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APPENDICES

APPENDIX A

List of patients in this study

Table A.1 Craniofacial genetics laboratory (CGL) DNA number and phenotype of all patients in this study.

CGL DNA number	Phenotypes	
022	Hypodontia of 31, 41 and microdontia of 12, 22	
024	Hypodontia of 13, 23	
029	Hypodontia of all premolars	
039	Hypodontia of ten permanent teeth	
169	Hypodontia of six permanent teeth	
193	Hypodontia of 32, 42	
194	Hypodontia of 25, 35, 45	
195	Hypodontia of 42	
206	Hypodontia of 13, 23	
208	Hypodontia of 41	
210	Hypodontia of 12, 22	
211	Hypodontia of 32, 42	

CGL DNA number	Phenotypes	
212	Hypodontia of 17, 27	
222	Hypodontia of 32	
228	Hypodontia of 18, 28, 38, 48	
231	Hypodontia of 72, 82	
233	Hypodontia of 32, 42	
330	Hypodontia of 14	
331	Hypodontia of 36	
332	Hypodontia of 35, 45	
335	Hypodontia of 14, 15, 16, 34, 35, 44, 45, 17, 27, 37, 47	
444	Hypodontia of 13, 23	
460	Hypodontia of 18, 28, 38	
462	Hypodontia of 32, 12, 22, 14, 15, 24, 25, 34, 35, 44, 45	
464	Hypodontia of 15, 25, 45	
467	Hypodontia of 12	
474	Hypodontia of 32, 42, 72, 82	
475	Hypodontia of 24, 25, 35, 45	
476	Hypodontia of 12, 22	
495	Hypodontia of nine permanent teeth	
180	Cleft lip and palate	
187	Cleft lip and palate	

CGL DNA number	Phenotypes	
189	Cleft palate	
197	Cleft lip and palate	
198	Cleft lip and palate Hypodontia of 12	Proband (Family II)
199	Cleft lip and palate	
200	Cleft lip and palate	
219	Cleft lip and palate Hypodontia of 12, 22, 25, 48 Polydactyly	Proband (Family I)
254	Cleft lip and palate	
258	Cleft palate	
267	Cleft lip and palate	
269	Cleft lip and palate	
284	Cleft lip and palate	
286	Cleft lip and palate	
288	Cleft lip and palate	
289	Cleft lip and palate	
296	Cleft lip	
298	Cleft lip and palate	
301	Cleft lip	
305	Cleft lip and palate	
306	Cleft lip and palate	

CGL DNA number	Phenotypes	
307	Cleft lip and palate	
506	Cleft lip and palate	
529	Cleft lip and palate	
564	Cleft lip and palate	
575	Cleft lip and palate	
576	Cleft lip and palate	
578	Cleft lip and palate	
608	Cleft lip and palate	
610	Cleft lip and palate	
611	Normal	Mother of 610
746	Hypodontia of 42	Father of 219 (Family I)
747	Hypodontia of 18, 28, 38, 48	Mother of 219 (Family I)
767	Unclear dental history	Grandfather of 219 (Family I)
768	Unclear dental history	Grandmother of 219 (Family I)
769	Hypodontia of 38, 48 Microdontia of 22	Brother of 219 (Family I)
775	Normal	Father of 198 (Family II)
776	Microdontia of 12	Mother of 198 (Family II)
777	Hypodontia of 48	Sister of 198 (Family II)

APPENDIX B

Coding sequence of *MSXI* (Exon 1 and 2)

1 tctagtcgcc agaggaaagt ttcccgggca ccccctctcc tcccctgcc tccgccgct
61 'gggccctgcc ctgcgtgccc ccaggcccag cgcgcctccg ggcgagtcce caggagcgcg
121 gcccaatgga tcgctccggg ccgccccct cgcgcgctga ttggccgccg ccccgctggc
181 ctgccttat tagcaagttc tctggggagc cgcggtaggg cccggagccg gcgagtgtc
241 ccgggaactc tgctgcgcg gcggcagcga ccggaggcca ggcccagcac gccggagctg
301 gcctgctggg gaggggcggg aggcgcgcgc gggagggtec gcccgccag ggccccggc
361 gctcgcagag gccggccgcg ctcccagccc gcccgagcc catgcccggc ggctggccag
421 tgctcggca gaagggggg cccgctctg catggcccc gctgctgaca tgacttcttt
481 gccactcggg gtcaaagtgg aggaactcgc cttcggcaag ccggcggggg gaggcgggg
541 ccaggcccc agcgcgcgcg cggccaaggc agccgccatg ggcgaggacg aggagggggc
601 caagcccaaa gtgtcccctt cgtcctgccc cttcagcgtg gaggcgctca tggccgacca
Exon 661 caggaagccg ggggccaagg agagcgcctt ggcgcctcc gagggcgtgc aggcggcggg
1 721 tggctcggcg cagccactgg gcgtcccgc ggggtcgtg ggagccccg acgcgccctc
781 ttcgccgcg cgcctcggc atttctcgtt ggggggactc ctcaagctgc cagaagatgc
841 gctcgtcaaa gccgagagcc ccgagaagcc cgagaggacc ccgtggatgc agagccccg
901 cttctccccg ccgccggcca gtgagtagcc agaaccagc gcgagaggga gggggccggg
961 tgggggcccg gtgggtgtg ggaccgcagg gctcctggtg gcctccggcg cctcgtacc
1021 tgcagccggt gctagggagc cgtgggctgc aaggccgggt cttgcgcctc cctccactcc
1081 caccagga gaaggttcca gacctctcg cttggccca gagacgctgc ggggtgggagt
1141 taacgatag gacaccgatg tctgggcacc ctgtcctct gccccacca aacgacctca
1201 ggggtccatg atccctcctc tgatcccaaa ctctgtttca tgggttcac cccagcggat
1261 gaatgtgtgt ggtgcggtat cttccctgca cccggagttt cactttctcg cagtaggagc
1321 tgggtgcccc cagccccctt tccctttcaa gtacctctt gcctagaggt tccgaagctc
1381 ctacagaatt ctacctccc atgcccttg agtttgaggc agatagttgg tgctttggg
1441 ggatggatga ttcaggggtg gggacattca ggttccagtg gagggggcgg ggcaccaagt

1501 caattagggg aaggcgcccc cgctaatacct atgggaagct cccaaacgtc taggactgag
 1561 ccattaaagt ggactccagg tgcccaaggc ggttcgctcc aaggcctcac ggccccctgg
 1621 ctgctctact cagagaacac gctcggagat atttcaggag cacgggaaat tcccaagttt
 1681 tcctcgtttc ctccgattat tttgctcggc ataatagcag ccagatttca atggcgtgat
 1741 gctgaggaat gatTTTTatc tggggattaa acgtctttga aaggccagtc cctccctaag
 1801 cctaattggcc ggagaagggtg gccccgctct gggttgtcgc cgctgaaggg agtgacgttt
 1861 ctctcggcgc ccgcccctcg ggcgccccgg cggaaagcta gttgggggcc aagcgcttcc
 1921 cggactcccc gtggcctcca gcaggaaga agcgggggtg taacacgaga tttcgtttga
 1981 ctcacatcct ggtggctga aagtccaaag gatcgttggt tttctttgt tttgtttgt
 2041 tttttctggt tgtttggtg tgttttttag agaggtgga aaaaatgcat acttaggcaa
 2101 aaccgcgctg gtgaaacatc ttcgatttga attcactttc tgccgggaaa gctgctgcat
 2161 aggcaaagtg tcctttccaa cgcttagggc cttgggcccc aagaccccga agtcaaagcg
 2221 atccccgctg tgttgggata atttgttcca cttttatcc gggggcagtc cccagcagac
 2281 cccatccccg acctgcacta gtctcgcct ctgatgcttc ttcactgtcc acccttgagg
 2341 tttattttga agccaaaaga aaaagacagc tgggcatggt gatgtctgct gactatgcca
 2401 caggttgagg ggagaggcga tctcaact cccccgcaa caacatcaac acacacacac
 2461 acacacaaac gtttgagtgg ggccagagg cctggcgc aggggtgaac gcgatccaac
 2521 agaggactga gacaatctaa agaaaaagcc cattagaata aagcagcccc tcgttctcct
 2581 ctccagatga cactttctgt ttctaagagg gctggccaca gtgcaccctc catgatggtc
 2641 tgcgctgctc catctctggt ctgcggaac tactcctaga atcccgtagg agcgaagtgt
 2701 tccggggaaa gtgtagaatt tgatttggat tctatgccac aaaactgcct agccccacac
 2761 tgaagcactc cgtgggcact gataaatggt tggccaacgc gtaaaactaa atgtgcctt
 2821 gggctgggag cagggcctct ttctgcatgt tcgtcaactg tattaacatc cacctttcct
 2881 ctggatggcc ctggaggag gcccgccatg aaggccttcc taagccgccc ggcagcacia
 2941 agtgatttc acatcttccc agctgtttag gcctaagatg tggacatcga gccttcaacg
 3001 tgggtatfff tctcctggaa tcttagtttc ttcatttgca aaaagtagac aggaacttct
 3061 cccctgcggg gttgcaatgg gaattggaga aaatatattt caagtgcctt gcgcatgcc
 3121 cggcaccgag gcaactggcg gcaactcaata tctggtattg tttggctatt attactactt
 3181 cttgggctga tcatgctcca atgcttctct ctttaaccct tgcttttttt ttctttcggc
 3241 cctcagggcg gctgagcccc ccagcctgca cctccgcaa acacaagacg aaccgtaagc
 3301 cgcggacgcc cttcaccacc gcgcagctgc tggcgctgga gcgcaagttc cgccagaagc
 3361 agtacctgtc catcgcgag cgcgaggagt tctccagctc gctcagcctc actgagacgc

3421 aggtgaagat atggttccag aaccgccgcg ccaaggcaaa gagactacaa gaggcagagc
Exon 3481 tggagaagct gaagatggcc gccaaagccca tgetgccacc ggctgccttc ggctctctct
2 3541 tccctctcgg cgccccgca gctgtagcgg ccgcccgggg tgcctcgcctc tacgggtgcct
 3601 ctggcccctt ccagcgcgcc gcgctgcctg tggcgcccgt gggactctac acggcccattg
 3661 tgggctacag catgtaccac ctgacataga gggccccagg togcccacct gtgggccagc
 3721 cgattcctcc agccctggtg ctgtaccccc ggacgtgctc cctgctcgg caccgccagc
 3781 cgccttcctt ttaacctca cactgctcca gtttcacctc tttgctcct gagttcactc
 3841 tccgaagtct gatccctgcc aaaaagtggc tggaagagtc ccttagtact cttctagcat
 3901 ttagagatct accctctcga gttaaaagat ggggaaactg agggcagaga ggttaacaga
 3961 tttatctaag gtccccagca gaattgacag tgtgaacaga gctagaggcc atgtctcctg
 4021 catagttttt cctgtcctg acaccaggca agaaagcga gagaaatcgg tgtctgacga
 4081 ttttggaatg agaacaatct caaaaaaaaa aaaaaaaaaa aaaaaaagaa aagagaaaaa
 4141 aaagactagc caccaggaag atgaatccta gcttcttcca ttggaaaatt taaagacaag
 4201 ttcaacaaca aaacatttgc tctggggggc agggaaaaca cagatgtgtt gcaaaggtag
 4261 gttgaaggga cctctctctt accagtacca gaaacacaat tgtaaaatta aaaaaaaaaa
 4321 aaactctttc tatttaacag tacatttgtg tggctctcaa acatcccttt ggaagggatt
 4381 gtgtgtacta tgtaatatac tgtatatatt aaattttatt atcatttata ttatagctat
 4441 atttgttaaa taaattaatt ttaagctaca aaaattatct ctttactgat tgagtctttt
 4501 attttaattt tgcttcttgc tatctcttta tgtgtacagt tgctttttca gtttgacggt
 4561 acttttacga cagcaacttt gaaaggatgat aacagggtat gcagggatag aggggggtgt
 4621 nctttttgaa caccacgac agatctgcat taggaaagtc aagtggagag tntgaagtaa
 4681 ttacctagga taattaggct caattttcca attcacttta agctgaaatc gacacttact
 4741 cgggcaacct ataacmwct cyttttaacc tgtctatgaa acaaagcaa atcgcccaaa
 4801 tggtttcatg agtgtggaca gctaattccag acaattcca cctccgcaaa tctgttttcc
 4861 cacgggaggc caggtggggg atgggaaggc gattggcggt gcctctctaa attaccccca
 4921 ctgtagacgc gttttgtact aattttgagc tgcagaatat aagaaccttc tcaccacctc
 4981 ttctggccgc taaaaacaca acacaacctc acaaagttta cagaaaagcc agggccacct
 5041 tccactctcg cctggaagtc ctctggagga gcctgtttc caaacctca caaactcggc
 5101 ttttttctt aagatcagtt ctccaggag aatatatttg cacacgcgcc aagtttagct
 5161 ttccggggaa ttc

The shaded parts indicate the coding regions.

APPENDIX C

MSX1 amino acid sequence

1 mtslplgvkv edsafgkpag ggagqapsaa aataaamgad eegakpkvsp slpfsveal
61 madhrkpgak esalapsegv qaaggsaqpl gvppgslgap dapssprplg hfsvggllkl
121 pedalvkaes pekpertpwm qsprfspppa rrlspactl rkhktnrkpr tpfttaqla
181 lerkfrqkqy lsiaeraefs ssslstetqv kiwfnrrak akrleaele klkmaakpml
241 ppaafglfsp lggpaavaaa agaslygasg pfqraalpva pvglytahvg ysmyhlt

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