



## APPENDICES

ลิขสิทธิ์มหาวิทยาลัยเชียงใหม่

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**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.

| <b>CGL DNA number</b> | <b>Phenotypes</b>                              |  |
|-----------------------|--|--|
| 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                           |  |

| <b>CGL DNA number</b> | <b>Phenotypes</b>  |  |
|-----------------------|--|--|
| 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                                     |  |

| <b>CGL DNA number</b> | <b>Phenotypes</b>   |                     |
|-----------------------|---|---------------------|
| 189                   | Cleft palate  |                     |
| 197                   | Cleft lip and palate  |                     |
| 198                   | Cleft lip and palate<br>Hypodontia of 12                            | Proband (Family II) |
| 199                   | Cleft lip and palate  |                     |
| 200                   | Cleft lip and palate  |                     |
| 219                   | Cleft lip and palate<br>Hypodontia of 12, 22, 25, 48<br>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  |                     |

| <b>CGL DNA number</b> | <b>Phenotypes</b>                         |                               |
|-----------------------|---|-------------------------------|
| 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<br>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 *MSX1* (Exon 1 and 2)

1 tctagtcgcc agaggaaagt ttcccgggca ccccctctcc tcccctgccc tccgccgcct  
61 gggccctgcc ctgctgccc ccaggcccag cgcgcctccg ggcgagtccc caggagcgcg  
121 gcccaatgga tcgctccggg cccgccccct cgcgcgctga ttggccgcgc ccccgctggc  
181 ctgccttat tagcaagttc tctggggagc cgcggtaggg cccggagccg gcgagtgtcc  
241 ccgggaactc tgcctgcgcg gcggcagcga ccggaggcca ggcccagcac gccggagctg  
301 gcctgctggg gaggggcggg aggcgcgcgc gggagggtcc gcccgccag ggccccgggc  
361 gctcgcagag gccggccgcg ctcccagccc gcccgagcc catgcccggc ggctggccag  
421 tgctgcgca gaagggggg cccggctctg catggccccg gctgctgaca tgacttcttt  
481 gccactcggg gtcaaagtgg aggactccgc cttcggcaag ccggcggggg gaggcgcggg  
541 ccaggcccc agcgcgcgcg cggccacggc agccgcatg ggcgcggacg aggagggggc  
601 caageccaaa gtgtcccctt cgctcctgcc cttcagcgtg gaggcgtca tggccgacca  
**Exon** 661 caggaagccg ggggccaagg agagcgcctt ggcgcctcc gagggcgtgc aggcgcgcgg  
**1** 721 tggctcggcg cagccactgg gcgtcccgcc ggggtcgtg ggagccccg acgcgcctcc  
781 ttcgccgcg cgcctcggcc atttctcggg ggggggactc ctcaagctgc cagaagatgc  
841 gctcgtcaa gccgagagcc ccgagaagcc cgagaggacc ccgtggatgc agagccccg  
901 cttctccccg ccgcccggca gtgagtagcc agaaccagg cgcagagga gggggccggg  
961 tgggggcccg gtgggggtgt ggaccagag gctcctggtg gcctccggcg cctgctacc  
1021 tgcagccggt gctaggagc cgtgggctgc aaggccgggt cttgcgcctc cctccactcc  
1081 caccagaa gaaggttcca gacctctcg cttggcca gagacgtgc ggggtggagt  
1141 taacgatag gacaccgatg tctgggacc ctgtcctcct gccccacca aacgacctca  
1201 ggggtccatg atccctcatc tgatccaaa ctctgtttca tcggettca cccagcggat  
1261 gaatgtgtgt ggtgcggtat cttccctgca cccggagttt cactttctcg cagtaggagc  
1321 tgggtgcccc cagcccctct tcctttcaa gtacctctt gcctagaggt tccgaagctc  
1381 ctacagaatt ctacctccc atgcccttg agtttgagc agatagttg tgctttgggc  
1441 ggatgatga ttcaggggtg gggacattca ggttccagt gagggggcgc ggcaccaagt

1501 caattagggg aaggcggccc cgctaactct atgggaagct cccaaacgtc taggactgag  
 1561 ccattaaagt ggactccagg tgcccaaggc ggttcgctcc aaggcctcac ggccccctgg  
 1621 ctgctctact cagagaacac gctcggagat atttcaggag cacgggaaat tcccaagttt  
 1681 tctctgtttc ctccgattat tttgctcggc ataatagcag ccagatttca atggcgtgat  
 1741 gctgaggaat gatttttatc tggggattaa acgtctttga aaggccagtc cctccctaag  
 1801 cctaattggc ggagaagggtg gccccgctct gggttgtcgc cgctgaaggg agtgacgttt  
 1861 ctctcggcgc ccgccccctg ggcggcccg cggaagcta gttgggggcc aagcgttcc  
 1921 cggactcccg gtggcctcca gcaggaaga agcgggtgt taacacgaga tttcgtttga  
 1981 ctcacatcct ggtggtctga aagtccaaag gatcgttggt ttttcttgt tttgtttgt  
 2041 ttttctgtt tgtttgggt tgtttttag agaggtgtga aaaaatgcat acttaggcaa  
 2101 aaccgcgtg gtgaaacatc ttcgattga attcacttcc tgccgggaaa gctgctgat  
 2161 aggcaaagtg tcctttccaa cgcttagggc cttgggcccc aagaccccg agtcaaagcg  
 2221 atcccgctg tggtgggata atttgtcca cattttatcc gggggcagtc cccagcagac  
 2281 cccatccccg acctgacta gtctgctct ctgatgcttc ttactgtcc acccttgagg  
 2341 tttatttga agccaaaaga aaaagacagc tgggcatggt gatgtctgct gactatgcca  
 2401 caggttgagg ggagaggcga tctcaact cccccgcaa caacatcaac acacacacac  
 2461 acacacaaac gtttgagtgg ggccagaggg cctggcgc aggggtgaa gcgatccaac  
 2521 agaggactga gacaatctaa agaaaaagcc cattagaata aagcagcccc tcgttctct  
 2581 ctccagatga cactttctgt ttctaagagg gctggccaca gtgcaccctc catgatggtc  
 2641 tgcgctgctc catctctggt ctgcgggaa tactctctaga atcccgtagg agcgaagtgt  
 2701 tccgggaaa gtgtagaatt tgatttgat tctatgccac aaaactgcct agccccacac  
 2761 tgaagcactc cgtgggact gataaatgtt tggccaacgc gtaaaactaa atgtgcctt  
 2821 gggctggcgc cagggcctct ttctgcatgt tcgtcaactg tattaacatc caccttctct  
 2881 ctggatggc ctgggaggag gcccgcatg aaggccttc taagccgccc ggcagacaaa  
 2941 aggtgatttc acatctccc agctgttag gctaagatg tggacatga gccttcaacg  
 3001 tgggtatctt tctcctggaa tcttagtttc ttcattgca aaaagtagac aggaacttct  
 3061 ccctgcggg gttgcaatgg gaattggaga aaatatatt caagtgcctt gcgcatgcc  
 3121 cggcaccgag gcaattggc gactcaata tctggtattg tttggctatt attactactt  
 3181 cttgggctga tcatgctcca atgcttctct cttaccct tgctttttt ttctttcggc  
 3241 cctcagggcg gctgagcccc ccagcctgca cctccgcaa acacaagacg aaccgtaagc  
 3301 cgcggacgcc cttaccacc gcgcagctgc tggcgctgga gcgcaagtcc gccagaagc  
 3361 agtacctgtc catgcggcag cgcgaggagt tctccagctc gctcagcctc actgagacgc

3421 aggtgaagat atggttccag aaccgccg ccaaggcaa gagactacaa gaggcagagc

**Exon** 3481 tggagaagct gaagatggcc gccaaagcca tgctgccacc ggctgccttc ggctctctct

**2** 3541 tccctctcgg cggccccgca gctgtagcgg ccgcgccggg tgcctcgtc tacggtgcct

3601 ctggccccctt ccagecgcgc gcgctgcctg tggcgcccggt gggactctac acggccccatg

3661 tgggctacag catgtaccac ctgacataga ggggccagg tcgccacct gtgggccagc

3721 cgattcctcc agccctgggtg ctgtaccccc ggacgtgctc ccctgctcgg caccgccagc

3781 cgccttccct ttaacctca cactgetcca gtttcacctc tttgctccct gagttcactc

3841 tccgaagtct gatccctgcc aaaaagtggc tggaaagatc ccttagtact cttctagcat

3901 ttagagatct accctctcga gttaaaagat ggggaaactg agggcagaga ggttaacaga

3961 tttatctaag gtccccagca gaattgacag tgtgaacaga gctagaggcc atgtctcctg

4021 catagttttt ccctgtcctg acaccaggca agaaagcga gagaaatcgg tgtctgacga

4081 ttttggaatg agaacaatct caaaaaaaaa aaaaaaaaa aaaaaaagaa aagagaaaa

4141 aaagactagc caccaggaag atgaatccta gcttcttcca ttggaaaatt taaagacaag

4201 ttcaacaaca aaacatttgc tctggggggc agggaaaaca cagatgtgtt gcaaaggtag

4261 gttgaaggga cctctctctt accagtacca gaaacacaat tgtaaaatta aaaaaaaaa

4321 aaactcttcc tatttaacag tacatttgtg tggctctcaa acatcccttt ggaagggatt

4381 gtgtgtacta tgtaataatac tgtatatttg aaattttatt atcatttata ttatagctat

4441 atttgttaaa taaattaatt ttaagctaca aaaattatct ctttactgat tgagtctttt

4501 attttaattt tgcttcttgc tatctcttta tgtgtacagt tgctttttca gtttgacggt

4561 acttttacga cagcaacttt gaaaggtgat aacaggtat gcagggatag aggggggtgt

4621 ncttttgaa caccacgac agatctgcat taggaaagtc aagtggagag tntgaagtaa

4681 ttacctagga taattaggct caattttcca attcaattta agctgaaatc gacacttact

4741 cgggcaacct ataaccmwct cyttttaacc tgtctatgaa aacaaagcaa atcgcccaaa

4801 tggtttcatg agtgtggaca gctaaccag acaattcca cctccgcaa tctgttttcc

4861 cacgggagc caggtggggg atgggaagc gattggcgtt gcctctctaa attacccca

4921 ctgtagacgc ggtttgtact aattttgagc tgcagaatat aagaacctc tcaccctca

4981 ttctggccgc taaaaacaca acacaaccta acaaagtta cagaaaagcc agggccacct

5041 tccactctcg cctggaagtc ctctggagga gccctgttc caaacctca caaactcgg

5101 ttttttctt aagatcagtt cttccaggag 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 sllpfsveal  
61 madhrkpgak esalapsegv qaaggsaqpl gvppgslgap dapssrplg hfsvvgllkl  
121 pedalvkaes pekpertpwm qsprfspppa rrlsppactl rkhktnrkpr tpfттаqlla  
181 lerkfrqky lsiaeraefs ssslstetqv kiwfqnrrak akrlqaele klkmaakpml  
241 ppaafglsfp lggpaavaaa agaslygasg pfqraalpva pvglytahvg ysmyhlt

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## CURRICULUM VITAE

|                                 |  |
|---------------------------------|--|
| <b>Name</b>                     | Ms.Onnida Wattanarat   |
| <b>Date of Birth</b>            | 24 September 1980  |
| <b>Education</b>                | Graduated High School from Montfort College in 1997<br>Graduated Doctor of Dental Surgery (D.D.S) from<br>Faculty of Dentistry, Chiang Mai University,<br>Chiang Mai in 2003 |
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