

# Sanger sequencing results

*Maurits Evers*

*Version 1.2 (29 March 2018)*

## Primer information

V1

QJL

1

2

3

4

E23

R

R

R

R

R

E27

F

F

F

F

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E33

F

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F

R

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E39

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F

F

F

F

```
## use default substitution matrix
## use default substitution matrix
## use default substitution matrix
## use default substitution matrix
```

```
## Warning in msaPrettyPrint(res.msa[[1]], output = "asis"): Cannot use
## default file name 'res.msa[[1]].asis' because it would contain invalid
## characters => resorting to 'msaPrettyPrintOutput.asis'!
```

	TAAAC	TTTCATG	CACACGTGTGCGAGT	CAGTCCATACCCAACAACACCTT	TCTTGC	
Father	.	.	.	.	.	0
Mother	.	.	.	.	.	0
Sister	TAAACTTTTCATG	CACACGTGTGCGAGT	CAGTCCATACCCAACAACACCTTTTCTTGCCGC			60
Brother	.	.	.	.	.	0
Proband	.	.	.	.	.TCCTGCACTG	10
RefSeq	.	.	.	.	.	0
consensus	.	.	.	.	.	

Father	.....CTTTGT.TGCCTTCTACAAA	20
Mother	.....	0
Sister	CTTTAGAGAGGGAGGGAGGGTTCCTGCAAGCAGTCTTTTCGGTATGTCTCACTCCAAA	120
Brother	.....	0
Proband	CAAAGAGCAATGGGAGAGCAGGTCCCTGCAAGCAGC..ATCCTGTATGGCTCCGTACAAA	68
RefSeq	.....	0
consensus		

	AAGCAATCACTAGTTTCTGCGTTGAGTAAACACAA.GCGCACATCTCTCCAGAGATG AAACAAAAAATATTTTTTTT.TGTGGGGAAGCCCCA...CGCACACTCCTCCCGGGGGG..	
Father	AAACAAAAAATATTTTTTTT.TGTGGGGAAGCCCCA...CGCACACTCCTCCCGGGGGG..	74
Mother	..	0
Sister	GAGCAATGAGTAGTTTCTTGTGCGTTGAGTAAACACAA.GCGCACATCTCTCCAGAGATG	179
Brother	..	0
Proband	AAGCGATGCTGAGTTATTCGTCTGAGTCAGCTGCAATGCCAGCTCACCTGCCGTGAGTTA	128
RefSeq	..	0
consensus	..	0

	G C   A A C G T C G G   G G T C C C A T   G G T T G A A G   G G G G   A G T T C	
Father	.GCTA.CAGAACCTCAGGT <b>TTCGTTCTTCCCTCCGG</b> .GTGAAG <b>TCTCGACGTCAGTTCA</b>	131
Mother	.....CGATGAT <b>TGCTCTCTCCTACGAGCGCGAGGGT</b> ....GAG <b>GTAAGCTCC</b>	44
Sister	AGCTAACGTAACGTCGGGT <b>TGGTTCTTCCATCTCGGTGTGGGGTCGTGGTG</b> .. <b>AGTCCT</b>	237
Brother	.....	0
Proband	CGCTAACG...TGCGAG <b>TAGGTTCTTTCATCAGAGG</b> ..T <b>CAGGTGGGGTGAGTGAGTCAT</b>	182
RefSeq	.....	0
consensus	* * *	** * ** *

logo

Father TTGTGGA.GGCATATCCA AAAATACCT.CCCAGGCAGGCACACATTCTTTCTC..AGAG 187  
 Mother TTTTGT..GACGCTGTTCAATAATTATTCCTCCCTGGC.GGCCACACATTCTTTCTCC..CTG 99  
 Sister TTGTGGATGGCACATCCAACAATAACTACCCACGC CGGCACACATTCTTTCTCTCAGTG 297  
 Brother ..... 0  
 Proband TTT.....ACGGAGCC.ATCATAACT.ACACACA CAGCCACACACTTCTTC.....AG 228  
 RefSeq ..... 0  
 consensus \*\* \* \* \*\* \* \* \* \* \* \* \* \*

logo

Father ACAGACAGAGGGATGAG.TGAGCTCAGTCAAGCGGGCGTGCTTTCTGTCTCAGGGGAGGG 246  
 Mother AGAAGCT.ATGGATGTC.GACCTCAGTCAACCGAGGAGCTTTCTGTCTGAG.GGAGGG 156  
 Sister AGAGGCTGAGGGATGAGATGAGCTCAGTCAAGCGGGCGTGCTTTCTGTCTCAGTGAGGG 357  
 Brother ..... 0  
 Proband ACAGACAGAGTGA TGAG.TAAGCTCAGTCAAGCGGGCGAGCTT.CTGTCTGGTGAGGG 286  
 RefSeq ..... 0  
 consensus \* \* \* \* \* \* \* \* \* \* \* \* \* \*

logo

Father GGGAAGGG.TTCATTCCGGCAAGCAAGGCAGGCAGCGTTCTTTGGAGGGGGGGGGGAGG 305  
 Mother GGAAGAGGATCTTTC CCGGCAGTCAAGGGAGGCGCGCTTCATTGGAGAAAAAGGGGAGGCG 216  
 Sister GGGGAGGGGTTCA TTCCGGCAATCAAGGCAGAGAGCGCTTCATTGGAGAGAGGGGGGAGG 417  
 Brother ..... 0  
 Proband AGAAGGGGAACC..TTCCGGCAGGCGAGGGAGGGGGCGGTGCGTTGAGGGAGGGGGAGAGG 344  
 RefSeq ..... 0  
 consensus \* \* \* \* \* \* \* \* \* \* \* \* \* \*

logo

Father AGCGGAGA.GTGAGTGG.GGGAGGCGTGAG.CAACC AACCCAGCCCTGCCACAAACCCATA 362  
 Mother AGCGAAAAATAATAGAGG.GACGCCCCGAAG.TGACAAGGCAAGCCCATCCACTATCGAGA 274  
 Sister AGCGGACA.GTCAGTGG.GGGGCGG.GAG.CAGCCAGCCCATCCCTAGTCACTAGCCATA 473  
 Brother ..... 0  
 Proband AGCAGAGA.GTGAGTGGAGGCACGCGCGAGGTGAGAAAGCCCTGCTTATCGACAGCCAGA 403  
 RefSeq ..... 0  
 consensus \*\*\* \* \* \* \* \* \* \* \* \* \* \* \* \*

logo

Father CTCATTCC.TGACCTTGCTGGCTGCCTCCATTTTATATAA..AGCTATGGAAACAGAGGG 419  
 Mother GTCATTCCGTTCAAGCCCGAGCTGCAACCAGATTGAAAAT..ACCTATAGAAAATGAGGG 332  
 Sister CTCATTCC.TGAGCTTGCTGGCCGCATCCAATTTAAATAT..AGGTATGGAAACAGAGGG 530  
 Brother ..... 0  
 Proband CTCATTCC.TGAGCTTCCGGGCCGCCCTCTTTTATTGATAGGGTAAGCAAAACAGAGGG 462  
 RefSeq ..... 0  
 consensus \* \* \* \* \* \* \* \* \* \* \* \* \*

logo

Individual	Sequence	Position
Father	GCCA.CA.TGTGCCTTTGGGCA..TGTAATATGTCACATGCAATGTGCTCTCTTTGTTTCC	475
Mother	GTCACCA.TGTGCCTTGGGCA...GAATATGACACAGGGCATGTGCGCTCTCTGTTTCC	388
Sister	AAAAACCA.TGTTGCCTTTGGGGGCGTATATGACACATGCAATGTGCTCTCTTTGTTTCC	589
Brother	.....TGTGCTCTCTCTGTTTCC	18
Proband	GGGCAACATGGTCCCTGGGGGCGTCTATGACACATGCGCATGTGCGCTCTCTGTTTCC	522
RefSeq	.....	0
consensus	*** ** *	

logo

Individual	Sequence	Position
Father	ATAGCTTTAAATAAAATGGGGCCAGCAAGGAAGCTCAGGAATGGGCTTGGCAATGGCAA	535
Mother	ATAGCTTTAAATAAAATGGGGCCAGCAAGGAAGCTCAGGAATGGGCTTGGCAATGGCAA	448
Sister	ATAGCTTTAAATAAAATGGGGCCAGCAAGGAAGCTCAGGAATGGGCTTGGCAATGGCAA	649
Brother	ATAGCTTTAAATAAAATGGGGCCAGCAAGGAAGCTCAGGAATGGGCTTGGCAATGGCAA	78
Proband	ATAGCTTTAAATAAAATGGGGCCAGCAAGGAAGCTCAGGAATGGGCTTGGCAATGGCAA	582
RefSeq	.....	0
consensus	*****	

logo

Individual	Sequence	Position
Father	GGCTTTGCTGCTCACCTCGGGCCTCCTCTGAGTCTCTGTCCCGCTCCTCCTCCTCTTCT	595
Mother	GGCTTTGCTGCTCACCTCGGGCCTCCTCTGAGTCTCTGTCCCGCTCCTCCTCCTCTTCT	508
Sister	GGCTTTGCTGCTCACCTCGGGCCTCCTCTGAGTCTCTGTCCCGCTCCTCCTCCTCTTCT	709
Brother	GGCTTTGCTGCTCACCTCGGGCCTCCTCTGAGTCTCTGTCCCGCTCCTCCTCCTCTTCT	138
Proband	GGCTTTGCTGCTCACCTCGGGCCTCCTCTGAGTCTCTGTCCCGCTCCTCCTCCTCTTCT	642
RefSeq	.....	0
consensus	*****	

logo

Individual	Sequence	Position
Father	CGAATGCCCTCTGCCTCCATTGCCGCCAGGAATGTTCCCTTTCCCTGAGCCGAGAGC	655
Mother	CGAATGCCCTCTGCCTCCATTGCCGCCAGGAATGTTCCCTTTCCCTGAGCCGAGAGC	568
Sister	CGAATGCCCTCTGCCTCCATTGCCGCCAGGAATGTTCCCTTTCCCTGAGCCGAGAGC	769
Brother	CGAATGCCCTCTGCCTCCATTGCCGCCAGGAATGTTCCCTTTCCCTGAGCCGAGAGC	198
Proband	CGAATGCCCTCTGCCTCCATTGCCGCCAGGAATGTTCCCTTTCCCTGAGCCGAGAGC	702
RefSeq	.....	0
consensus	*****	

logo

Individual	Sequence	Position
Father	ATGCCCTGGGCTTGACGGTGCTCATCCCTCAACTTGTCTCTCAAGGAGAAAGTGTGTGG	715
Mother	ATGCCCTGGGCTTGACGGTGCTCATCCCTCAACTTGTCTCTCAAGGAGAAAGTGTGTGG	628
Sister	ATGCCCTGGGCTTGACGGTGCTCATCCCTCAACTTGTCTCTCAAGGAGAAAGTGTGTGG	829
Brother	ATGCTCCTGGGCTTGACGGTGCTCATCCCTCAACTTGTCTCTCAAGGAGAAAGTGTGTGG	258
Proband	ATGCCCTGGGCTTGACGGTGCTCATCCCTCAACTTGTCTCTCAAGGAGAAAGTGTGTGG	762
RefSeq	.....GAGAAAGTGTGTGG	14
consensus	*****!!!!!!!!!!!!	

logo CCTGTGTGGGAATTTTGATGGCATCCAGAACAAATGACCTCACCAGCAGCAACCTCCAAGT

Father	CCTGTGTGGGAATTTTGATGGCATCCAGAACAAATGACCTCACCAGCAGCAACCTCCAAGT	775
Mother	CCTGTGTGGGAATTTTGATGGCATCCAGAACAAATGACCTCACCAGCAGCAACCTCCAAGT	688
Sister	CCTGTGTGGGAATTTTGATGGCATCCAGAACAAATGACCTCACCAGCAGCAACCTCCAAGT	889
Brother	CCTGTGTGGGAATTTTGATGGCATCCAGAACAAATGACCTCACCAGCAGCAACCTCCAAGT	318
Proband	CCTGTGTGGGAATTTTGATGGCATCCAGAACAAATGACCTCACCAGCAGCAACCTCCAAGT	822
RefSeq	CCTGTGTGGGAATTTTGATGGCATCCAGAACAAATGACCTCACCAGCAGCAACCTCCAAGT	74
consensus	!!	

logo GGAGGAAGACCCCTGTGGACTTTGGGAACTCCTGGAAAGTGAGCTCGCAGTGTGCTGACAC

Father	GGAGGAAGACCCCTGTGGACTTTGGGAACTCCTGGAAAGTGAGCTCGCAGTGTGCTGACAC	835
Mother	GGAGGAAGACCCCTGTGGACTTTGGGAACTCCTGGAAAGTGAGCTCGCAGTGTGCTGACAC	748
Sister	GGAGGAAGACCCCTGTGGACTTTGGGAACTCCTGGAAAGTGAGCTCGCAGTGTGCTGACAC	949
Brother	GGAGGAAGACCCCTGTGGACTTTGGGAACTCCTGGAAAGTGAGCTCGCAGTGTGCTGACAC	378
Proband	GGAGGAAGACCCCTGTGGACTTTGGGAACTCCTGGAAAGTGAGCTCGCAGTGTGCTGACAC	882
RefSeq	GGAGGAAGACCCCTGTGGACTTTGGGAACTCCTGGAAAGTGAGCTCGCAGTGTGCTGACAC	134
consensus	!!	

logo CAGAAAAGTACGTCTGGGTCTCTGTGTGGACAGAGCCCGAGAGCTTGCTTCCTGGAATGT

Father	CAGAAAAGTACGTCTGGGTCTCTGTGTGGACAGAGCCCGAGAGCTTGCTTCCTGGAATGT	895
Mother	CAGAAAAGTACGTCTGGGTCTCTGTGTGGACAGAGCCCGAGAGCTTGCTTCCTGGAATGT	808
Sister	CAGAAAAGTACGTCTGGGTCTCTGTGTGGACAGAGCCCGAGAGCTTGCTTCCTGGAATGT	1009
Brother	CAGAAAAGTACGTCTGGGTCTCTGTGTGGACAGAGCCCTAGAGCTTGCTTCCTGGAATGT	438
Proband	CAGAAAAGTACGTCTGGGTCTCTGTGTGGACAGAGCCCGAGAGCTTGCTTCCTGGAATGT	942
RefSeq	CAGAAAA.....	141
consensus	!!!!!!!!*****	

logo CCCTCTGTCCCCATGTCATGGGCTGAAAGGGAGTG

Father	CCCTCTGTCCCCATGTCATGGGCTGAAAGGGAGTG	931
Mother	CCCTCTGTCCCCATGTCATGGGACTGAAAGCTA...	840
Sister	CCCTCTGTCCCCATGTCATGGGACTGAAAGGG.GTA	1044
Brother	CCCTCTGTCCCCATGTCATGGGCTGAAATGGGGTTA	474
Proband	CCCTCTGTCCCCATGTCATGGGTCTGAAACGGGCGT	978
RefSeq	.....	141
consensus	*****	

☐ non conserved  
☒ ≥ 50% conserved