APPENDICES

APPENDIX A

List of patients and their phenotypes

CGL DNA No.	Sex	Phenotype
779	M	XLHED
785	M	XLHED
786	M	XLHED
805	M	Normal
806	F	Carrier
814	M	XLHED
815	F	Carrier
816	M	Normal
738	M	XLHED
739	M	Normal
740	F	Normal
787	M	XLHED
813	M	XLHED
807	F	Carrier
808	M	Normal
21	F	Hypodontia of multiple permanent teeth
24	M	Hypodontia of 13, 23
431	F	Hypodontia of 13, 23
811	M	Hypodontia of 18, 28
812	F	Normal
436	F	Hypodontia of 32 Fusion of 41 and 42
444	F	Hypodontia of 13, 23
462	F	Hypodontia of 32, 12, 22, 14, 15, 24, 25, 34, 35, 44, 45
463	M	Hypodontia of 42
467	M	Hypodontia of 12
474	M	Hypodontia of 32, 42, 72, 82
495	M	Hypodontia of nine permanent teeth
728	F	Hypodontia of 13, 23,15,25,35,45
809	F	Hypodontia of 18, 28, 38, 48
810	M	Hypodontia of 12
788	F	Hypodontia of 15,34,35,42,44,45

APPENDIX B

Sequences of each exon of EDA

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   1 attccctcgg cgggccgagc ctccctctc tcccgccct cctcctccct ttcccacccc
  61 tcggagtaga gctgcacatg cggctgctcc ctgctccgtc ccgcccagcc actgtcgcgc
 121 aggaacgggt ccctgcagcc cccagccgat ggcaggacag tagccgcctg tcagaggtcg
 181 tgaacggctg aggcagacgc agcggctccc gggcctcaag agagtgggtg tctccggagg
 241 ccatgggcta cccggaggtg gagcgcaggg aactcctgcc tgcagcagcg ccgcgggagc
 301 gagggagcca gggctgcggg tgtggcgggg cccctgcccg ggcgggcgaa gggaacagct
 361 gcctgctctt cctgggtttc tttggcctct cgctggccct ccacctgctg acgttgtgct
 421 gctacctaga gttgcgctcg gagttgcggc gggaacgtgg agccgagtcc cgccttggcg
 481 gctcgggcac ccctggcacc tctggcaccc taagcagcct cggtggcctc gaccctgaca
 541 gccccatcac cagtcacctt gggcagccgt cacctaagca gcagccattg gaaccgggag
 601 aagccgcact ccactctgac tcccaggacg ggcaccagat ggccctattg aatttcttct
 661 tccctgatga aaagccatac tctgaagaag aaagtaggcg tgttcgccgc aataaaagaa
 721 gcaaaagcaa tgaaggagca gatggcccag ttaaaaacaa gaaaaaggga aagaaagcag
 781 gacctcctgg acccaatggc cctccaggac ccccaggacc tccaggaccc cagggacccc
 841 caggaattcc agggattcct ggaattccag gaacaactgt tatgggacca cctggtcctc
 901 caggtcctcc tggtcctcaa ggacccctg gcctccaggg accttctggt gctgctgata
 961 aagctggaac tcgagaaaac cagccagctg tggtgcatct acagggccaa gggtcagcaa
1021 ttcaagtcaa gaatgatctt tcaggtggag tgctcaatga ctggtctcgc atcactatga
1081 accccaaggt gtttaagcta catccccgca gcggggagct ggaggtactg gtggacggca
1141 cctacttcat ctatagtcag gtagaagtat tactacatcaa cttcactgac tttgccagct
1201 atgaggtggt ggtggatgag aagcccttcc tgcagtgcac acgcagcatc gagacgggca
1261 agaccaacta caacacttgc tataccgcag gcgtctgcct cctcaaggcc cggcagaaga
1321 tcgccgtcaa gatggtgcac gctgacatct ccatcaacat gagcaagcac accacgttct
1381 ttggggccat caggctgggt gaagcccctg catcctagat tccccccatt ttgcctctgt
1441 ccgtgccccc tccctgggtt tgggagccag gactcccaga acctctaagt gctgctgtgg
1501 agtgaggtgt attggtgttg cagccgcaga gaaatgcccc agtgttattt attccccagt
1561 gactccaggg tgacaaggcc tgcttgactt tccagaatga ccttgagtta acaggacagt
1621 tgatggagcc ccagggttta catgaagcag aaccttcttt ggttccatgt tgactgactt
1681 atggcatgac tcttcaaccc cgaggtccct gttgtcagat ctattgtttg ttgcactaaa
1741 atgaggatcc agggcagcag gccagagaaa gcaaaggtgc actccagact ctgggggtgg
1801 acatctgacc ccaaggggcc tgctgctcct ctcttgggta gggtagtggc tggggtggag
1861 tgggaaggga gcattgcagc ctaagaagaa ggccagagag ggaaaaggca ggtgcttttg
1921 gcagagacca taagagaaac ctgccaagga gcatccttgg cagtgggaat gttctttctg
1981 ctctatactg tggcctgcag gagggttgga gtgctcttcc cactccagct gacagccaca
2041 ccgtggcagc ttgctgggct ttgggaagtt tgctgtgctt tggaacaatc acagggaatg
2101 gccacaaacc tgcccgccta agaccctgaa tccgtacttg ggtcacatga ctctcatttt
2161 atttacagct gtgctccaca ctcagaaaat tccctggggt caccttctag ttgcccccat
2221 tcccagcctg actagaactc ctgtcttctt tctccatgga gcctacctct gtctgagaca
2281 ggtgcctaac ctgggacctg tggtcatgtg agtctgggat attctttagc ttacctgggc
2341 acaaacagaa ttttccattt attaagcagt acaaatgttt ttcatccatt cctaatcaaa
2401 ttctgtctgg ggacgaaggg ttggacggga tgacctccag aagtcccttc aatttctagt
2461 acctgtgact cttagccctc accacagcct tctaaattcc caaatcctag actgctcctg
2521 ggcattagca aggcagagcc tttttacctg gcctagaaag ggcaaggggt gaggatagga
2581 cagagggatt ttgttcaagt ttgctgcaac ccaagtggac gttaggccag gccttatctg
2641 aaaggccagc agctgatgct gtactaaccc agtctttctt cactctggct tcaaaaagcc
2701 acagcagagc attgtcaccg caggtgctca tgctgctccc ctaaagccag gctcaggaga
2761 agccagtgtc taggcactga gcagggatct gccccctagt tcaggtccaa attcaccttc
2821 ccctaaaccc caagcttccc aacagatcat atggtaggac cctcgagagc cttacttcaa
2881 agtgcctggg ctcagcctgg tttctgggtg ctagatccag cccaaacctg ggaaggccag
2941 ccttgtacag tctgctcctc ttgttcctga aatgtgtttc cttttcagga gatggggaat
3001 aatttccttc aggcagctga aattcaccaa gaacagcggg tacttatttc tcagctgtgc
3061 cttccctttc taagcaacca cactgcttgg cccttcaagg gtcagggtga gacgtgatgg
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3121 gctaggcctc cgttgtctgg ttgctaatga cagccttgca acccaaggtg aggtgaactc
3181 caggcatgtg tctggcccta actcctataa agtgcctcgg acagtccgca gttgtagcag
3241 aaaccaacaa gaaccactcc ttcatgtttg gaaaataatt tctcttgtat tatctccttt
3301 gaagaaggca aggctgataa tatgacaaac atcattgttt agatgaggct cagagaggta
3361 gcactctcag agtgttttga ccagtttaag ccgcagacct ggagcttcag ccaggtctga
3421 ctccaaagct gttccattac accacagcat tgtgtggaat ttgaggtcta gagagaacca
3481 ataaaagtgg taattgggaa ctgaaatcct tgagagttcc ggggagaaac ccagagatgc
3541 ctgatttcat tcctcgatgg taatacccgt cctctcggct gccaggggct ctgtggcaaa
3601 aagagtcaga catttctttg gaaaacagcg aacagcctta gagctcttgt gttcagaaga
3661 atcttcctgg cacaatgttg gagcagcagg cctctgggac ccacagaact tgtggccttt
3721 atgttctttc acccatccta ggaaccagcc aaccatcatg tgtagagccc ctactgtggg
3781 caaagtcctc ctttcattac cctacagaca gcttacagga gccagcctgc ttcccacaac
3841 tactagtgtg actccttatc tctttccacc ataccttaga gactttgata ctaccagggt
3901 ctctcaggga tggagggaag acctgaaaga gaggactggt tctgaggcca gaaaggtgtg
3961 aggagagagg aggaaaagtc ttcctaattg tgcccctaaa gagcatcctg ataccattct
4021 attctccaga catggagggg atgataaagg aaataggatc tccactggac ccttgattca
4081 ttctgaaccc tccaaaggaa ctctagaggg cgagggatga tgagggaagc aataggtagc
4141 tggggagccc tattgctgct aagtcattgg caaagtgaca aagcaattta ctgatgagag
4201 aatgtggaaa tagatgtgca gtttggaatt atgttggtgt gaatttgcca gaggaccaat
4261 gcttgcatgg agaatgggac gaggacattt gtgggcaagc agatgacaga ggtttgaagg
4321 agaatggcat ggcaggagtc tctgccagtt acttgggctt caacagccaa gctggcacaa
4381 aagacagctg gcggaggctg ctcggctact ggttacctgg agaagtagta tttgcctatt
4441 tccccccttca tccatcctga gccaaatttc ttttgctgaa caggaaagag ctaggaaccc
4501 tggaggtaaa caaagacttt gatccatgta tgagtgtatg tgtttatgta acttcctgtg
4561 gatgcaaata gattcagaga aatttagagc taaaaaggcc cttagaggga atctagccca
4621 acctacattc caccctgtta cttatgtaga aactgaggcc cagagaggga agatgacctg
4681 ccccaagtgg tgagcaagca ccaacctcca gactcagcag agtgaggggg taaagcagtt
4741 cctgtcccac atggccatct tctttcttcc acccacaaac tccaggctgg aagtacttgg
4801 ccccccttcag gagcctggcc aggcagggag agagtagctg cagccttcat cagaactctt
4861 cctcctccca aggcattctc ccagctctag cctctggact ggaaagcaca agactggccc
4921 agtgccagca agtccttagg ctactgtaat gctgcctcag gacccatccc tgcctggagg
4981 ctcctctagg ccctgtgagc acaaagaaga aagctgattt ttgtctttta atccatttca
5041 ggactctctc caggagggct cggggtgtgt catttctata ttcctccagc tgggattggg
5101 gggtgggctt tgttgtgaga atggcctgga gcaggcccaa tgctgctttt gggggtcagc
5161 atccagtgtg agatactgtg tatataaact atatataatg tatataaact gggatgtaag
5221 tttgtgtaaa ttaatggttt attctttgca aataaaacgc tttccccgtc tgttcttgaa
5281 aaaaaaaaaa aaaaaa
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The gray highlight ( ) represents exon1, the yellow ( ) represents exon3, the green ( ) represents exon4, the turquoise ( ) represents exon5, the pink ( ) represents exon6, the blue ( ) represents exon7, the red ( ) represents exon8 and the purple ( ) represents exon9.

APPENDIX C

Amino acid sequences of EDA

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  1  MGYPEVERRE LLPAAAPRER GSQGCGCGGA PARAGEGNSC LLFLGFFGLS LALHLLTLCC
 61  YLELRSELRR ERGAESRLGG SGTPGTSGTL SSLGGLDPDS PITSHLGQPS PKQQPLEPGE
121  AALHSDSQDG HQMALLNFFF PDEKPYSEEE SRRVRRNKRS KSNEGADGPV KNKKKGKKAG
181  PPGPNGPPGP PGPPGPQGPP GIPGIPGIPG TTVMGPPGPP GPPGPQGPPG LQGPSGAADK
241  AGTRENQPAV VHLQGQGSAI QVKNDLSGGV LNDWSRITMN PKVFKLHPRS GELEVLVDGT
301  YFIYSQVEVY YINFTDFASY EVVVDEKPFL QCTRSIETGK TNYNTCYTAG VCLLKARQKI
361  AVKMVHADIS INMSKHTTFF GAIRLGEAPA S
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APPENDIX D

Coding sequences and their corresponding amino acid sequences of EDA

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  1 attccctcgg cgggccgagc ctcccctctc tcccgcccct cctcctccct ttcccacccc
 61 tcggagtaga gctgcacatg cggctgctcc ctgctccgtc ccgcccagcc actgtcgcgc
121 aggaacgggt ccctgcagcc cccagccgat ggcaggacag tagccgcctg tcagaggtcg
181 tgaacggctg aggcagacgc agcggctccc gggcctcaag agagtgggtg tctccggagg
241 cc
243 atg ggc tac ccg gag gtg gag cgc agg gaa ctc ctg cct gca gca gcg
     M   G   Y   P   E   V   E   R   R   E   L   L   P   A   A   A  16
291 ccg cgg gag cga ggg agc cag ggc tgc ggg tgt ggc ggg gcc cct gcc
     P   R   E   R   G   S   Q   G   C   G   C   G   G   A   P   A  32
339 cgg gcg ggc gaa ggg aac agc tgc ctg ctc ttc ctg ggt ttc ttt ggc
     R   A   G   E   G   N   S   C   L   L   F   L   G   F   F   G  48
387 ctc tcg ctg gcc ctc cac ctg ctg acg ttg tgc tgc tac cta gag ttg
     L   S   L   A   L   H   L   L   T   L   C   C   Y   L   E   L  64
435 cgc tcg gag ttg cgg cgg gaa cgt gga gcc gag tcc cgc ctt ggc ggc
     R   S   E   L   R   R   E   R   G   A   E   S   R   L   G   G  80
483 tcg ggc acc cct ggc acc tct ggc acc cta agc agc ctc ggt ggc ctc
     S   G   T   P   G   T   S   G   T   L   S   S   L   G   G   L  96
531 gac cct gac agc ccc atc acc agt cac ctt ggg cag ccg tca cct aag
     D   P   D   S   P   I   T   S   H   L   G   Q   P   S   P   K 112
579 cag cag cca ttg gaa ccg gga gaa gcc gca ctc cac tct gac tcc cag
     Q   Q   P   L   E   P   G   E   A   A   L   H   S   D   S   Q 128
627 gac ggg cac cag atg gcc cta ttg aat ttc ttc ttc cct gat gaa aag
     D   G   H   Q   M   A   L   L   N   F   F   F   P   D   E   K 144
675 cca tac tct gaa gaa gaa agt agg cgt gtt cgc cgc aat aaa aga agc
     P   Y   S   E   E   E   S   R   R   V   R   R   N   K   R   S 160
723 aaa agc aat gaa gga gca gat ggc cca gtt aaa aac aag aaa aag gga
     K   S   N   E   G   A   D   G   P   V   K   N   K   K   K   G 176
771 aag aaa gca gga cct cct gga ccc aat ggc cct cca gga ccc cca gga
     K   K   A   G   P   P   G   P   N   G   P   P   G   P   P   G 192
819 cct cca gga ccc cag gga ccc cca gga att cca ggg att cct gga att
     P   P   G   P   Q   G   P   P   G   I   P   G   I   P   G   I 208
867 cca gga aca act gtt atg gga cca cct ggt cct cca ggt cct cct ggt
     P   G   T   T   V   M   G   P   P   G   P   P   G   P   P   G 224
915 cct caa gga ccc cct ggc ctc cag gga cct tct ggt gct gct gat aaa
     P   Q   G   P   P   G   L   Q   G   P   S   G   A   A   D   K 240
963 gct gga act cga gaa aac cag cca gct gtg gtg cat cta cag ggc caa
     A   G   T   R   E   N   Q   P   A   V   V   H   L   Q   G   Q 256
1011 ggg tca gca att caa gtc aag aat gat ctt tca ggt gga gtg ctc aat
     G   S   A   I   Q   V   K   N   D   L   S   G   G   V   L   N 272
1059 gac tgg tct cgc atc act atg aac ccc aag gtg ttt aag cta cat ccc
     D   W   S   R   I   T   M   N   P   K   V   F   K   L   H   P 288
1107 cgc agc ggg gag ctg gag gta ctg gtg gac ggc acc tac ttc atc tat
     R   S   G   E   L   E   V   L   V   D   G   T   Y   F   I   Y 304
1155 agt cag gta gaa gta tac tac atc aac ttc act gac ttt gcc agc tat
     S   Q   V   E   V   Y   Y   I   N   F   T   D   F   A   S   Y 320
1203 gag gtg gtg gtg gat gag aag ccc ttc ctg cag tgc aca cgc agc atc
     E   V   V   V   D   E   K   P   F   L   Q   C   T   R   S   I 336
1251 gag acg ggc aag acc aac tac aac act tgc tat acc gca ggc gtc tgc
     E   T   G   K   T   N   Y   N   T   C   Y   T   A   G   V   C 352
1299 ctc ctc aag gcc cgg cag aag atc gcc gtc aag atg gtg cac gct gac
     L   L   K   A   R   Q   K   I   A   V   K   M   V   H   A   D 368
1347 atc tcc atc aac atg agc aag cac acc acg ttc ttt ggg gcc atc agg
     I   S   I   N   M   S   K   H   T   T   F   F   G   A   I   R 384
1395 ctg ggt gaa gcc cct gca tcc tag attccccccatt ttgcct..........
     L   G   E   A   P   A   S
5281 aaaaaaaaaa aaaaaa
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CURRICULUM VITAE

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