408delC;p.(Tyr1137Thrfs*114) | Frameshift small indels | Ectodomain |
| c.3481dupT;p.(Tyr1161fs*2) | Frameshift small indels | Ectodomain |
| c.3482_3483del;p.(Tyr1161*) | Frameshift small indels | Ectodomain |
| c.3487G>T;p.(Glu1163*) | Premature termination codon | Ectodomain |

| | | |
|---|---|---|
| c.3496_3497delTC;p.(Ser1166Leufs*6) | Frameshift small indels | Ectodomain |
| c.3505C>T;p.(Arg1169*) | Premature termination codon | Ectodomain |
| c.3509-1G>C | Splice | Ectodomain |
| c.3513delC;p.(Glu1172fs) | Frameshift small indels | Ectodomain |
| c.3514ins25 | Frameshift small indels | Ectodomain |
| c.3539dupC;p.(Pro1180Profs*62) | Frameshift small indels | Ectodomain |
| c.3548delC;p.(Pro1183Argfs*68) | Frameshift small indels | Ectodomain |
| c.3569dupG;p.(Asn1191Glnfs*51) | Frameshift small indels | Ectodomain |
| c.3569_3570insT;p.(Asn1191Glnfs*51) | Frameshift small indels | Ectodomain |
| c.3579G>A;p.(Trp1193*) | Premature termination codon | Ectodomain |
| c.3600-3601delCT | Frameshift small indels | Ectodomain |
| c.3613_3616delTTAC;p.(Leu1205Ilefs*45) | Frameshift small indels | Ectodomain |
| c.3615_3619dupACATA | Frameshift small indels | Ectodomain |
| c.3619+2T>C | Splice | Ectodomain |
| c.3673_3674insT;p.(Pro1225fs) | Frameshift small indels | Ectodomain |
| c.3676C>T;p.(Arg1226*) | Premature termination codon | Ectodomain |
| c.3686C>T;p.(Pro1229Leu) | Missense | Ectodomain |
| c.3689dup;p.(V1231Cfs*11) | Frameshift small indels | Ectodomain |
| c.3730G>A;p.(Asp1244Asn) | Missense | Ectodomain |
| c.3740G>A;p.(Arg1247Gln) | Missense | Ectodomain |
| c.3766+1G>A | Splice | Ectodomain |
| c.3766+1G>C | Splice | Ectodomain |
| c.3782G>C;p.(Ser1261Thr) | Missense | Ectodomain |
| c.3786_3789delCATT;p.(Phe1262Leufs*49) | Frameshift small indels | Ectodomain |
| c.3801insC;p.(Gly1268Argfs*25) | Frameshift small indels | Ectodomain |
| c.3806delC;p.(Pro1269Leufs*43) | Frameshift small indels | Ectodomain |
| c.3821_3829delGACCCCCTGinsC;p.(Gly1274Alafs*16) | Frameshift small indels | Ectodomain |
| c.3827dupC;p.(Gly1277Trpfs*16) | Frameshift small indels | Ectodomain |
| c.3865dupA;p.(Ser1289Lysfs*4) | Frameshift small indels | Ectodomain |
| c.3871+1G>A | Splice | Ectodomain |
| c.3871+1G>C | Splice | Ectodomain |
| c.3897_3900delATCT;p.(Val1301Glyfs*10) | Frameshift small indels | Ectodomain |
| c.3899_3900delCT;p.(Ser1300Cysfs*29) | Frameshift small indels | Ectodomain |
| c.3908G>A;p.(Arg1303Gln) | Missense | Ectodomain |
| c.3922delA;p.(Ser1308Alafs*4) | Frameshift small indels | Ectodomain |
| c.4003delTC | Frameshift small indels | Ectodomain |
| c.4011T>A;p.(Tyr1337*) | Premature termination codon | Ectodomain |
| c.4041T>G;p.(Tyr1347*) | Premature termination codon | Ectodomain |
| c.4045dupG;p.(Ala1349Glyfs*18) | Frameshift small indels | Ectodomain |
| c.4080insGG;p.(Gly1361fs) | Frameshift small indels | Ectodomain |
| c.4088delT;p.(Leu1363fs) | Frameshift small indels | Ectodomain |
| c.4100_4101delTT;p.(Phe1367Cysfs*8) | Frameshift small indels | Ectodomain |
| c.4144del4;p.(Glu1382fs) | Frameshift small indels | Ectodomain |
| c.4145_4148delAGAG;p.(Glu1382Alafs*40) | Frameshift small indels | Ectodomain |
| c.4149_4150insG;p.(Met1384fs) | Frameshift small indels | Ectodomain |
| c.4153C>T;p.(Gln1385*) | Premature termination codon | Ectodomain |

| | | |
|---|---|---|
| c.4156+1G>C | Splice | Ectodomain |
| c.4156+1G>A | Splice | Ectodomain |
| c.4159C>T;p.(Gln1387*) | Premature termination codon | Ectodomain |
| c.4207C>T;p.(Gln1403*) | Premature termination codon | Ectodomain |
| c.4230delC;p.(Gly1411Alafs*12) | Frameshift small indels | Ectodomain |
| c.4231G>A;p.(Gly1411Ser) | Missense | Ectodomain |
| c.4237_4238insC;p.(Ser1413Thrfs*42) | Frameshift small indels | Ectodomain |
| c.4261+1G>C | Splice | Ectodomain |
| c.4265_4266insTT;p.(Thr1423*) | Frameshift small indels | Ectodomain |
| c.4295-1G>C | Splice | Ectodomain |
| c.4307_4310dupTTCA;p.(Gln1437Hisfs*19) | Frameshift small indels | Ectodomain |
| c.4319dupC;p.(Gly1441Trpfs*14) | Frameshift small indels | Ectodomain |
| c.4320delT;p.(Gln1442Lysfs*70) | Frameshift small indels | Ectodomain |
| c.4321delT;p.(Gln1442fs) | Frameshift small indels | Ectodomain |
| c.4324C>T;p.(Gln1442*) | Premature termination codon | Ectodomain |
| c.4335delC;p.(Met1446fs) | Frameshift small indels | Ectodomain |
| c.4410-4413dupCATT | Frameshift small indels | Ectodomain |
| c.4424-5insC | Splice | Ectodomain |
| c.4425_4426insC;p.(Lys1476fs) | Frameshift small indels | Ectodomain |
| c.4425delT | Frameshift small indels | Ectodomain |
| c.4460G>A;p.(Arg1487Gln) | Missense | Ectodomain |
| c.4463-1G>A | Splice | Ectodomain |

**Table S2:** *COL17A1* variants associated with AI (This study and from database search).

| Variant | Exonic Function | Domain | References |
|---|---|---|---|
| c.340;p.(Ser114Valfs*60) | Frameshift small indels | Endodomain | Leeds AI group |
| c.460C>T;p.(Arg154*) | Premature termination codon | Endodomain | Leeds AI group |
| c.541_550del;p.(Asn181Profs*13) | Frameshift small indels | Endodomain | Leeds AI group |
| c.1141+1G>A | Splice | Endodomain | Prasad, M. K., et al. (2016) |
| c.1646G>A;p.(Trp549*) | Premature termination codon | Ectodomain | Prasad, M. K., et al. (2016) |
| c.1745-2A>C | Splice | Ectodomain | Bloch-Zupan, A., et al. (2023) |
| c.1861G>A; p.(Gly621Ser) | Missense | Ectodomain | Leeds AI group |
| c.1873C>T;p.(Arg625*) | Premature termination codon | Ectodomain | Prasad, M. K., et al. (2016) |
| c.2011G>A;p.(Gly671Ser) | Missense | Ectodomain | Leeds AI group |
| c.2030G>A;p.(Gly677Asp) | Missense | Ectodomain | Leeds AI group |
| c.2407G>T;p.(Gly803*) | Premature termination codon | Ectodomain | Prasad, M. K., et al. (2016) |
| c.2435-1G>A | Splice | Ectodomain | Leeds AI group |
| c.2912del;p.(Pro971Glnfs*95) | Frameshift small indels | Ectodomain | Leeds AI group |
| c.2947+2T>C | Splice | Ectodomain | Leeds AI group |
| c.3277+1G>A | Splice | Ectodomain | Leeds AI group |
| c.3297C>A;p.(Tyr1099*) | Premature termination codon | Ectodomain | Leeds AI group |
| c.3327del;p.(Pro1110Argfs*21) | Frameshift small indels | Ectodomain | Bloch-Zupan, A., et al. (2023) |
| c.3397C>T;p.(Arg1133Cys) | Missense | Ectodomain | Leeds AI group |
| c.3456del;p.(Pro1154Leufs*97) | Frameshift small indels | Ectodomain | Leeds AI group |
| c.3462_3463del;p.(Gly1155fs*7) | Frameshift small indels | Ectodomain | Leeds AI group |
| c.3595G>C;p.(Glu1199Gln) | Missense | Ectodomain | Leeds AI group |
| c.3605C>T;p.(Ser1202Leu) | Missense | Ectodomain | Leeds AI group |
| c.4147_4148del;p.(Ser1383Hisfs*71) | Frameshift small indels | Ectodomain | Leeds AI group |

**Table S3:** *COL17A1* variants associated with ERED (From database search).

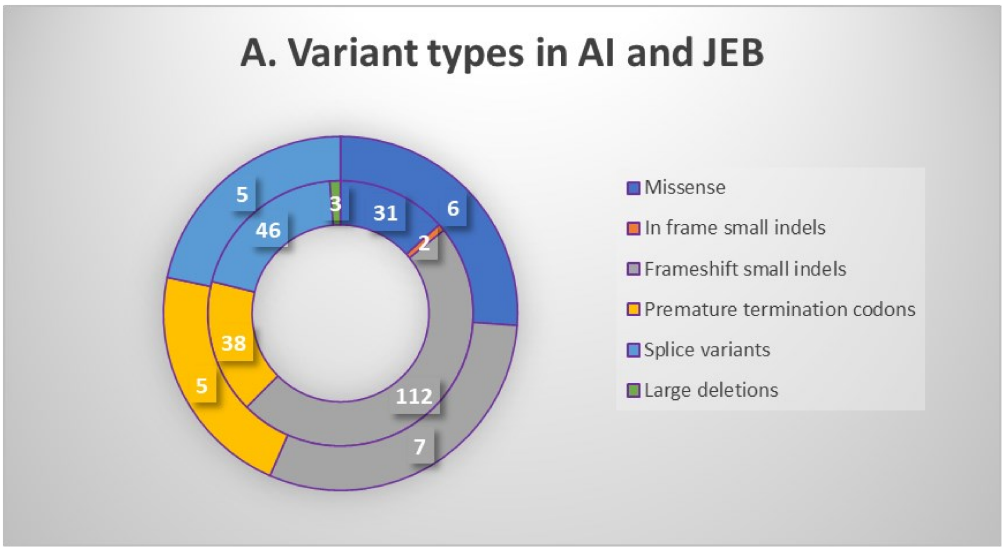
| Variant | Exonic Function | Domain |
|---|---|---|
| c.2816C>T;(Thr939Ile) | Missense | Ectodomain |
| c.3156C>T | Splice | Ectodomain |
| c.3554C>T;(Pro1185Leu) | Missense | Ectodomain |

**Table S4:** Genes included in the smMIP reagent. Reference genome GRCh37/hg19.

| Gene Name | Gene Symbol | OMIM | Genomic Coordinates | Cytoband |
|---|---|---|---|---|
| LAMININ, BETA-3 | *LAMB3* | 150310 | chr1:209,788,218-209,825,820 | 1q32.2 |
| INTEGRIN, BETA-6 | *ITGB6* | 147558 | chr2:160,958,233-161,056,589 | 2q24.2 |
| AMELOTIN | *AMTN* | 610912 | chr4:71,384,298-71,398,459 | 4q13.3 |
| AMELOBLASTIN | *AMBN* | 601259 | chr4:71,457,975-71,473,004 | 4q13.3 |
| ENAMELIN | *ENAM* | 606585 | chr4:71,494,461-71,512,536 | 4q13.3 |
| ODONTOGENESIS-ASSOCIATED PHOSPHOPROTEIN | *ODAPH* | 614829 | chr4:76,481,258-76,491,103 | 4q21.1 |
| FAMILY WITH SEQUENCE SIMILARITY 83 | *FAM83H* | 611927 | chr8:144,806,103-144,815,914 | 8q24.3 |
| COLLAGEN, TYPE XVII, ALPHA-1 | *COL17A1* | 113811 | chr10:105,791,046-105,845,638 | 10q25.1 |
| RECEPTOR EXPRESSED IN LYMPHOID TISSUES | *RELT* | 611211 | chr11:73,087,405-73,108,519 | 11q13.4 |
| MATRIX METALLOPROTEINASE 20 | *MMP20* | 604629 | chr11:102,447,566-102,496,063 | 11q22.2 |
| G PROTEIN-COUPLED RECEPTOR 68 | *GPR68* | 601404 | chr14:91,698,876-91,710,852 | 14q32.11 |
| SOLUTE CARRIER FAMILY 24 (SODIUM/POTASSIUM/CALCIUM EXCHANGER), MEMBER 4 | *SLC24A4* | 609840 | chr14:92,790,152-92,967,825 | 14q32.12 |
| WD REPEAT-CONTAINING PROTEIN 72 | *WDR72* | 613214 | chr15:53,805,938-54,051,859 | 15q21.3 |
| TRANSCRIPTION FACTOR Sp6 | *SP6* | 608613 | chr17:45,922,280-45,928,516 | 17q21.32 |
| DISTAL-LESS HOMEOBOX 3 | *DLX3* | 600525 | chr17:48,067,369-48,072,588 | 17q21.33 |
| FAMILY WITH SEQUENCE SIMILARITY 20, MEMBER A | *FAM20A* | 611062 | chr17:66,531,257-66,597,095 | 17q24.2 |
| ACID PHOSPHATASE 4 | *ACP4* | 606362 | chr19:51,293,672-51,298,481 | 19q13.33 |
| KALLIKREIN-RELATED PEPTIDASE 4 | *KLK4* | 603767 | chr19:51,409,608-51,413,994 | 19q13.41 |
| AMELOGENIN | *AMELX* | 300391 | chrX:11,311,533-11,318,881 | Xp22.2 |

**Figure S1:** A comparison of *COL17A1* variants detected in AI and JEB.

A. Collagen XVII variant types reported in AI (outer ring) and JEB (inner ring) patients: Most variants detected in both AI and JEB are those that lead to frameshift and PTC. Fewer variants are reported in AI (n=23) than in JEB (n=232). The differences observed were not significant.



B. Variant distribution in different domains of collagen XVII reported in AI (outer ring) and JEB (inner ring) patients: A total of 23 variants are reported to be associated with AI, 19 of them occurring in the ectodomain and 4 of them in endodomain. A total of 232 variants were reported to be associated with JEB, 172 of them were occurring in ectodomain, 57 in endodomain and 3 of them in transmembrane domain.

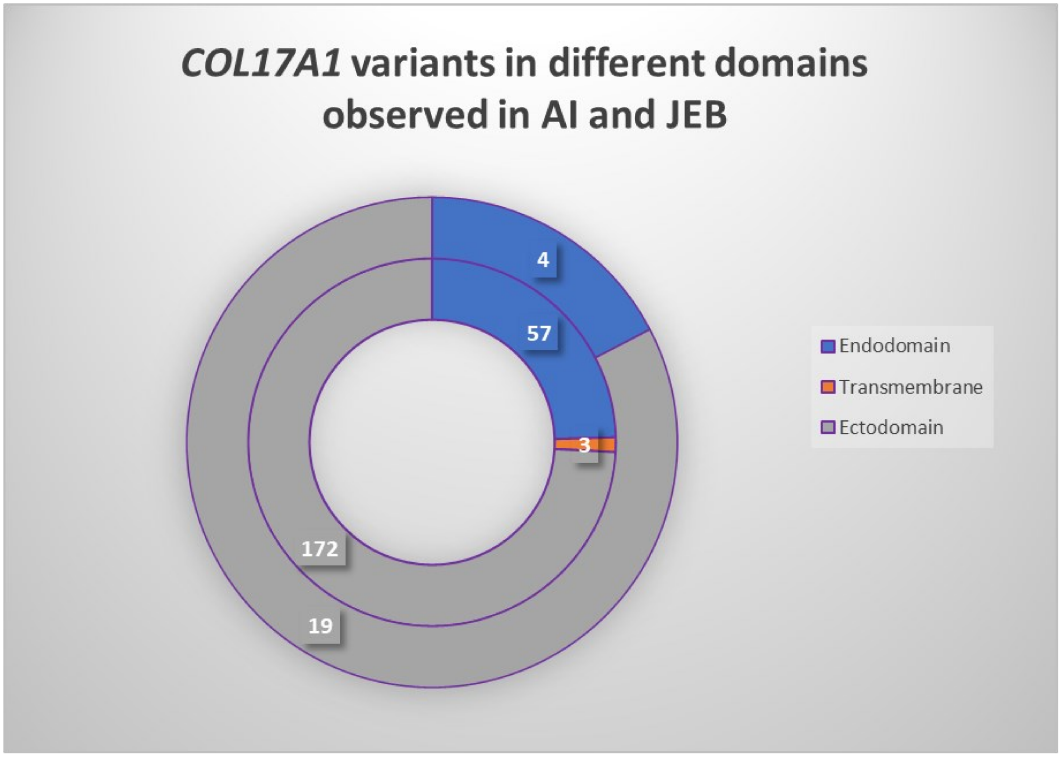


**Figure S2:** Family pedigree and IGV trace of the variant c.340; p.(Ser114Valfs*60) detected in the family 19, generated by using human reference genome hg19.

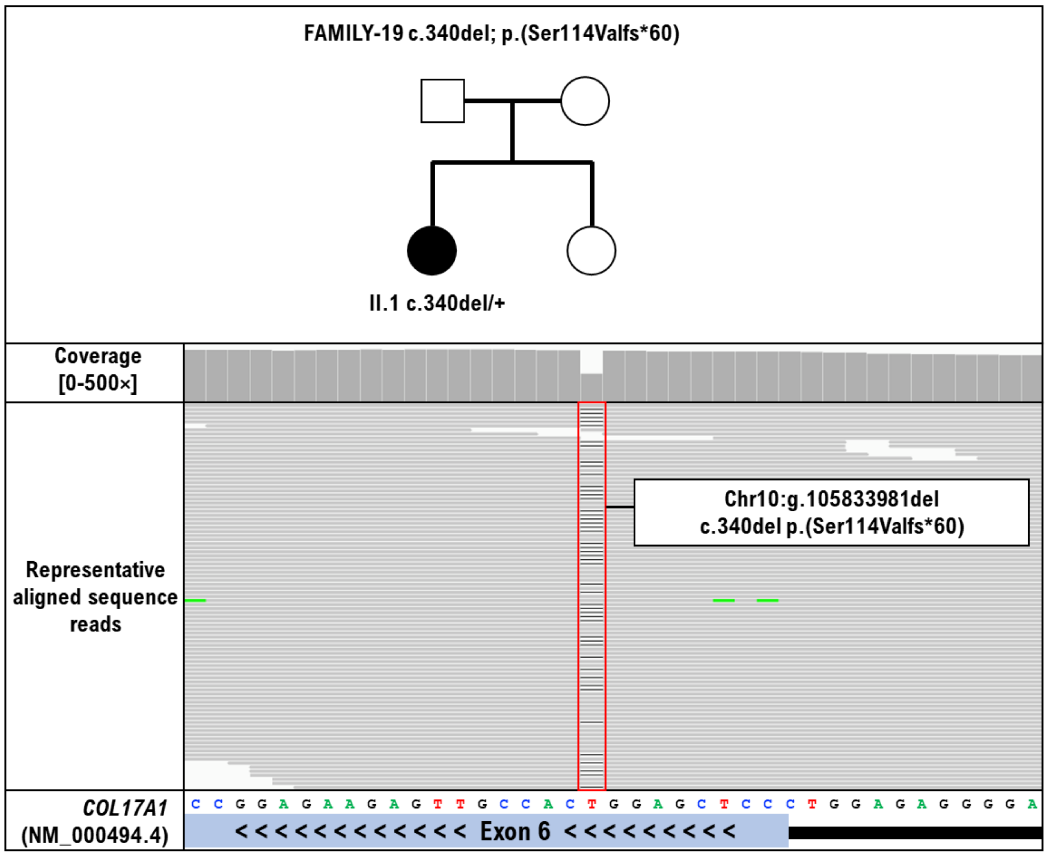
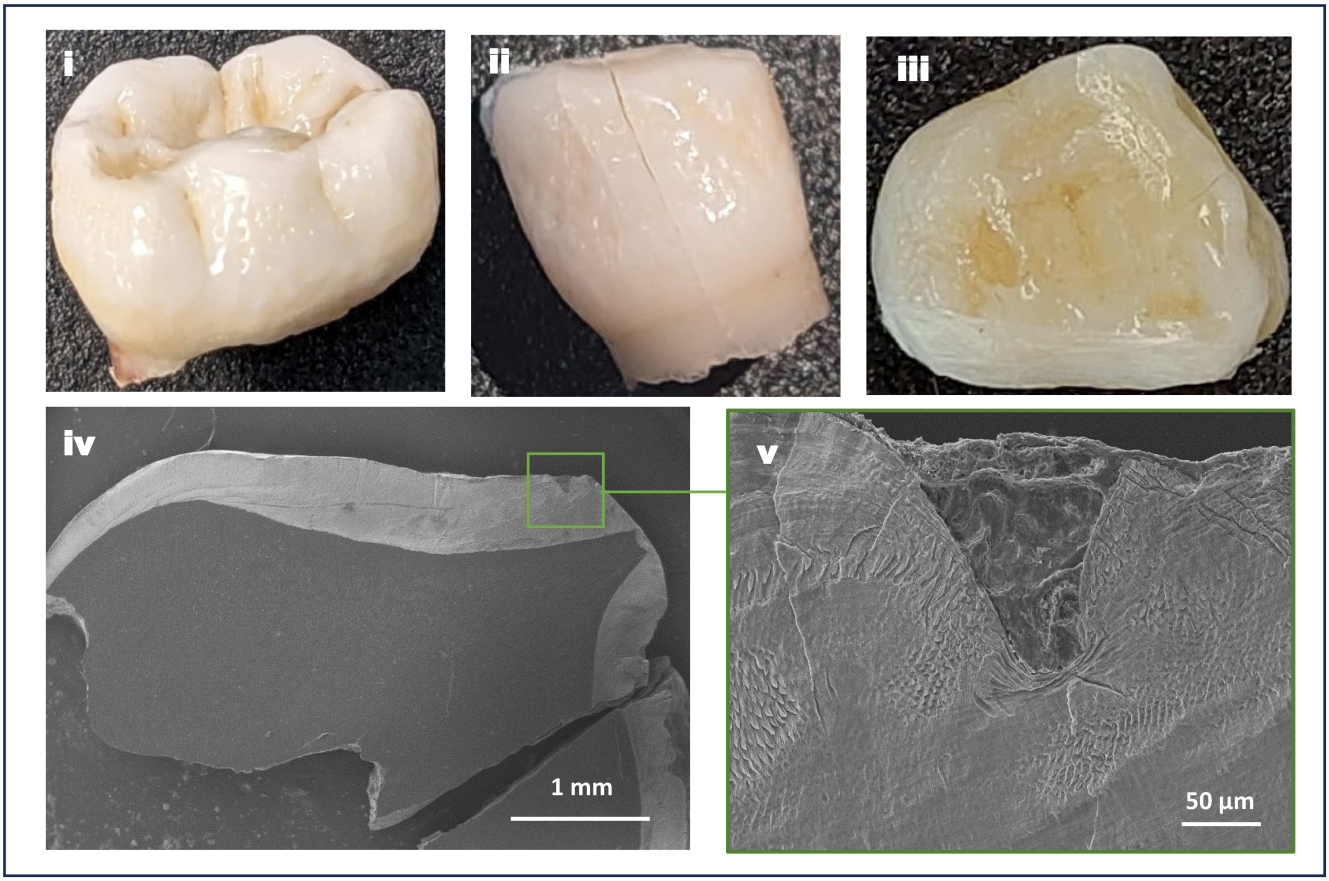


**Figure S3:** Surface pitting observed in primary teeth from different families.
Whole tooth images from the proband of (i) F14, (ii and iii) F9. SEM cross-section images showing surface pit (iv) F9 (inset), (v) Higher resolution of the pit.



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