



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.

CGLDNA number	Phenotypes	
118	Cleft palate with ankyloglossia	
249	Cleft palate with ankyloglossia	
250	Cleft lip and palate with tongue tie	
251	Cleft lip	
252	Cleft palate	
253	Cleft palate with tongue tie	
254	Cleft lip and palate	Daughter of 255
255	Ankyloglossia	Mother of 254
256	Cleft lip and palate	
257	Cleft lip and palate with missing 12,22	
258	Cleft palate	Son of 259
259	Ankyloglossia	Mother of 258
260	Cleft palate with ankyloglossia	Daughter of 261
261	Ankyloglossia	Mother of 260
262	Cleft palate with ankyloglossia	Daughter of 263
263	Ankyloglossia	Mother of 262
264	Cleft lip and palate with tongue tie	Son of 265
265	Ankyloglossia	Father of 264
266	Cleft palate	
267	Cleft lip and palate	
269	Cleft lip and palate	Son of 270
270	Ankyloglossia	Father of 269
271	Cleft lip and palate with tongue tie	
272	Cleft lip and palate	
273	Cleft lip and palate	
274	Ankyloglossia	Mother of 275
275	Cleft palate and Hemifacial microsomia	Son of 274
284	Cleft lip and palate	
286	Cleft lip and palate	
287	Cleft lip	

CGLDNA number	Phenotypes	
288	Cleft lip and palate, Syndactyly	
290	Cleft palate	Daughter of 291
291	Ankyloglossia	Mother of 290
292	Cleft lip and palate	Son of 293
293	Ankyloglossia	Mother of 292
294	Cleft lip	หลาน 295
295	Ankyloglossia	Aunt of 294
298	Cleft lip and palate	Son of 299
299	Ankyloglossia	Mother of 298
300	Ankyloglossia	Mother of 316
301	Cleft lip	Son of 302
302	Ankyloglossia	Mother of 301
303	Facial cleft +BCLP	Daughter of 304
304	Ankyloglossia	Mother of 303
305	Cleft lip and palate	
306	Cleft lip and palate	
307	Cleft lip and palate	Daughter of 308
308	Ankyloglossia	Mother of 307
309	Cleft lip and palate	Son of 310
310	Ankyloglossia	Mother of 309
311	Cleft lip and palate	
312	Ankyloglossia	Grandmother of 317
314	Missing 12, 22	
316	Cleft lip and palate	Daughter of 300
317	Cleft palate	Daughter of 318
318	Ankyloglossia	Mother of 317
322	Missing 28	
421	Ankyloglossia	
422	Missing 18	
423	Missing 18, 28	
427	Missing 48	
428	Ankyloglossia	
429	Ankyloglossia	
430	Ankyloglossia and missing on 32	
506	Cleft lip and palate with Ankyloglossia	
510	Cleft lip and palate	
518	Cleft lip and palate	
524	Cleft lip and palate	
527	Incomplete cleft palate+ hypotonia	
530	Incomplete CL	
533	Cleft lip and palate	
535	Cleft palate	
538	Cleft lip and palate	

<b>CGLDNA number</b>	<b>Phenotypes</b>	
540	Bifid uvula	
544	Cleft lip and palate	
545	Cleft lip and palate	
547	Cleft lip and palate	
548	Cleft palate with ankyloglossia	
550	Cleft lip and palate	
555	Cleft lip and palate	
557	Cleft lip and palate	
560	Cleft lip and palate	
561	Cleft palate with small mandible	
564	Cleft lip and palate	
581	Cleft lip with ankyloglossia	
586	Cleft lip and palate with ankyloglossia	
605	Ankyloglossia	
608	Cleft lip and palate	
610	Cleft lip and palate	
061	Cleft lip with ankyloglossia and missing	
639	Normal	Mother of 264
643	Short frenum	Grandmother of 264
644	Normal	Grandfather of 264

## Appendix B

### Coding sequence of *TBX22* (Exon1-8)

1 cttctctgagg ctcttgagaa gagctgctgc aggcatttgc agagtgaatg agctctgact  
61 gagacttgac ttcagaacca ctgggctggc ctggcctctt gacacagcaa cacactgctt  
121 cttgccttcc aggtaaatac ctctgcctta ctgacctgtt tatgggtttt agtgggacat  
181 cagtacaggg attctoctca gcctcttctc ctctgttaa ctatcactaa ccaagtcctt  
241 aacattagga aagtccaact gataactgtc agtttcatct ttcattaact gggggttttg  
301 tgacagagat cttgcaatca ctacttctcg ggggactgga agtagaatat tagtgctctt  
361 taaaattaat gaaggctaga cttgtccagt ctgttaagag tatgctgtaa taccxaaaga  
421 agccagcaat agtccctctc ctaggcagca atgcagccag aaggctccca ggccaggat  
481 actatgcatt agtgctcaca gatcttgttt agggagctag ggagcaagg gaagggacgg  
541 gagtgcctcc tgggaaggtga ggactgaagc caacgtggaa gtcactgagc tgagtggcag  
601 gtgtctaaca ctgaaaagtg gccctagcca agggctctaa gtagtggtag tttctaccga  
661 ggctcttccc caaggagctc tagatccaca gaggcagaag gattatttga tcttctttgc  
721 actcatttga tttcaggagg aggtctgaag tcccaaagca tgggcattgg ggtgagagag  
781 ggaacatcaa agtgtgcaa aaaagattgt tttatcttac ttattctga tttttcttac  
841 cagagcttga gctcatacac cgcattgtaa cacaaacaca caagcacaca taccacacac  
901 acacacatgc cttcaaaacta gagaagggga ctacagaaag cacctggcca taaaaggcat  
961 gtcaaggagt ttgctgtagg gtggtgatat ggttgctttg tttcagttca gcagacttat  
1021 ctccatgaag accttattgt tttctgtgat ttctcttttg ttttttttaa aatttgtctt  
1081 tttttcttaa ttattaaaac cttccctttt gccctcggga gcattgatca gggtttacta  
1141 actacaaata tttacttcat gaaggagcag ttttttctct agtaataata atgtggggac  
1201 cctgggaagg aagacagtgt atgaaagaag gcatagattg gcttcaagca caggcagat  
1261 aggtgtttgc ctatatttgg tgttttgata gacttaggga aaatccctca gcctactacc  
1321 aaatattttt gagtgtcagt acatctgtgg ttgtaggta ttgaaaagcc ctgattaaat  
1381 acagataatt atgagaatat ggaagaagc ctttagtacc ttattcgcac gtcctaggaa  
1441 tgatgaggtg ggtgccaag ttaagaagag gaaaatata tgtaaatatc caggagtaag  
1501 taaatagtga aaagattagc acgagatttg gtgggcccaa tggagttcag gatgatttct  
1561 ttcttaacag aaggatctgc ttctgttggg ctcatggctg ccagaattag gcagagccag  
1621 atgatgactc actgatctga gaactgagtg gactcatttg gtcagaagc gtgtgtaggg  
1681 gaagaggtg taaagcaagc attttctag ttttaggaag ataaaaaat tctcataaa  
1741 cagtattttt tttcaataat tcattgatgc aattaacttg gcatactctc gttcaagtga  
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1861 acctgcagtc atttttcaaa tcttagccca ctttctttga ggctgatgca tatagcatat  
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1981 tggctcaagc ctgtaatccc agcacttttg gaggctgagg caggcagatc acctgaggtc  
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Exon 3	9361	agtaccatgt	ggccatcgat	gtggtgccgg	tggattccaa	acgctatagg	taatgggccc
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10801 ccagcttttg cctgctgcac ctaatgccac agcatgtgtt ttttcaggta cgtctatcac  
**Exon 4** 10861 agctcacagt ggatggtagc tgggaataca gaccatttgt gcatcattcc tagattctat  
10921 gttcaccggg actcaacctg ctcgggagag acctggatgc ggcagatcat cagctttgat  
10981 cgcataaaac tcaccaacaa tgagatggat gacaaaggcc acgtactgta gcacaatcct  
11041 ttaccccagag gagggagggt ccaaggctcc tgcccactgg tcaccctgga gaggccaggg  
11101 aggcctttttg cagatgcaca tgttgtgttt ttttaggaact gttcaagccc tgcacctgta  
11161 agtaaagtag ctaacctagg gctcctgaaa cgtttgttta gcatctgaga tgctggcgac  
11221 tggggatggt gaatataggg agtttgagga cagaaggaga cctgtttaag ccagaatgcc  
11281 tgggtctggga gagtaggagg gaaccattca gggagttctc aggcacctta gcctccttgg  
11341 caatcactat gatctctgat tgcctcctg tagtccacgc agttggatg gcaccagtg  
11401 tatggtatgt gtgatggaag gatacaagtg ggggaatctc agactgcatc ctggaaatct  
11461 cccaaacaac ctaccctcat aggtcctatt gatcttgac attctacta ggtgaccccc  
11521 agcttctgtc ctccccagtc tggcactgtg ccagctacaa aaggaaagag ggaagctccc  
11581 aagattagga ggtagggtat gtcaggttac ccaccatata caagcctatc agttactgcc  
11641 atgggaactt gtagcaggct agcagatccc agtgccactt ctacaccaga tggctgccag  
11701 tgtttgaagc aggaaaaggc cccaatctgg aatggaatc actgctgact ggtgcogctg  
11761 aggtgtcttt ctctggaat gtttttagct cggagcgctt cgtttggagg ccaaatggca  
11821 caacaaagta gttacctaag ctctcatcatt gcctttttgt gtgcatgag tggagggtgt  
11881 caggagaggg aactagggtt tgggactgaa gccagttttt ctaacagcat tgatcatttc  
11941 tctccagat cattctgcaa tccatgcata agtacaacc cagagtgcac gtgatagagc  
**Exon 5** 12001 aaggcagcag tgttgacctg tcccagattc agtccttggc cactgaaggt gttaaaacat  
12061 tctcctttta agaaactgag ttcaccacag taacggctta ccaaaaccaa caggtaaact  
12121 tggatcatgct tcaggcgaat ggaatggctt ccagaggagc cttggattag aggcattagag  
12181 gctaactgcc atcaattgca ttttccacaa ttctaataa aatcatagat tttattgtat  
12241 tgtcatccat gcaagtttga taatgaattg tataaatctt atgggagaag gcaactccag  
12301 aaggtaattt ataaaagtaa tgatttataa ataggagcta accattctaa atatgacttg  
12361 caatatggtt acatttattt aatgtgcat ataacgtgt tcaacatct ctctgggtgg  
12421 ttgtaaccac tataagcaat ggcaacagtg ttcttactga aagcaatgac tttatctttc  
12481 tctctctatt gaatccatag aatagcaaa taaaaataga aagaaatcct tttgctaagg  
**Exon 6** 12541 gatattagaga tactggaaga aacagtaag cagcatagca atgatctcta atattgatgt  
12601 agctagctat tttcaaaagt tttttttttt ttttttttcc tgagatggag totcactctg  
12661 ttgcccaggc tggagtgcaa tggcacgata tcagctcact gcaactttca cctccctggg  
12721 tcaagcaatt ctctgcctc agcctcccaa gtagctagga ttacaggcat gtgccaacag  
12781 gcctggctaa tttttgtatt ttttagtagag atggggtttc accatattgg tcaggctggg  
12841 cctgaaacacc tgacctcgtg atctgcccgc ctgagcctcc caaagtctg ggattacagg  
12901 cgtgagccac tgcacctggg cccccacaag ttttaaggat aagaaagctg actcagagac  
12961 tcaaaagctc taagttttgt aatagtagg tggtagaaga ggaaccaga tgttttgggt  
13021 tgaaaacatt acttctctcc actcctttgc tactcactat taagacctg cttattttag  
13081 aattctaggt ttgactgaat tttattcaga agtctagcta tataaaagag aatatattaa  
13141 aatagttaca tctgaaagat atagcaaat aactcaaat cattattttc agaaattgca  
13201 ttctggggat gctgaaagtt gactctcttt ttttaggggtg tattggatgg gcttttagag  
**Exon 7** 13261 acctaccat ggaggccttc tttcactctc gatttttaaaa cctttggcgc agacacacaa  
13321 agtaagaaaa cttggaacgt ttgttttatt tttacatac aattaaataa caacatccgg  
13381 aacccttgta ccaataacta cacaagggga tatgtacatg caagaaaatg tctacttatg  
13441 ggtaactgcg tgtacatttg cctgaatgac gaggtaagag gcataaaggg agagaagctg  
13501 gcatgaagga agagaataga aatgtgataa ttgtagactt attgacatat gttataaagt  
13561 tatcttacac cctgtgattt tcagatttca tctggatttt tcagcctctc actgtttcac  
13621 attcttttag atatgctgtt ggtgcatttg cagcctgatt tgcaaatatg ttactattga  
13681 aatgaatagt ttatctttta tgaggacctt tcagtacatg ttaaaatgac tttcattaaa  
13741 ttcactactg agctctaatt ttctctttt ttggtcatgg ggaaagttaa tgttaaaaat  
13801 aatactaagt ctccacaatt taattgccac aagcatttaa aatatattgc aagatattta  
13861 tctcagatg gtacattaca tgacaaaata ataataatta aataatgcag gctttctggg  
13921 ggaaggatac cttaatttgc tctgatagtt aataataaat agaataattt ttatttgaaa  
13981 atgacatgga ttttttttat ccctttgtct ttactccatc gaagttgctt gcttttgacc  
14041 attaattcaa ggttacaatt gccaaagcaa accaacatta tagttaggtt gagttttaca  
14101 tgatgccata acaactaact gaaaagggaa gaagtgaagt ctaactattt ggtgaaatat  
14161 ttcactggaa tggctgtctt gatgtgggtt agggaaattgc atgaaaaaaa aaacacttat  
14221 tttacagtgt cgatttaact aatagcatct agaaagcata ctttttaata tccaagtcaa  
14281 acaaatgtg cactaataaa gaaaaagtta aagatgatca tgaaatgata atttaaagta  
14341 cacagttttc ttaattattt aacatatcca aatgctctc aaattgcaaa taaatgagct  
14401 actagtttct taaataactt ttaaatgctt tattataaaa gatatttgcc atcttgggtg  
14461 tgaaggttta ggatacgtct acatacacia aatagttaag aaaacagcaa cactaaaaaa  
14521 gaaacaactt ggtcaaatcc agccaataac caattgtttg aattcagaca tttacaaaaa



14581 taagcttttag agtttagcgt ttcttgacc ttataattat cctagtttt agtagatgct  
 14641 aggcaaacat aaaaaatcag caaacgcat tgatatgac acttacattc atccttatat  
 14701 ttcttaactt gcaaaaattat ttccaactct taagatataa tgcagtatgg aaattgcccc  
 14761 attcattggt ttgtataag aaaaaagct aaggctttta aatatattgc tttcatttat  
 14821 acaagactat ttcatgttc actaattttt taaatcagtg gtcagaggaa ttttttaaat  
 14881 cagtgtacct ttggtcagag gaatttaaaa aatcaacgta catgaaatct atacattcta  
 14941 tctataaact aaatctatat aatcccaaac aaccaatcaa tttgtgcatc ttaactactt  
 15001 attttagtaa ctttttaagg ttttaaaata ataaaactag tttgtgtttg taagatataa  
 15061 ttgacgctaa gtcataattg gacttctttg taattctttg tacaaatgtg gttcaagtgc  
 15121 ctagcacagt atttgagcaa atgaatgctt aatacatgat acatggtagt agtgctaaaa  
 15181 gtaatagtgg tagtagtata gatcagtggg cccaacctt tttggtacca ggggccagtt  
 15241 ttgtgaaaga caatttttcc acgaatggtg ggggttgga ggggatggtt tcgggatgaa  
 15301 actgtttccac ctccagatcat caggcattag attatcatta aagggcactc aacctagatc  
 15361 cctcacatgc acagttcaca atagagtgtg cgctcctgtg agaatctaact gctgcgctg  
 15421 atctgaaagg aggcagagct caggcaatga ggtaatgctc actggcctgc tgatcacctc  
 15481 ttgctgtcca gcttgattca taacaggcta cagacctgta ccagtgggta acccaggcat  
 15541 tagcgatacc tagtatagat actatattca acttttagtca gaggaatgag caggtcctgt  
 15601 ctcaaagaag ctaaaagaac tgtgaactca caaggaaaat taatctaagg atgaagcaca  
 15661 gatagtgaag gatatgatta ctaatattgc aggaacatca aatgtcaagt tcattattca  
 15721 ttggctgaat tgtcattcac aggtggaagc agtggctcat ctccagtgac ctctagtgga  
 15781 ggggccccct ctcttttgaa ctcttactt tctccacttt gcttttcacc tatgtttcat  
 15841 ttacctacaa gctcccttgg aatgcctgt ccagaggcat acctgcccaa tgtcaacctg  
 15901 cctctatgct acaagatttg tccaactaat ttttggaac agcaacctct tgttttaccg  
 15961 gtcctgaaa gactagcaag cagcaacagt tctcagtct tagccccact catgatggaa  
 16021 gtgcctatgt tatcttccct ggggtcacc aattcaaaaa gcggttcac tgaagactcc  
 16081 agtgatcagt atctacaagc acctaatctt accaatcaaa tgttatatgg attacagtca  
 16141 cctggaaata tttttctgcc aaactccatc accccagaag cacttagttg ctctttcat  
 16201 ccttccatg acttttatag atacaatttc tctatgcoat ctagactgat aagtggttcc  
 16261 aaccatctta aagtgaatga cgacagtcaa gtttcttttg gagaaggcaa atgtaatcat  
 16321 gttcattggt atccagcaat taaccattac ctttagtaag acaatagcat ttctagaaca  
 16381 attacatgta aacaaatatt ttctttattt gtagccaaag aaatttcaac agttattggg  
 16441 cttaaaaagc atcattacaa tacagtattt ctttgttata catttaaaga tttaaagtgc  
 16501 cttatcaaat aatattcatg aagagttggt tataatgtca aatgaaacct acaggaatct  
 16561 ctgattacag tggccttgag cttcaaatg agatagcaa taaatattat ttgatgatac  
 16621 tccaccagtg aaaattgatg ctaagtgatg ggattttcaa ttatactgaa gctagttcac  
 16681 cacgttaact gcattttaca cattgacaat gacaaaaaga agatggatgt aattctcatg  
 16741 aaagcagtga agcaatttca gttttaaaa tgaagatcgg ctttcatgta attatctagt  
 16801 agttgtagaa gaaaatttaa ttattggtt gcctcatgcc tttatacttt gctggtgaag  
 16861 aaactactaa tctcaattta agatacaaat aaggacaaa actttcaagt attatatttt  
 16921 atttatcttt gtagcctaaa gaccatttaa tcttgaagga acacaaagat caaatgaaaa  
 16981 gtaaaacact ctaataaat ttgcttttat tttc

Exon 8

The shaded parts indicate the coding regions.

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## Appendix C

### *TBX22* amino acid sequence

```
1  atg gct ctg agc tct cgg gcg cgt gcc ttc tcc gtg gaa gcc ttg
   M  A  L  S  S  R  A  R  A  F  S  V  E  A  L
16  gtg ggg aga ccc agc aaa aga aaa ctc caa gac cca ata cag gcg
   V  G  R  P  S  K  R  K  L  Q  D  P  I  Q  A
31  gag cag cct gag ctg cgg gag aaa aag ggc gga gag gaa gag gag
   E  Q  P  E  L  R  E  K  K  G  G  E  E  E  E
46  gag aga agg agc agc gct gca ggg aag agc gag ccg ctt gaa aaa
   E  R  R  S  S  A  A  G  K  S  E  P  L  E  K
61  caa cct aag aca gag ccc tca aca tct gct tcc tct ggc tgc ggc
   Q  P  K  T  E  P  S  T  S  A  S  S  G  C  G
76  agc gac agc ggc tac ggc aac agc tct gaa agt ctg gaa gag aaa
   S  D  S  G  Y  G  N  S  S  E  S  L  E  E  K
91  gat att caa atg gag ctt caa gga tct gaa ctg tgg aaa aga ttc
   D  I  Q  M  E  L  Q  G  S  E  L  W  K  R  F
106 cat gac atc ggg act gag atg atc att act aaa gcg ggc agg cgg
   H  D  I  G  T  E  M  I  I  T  K  A  G  R  R
121 atg ttc ccc tct gtt cgg gtc aag gtg aaa ggg ttg gat cca ggg
   M  F  P  S  V  R  V  K  V  K  G  L  D  P  G
136 aag cag tac cat gtg gcc atc gat gtg gtg ccg gtg gat tcc aaa
   K  Q  Y  H  V  A  I  D  V  V  P  V  D  S  K
151 cgc tat agg tac gtc tat cac agc tca cag tgg atg gta gct ggg
   R  Y  R  Y  V  Y  H  S  S  Q  W  M  V  A  G
166 aat aca gac cat ttg tgc atc att cct aga ttc tat gtt cac ccg
   N  T  D  H  L  C  I  I  P  R  F  Y  V  H  P
181 gac tca ccc tgc tgc gga gag acc tgg atg cgg cag atc atc agc
   D  S  P  C  S  G  E  T  W  M  R  Q  I  I  S
196 ttt gat cgc atg aaa ctc acc aac aat gag atg gat gac aaa ggc
   F  D  R  M  K  L  T  N  N  E  M  D  D  K  G
211 cac atc att ctg caa tcc atg cat aag tac aaa ccc cga gtg cac
   H  I  I  L  Q  S  M  H  K  Y  K  P  R  V  H
```

226 gtg ata gag caa ggc agc agt gtt gac ctg tcc cag att cag tcc  
       V I E Q G S S V D L S Q I Q S  
 241 ttg ccc act gaa ggt gtt aaa aca ttc tcc ttt aaa gaa act gag  
       L P T E G V K T F S F K E T E  
 256 ttc acc aca gta acg gct tac caa aac caa cag att acg aaa cta  
       F T T V T A Y Q N Q Q I T K L  
 271 aaa ata gaa aga aat cct ttt gct aaa gga ttt aga gat act gga  
       K I E R N P F A K G F R D T G  
 286 aga aac agg ggt gta ttg gat ggg ctt tta gag acc tac cca tgg  
       R N R G V L D G L L E T Y P W  
 301 agg cct tct ttc act ctc gat ttt aaa acc ttt ggc gca gac aca  
       R P S F T L D F K T F G A D T  
 316 caa agt gga agc agt ggc tca tct cca gtg acc tct agt gga ggg  
       Q S G S S G S S P V T S S G G  
 331 gcc ccc tct cct ttg aac tcc tta ctt tct cca ctt tgc ttt tca  
       A P S P L N S L L S P L C F S  
 346 cct atg ttt cat tta cct aca agc tcc ctt gga atg ccc tgt cca  
       P M F H L P T S S L G M P C P  
 361 gag gca tac ctg ccc aat gtc aac ctg cct cta tgc tac aag att  
       E A Y L P N V N L P L C Y K I  
 376 tgt cca act aat ttt tgg caa cag caa cct ctt gtt tta ccg gct  
       C P T N F W Q Q Q P L V L P A  
 391 cct gaa aga cta gca agc agc aac agt tct cag tct tta gcc cca  
       P E R L A S S N S S Q S L A P  
 406 ctc atg atg gaa gtg cct atg tta tct tcc ctg ggg gtc acc aat  
       L M M E V P M L S S L G V T N  
 421 tca aaa agc ggt tca tct gaa gac tcc agt gat cag tat cta caa  
       S K S G S E D S S D Q Y L Q  
 436 gca cct aat tct acc aat caa atg tta tat gga tta cag tca cct  
       A P N S T N Q M L Y G L Q S P  
 451 gga aat att ttt ctg cca aac tcc atc acc cca gaa gca ctt agt  
       G N I F L P N S I T P E A L S  
 466 tgc tcc ttt cat cct tcc tat gac ttt tat aga tac aat ttc tct  
       C S F H P S Y D F Y R Y N F S  
 481 atg cca tct aga ctg ata agt ggt tcc aac cat ctt aaa gtg aat  
       M P S R L I S G S N H L K V N  
 496 gac gac agt caa gtt tct ttt gga gaa ggc aaa tgt aat cat gtt  
       D D S Q V S F G E G K C N H V

511 cat tgg tat cca gca att aac cat tac ctt tag  
H W Y P A I N H Y L STOP



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