

Supplementary Material for the article:Inagaki et al., *Human Mutation***A Palindromic AT-rich Repeat in the *NFI* Gene is Hypervariable in Humans and Evolutionarily Conserved Among Primates**

Hidehito Inagaki, Tamae Ohye, Hiroshi Kogo, Kouji Yamada, Hiroe Kowa, Tamim H. Shaikh, Beverly S. Emanuel, and Hiroki Kurahashi

Supplementary Table S1. SNP genotyping of the individuals, cell lines and primates

No. or name	ethnicity	PATRR type		SNPs (distance from 17PATRR)			
				dbSNP2854306* (-14 kb)	2854308* (+4 kb)	964288 (+17 kb)	
Individuals	1	Japanese	S/S	A/A	G/G	A/A	
2	Japanese	L/S		A/G	G/A	A/G	
3	Japanese	L/L		G/G	A/A	G/G	
4	Japanese	L/S		A/G	G/A	A/G	
5	Japanese	L/S		A/G	G/A	A/G	
6	Japanese	L/S		A/G	G/A	A/G	
7	Japanese	L/S		A/G	G/A	A/G	
8	Japanese	L/L		G/G	A/A	G/G	
9	Japanese	L/S		A/G	G/A	A/G	
10	Japanese	L/S		A/G	G/A	A/G	
11	Japanese	S/S		A/A	G/G	A/A	
12	non-Jpn	L/S		A/G	G/A	A/G	
13	Japanese	L/S		n.d.	n.d.	A/G	
14	Japanese	L/L		G/G	A/A	G/G	
15	Japanese	L/L		G/G	A/A	G/G	
16	Japanese	L/L		G/G	A/A	G/G	
17	Japanese	S/S		A/A	G/G	A/A	
18	non-Jpn	L/S		A/G	G/A	A/G	
19	non-Jpn	L/S		A/G	G/A	A/G	
20	Japanese	L/S		A/G	G/A	A/G	
Cell lines	293		L/S	A/G	G/A	A/G	
	HeLa	Black	L/S	A/G	G/A	A/G	
	HepG2	Caucasian	L/S	A/G	G/A	A/G	
	HT1080	Caucasian	L/S	A/G	G/A	A/G	
	THP1		L/L	G/G	A/A	G/G	
Putative SNP type		17-L-PATRR		G	A	G	
		17-S-PATRR		A	G	A	
Primates	Gorilla			A/A	G/G	A/A	
	Chimpanzee (database)			A/A	A	G	A
	Rhesus monkey			A/A	G/G	A/A	
	African green monkey			A/A	A/A	G/G	A/A
	Tamarin			n.d.	n.d.	A/A	
	Owl monkey			n.d.	n.d.	n.d.	A/A

n.d.: not determined

*Two SNPs were genotyped by PCR amplification and sequencing. To amplify from both human and primates, PCR primer pairs were designed on conserved exons and long-PCR reactions were carried out. The PCR and sequencing primers were the followings:

dbSNP2854306: PCR forward 5'-GGTATAGTTTGCTTTTGTTCAGG-3'

PCR reverse 5'-GACAACACTAGTCTAGCCAGAATGG-3'

Sequencing 5'-GCTTGGACATTACAAATTCTACTG-3'

dbSNP2854308: PCR forward 5'-AGTTGCTTAAAAGGACCTGACACTTA-3'

PCR reverse 5'-GTGTATTCACCTCAAATTTGTCACA-3'

Sequencing 5'- ATGGCATGGTATTACCATCCAG -3'

Figure S1

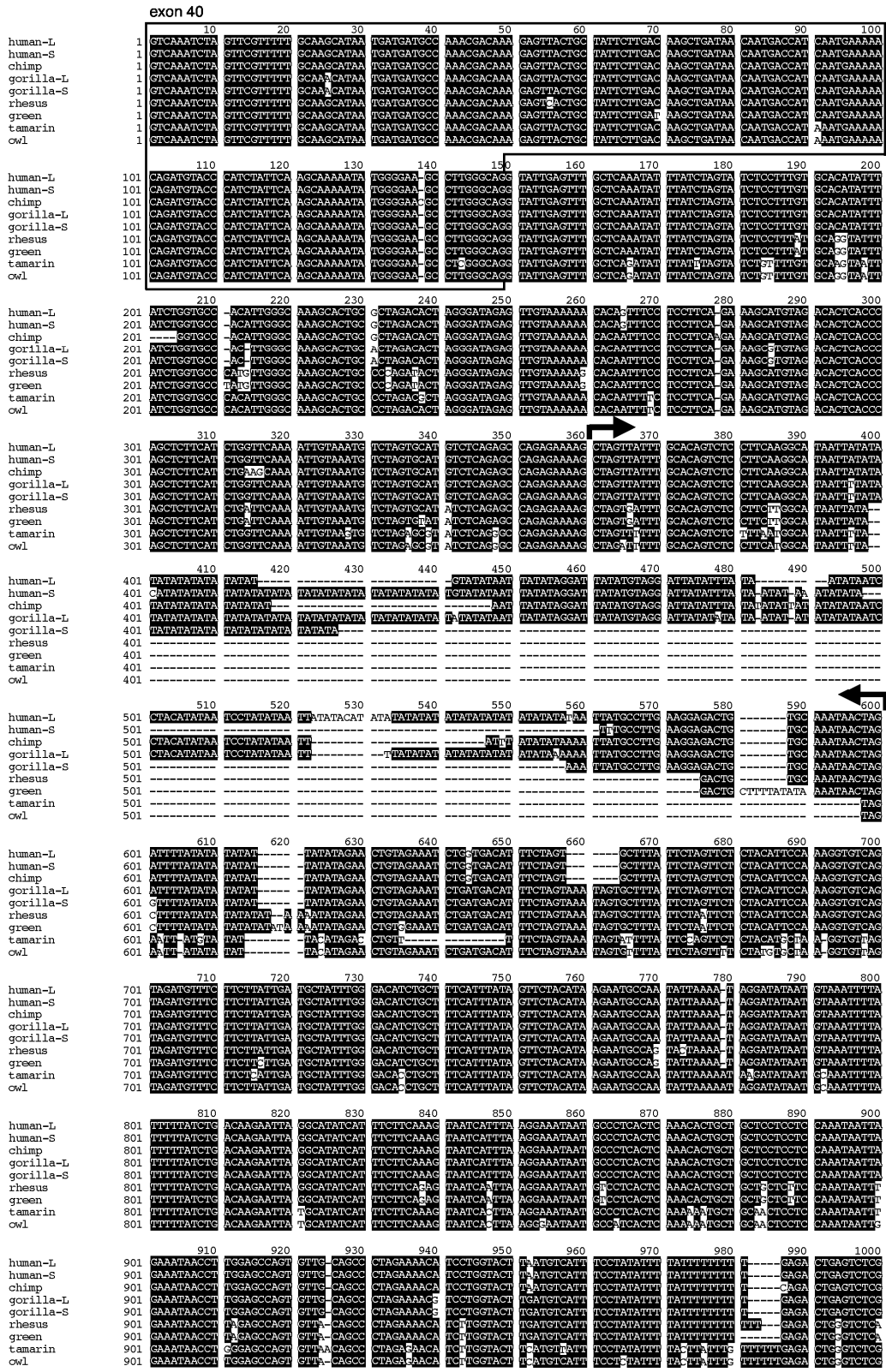


Figure S1 (continued)

	1010	1020	1030	1040	1050	1060	1070	1080	1090	1100		
human-L	1001	CCTGTCCACC	CAGGCTGGA	GTGAGTGGT	ACAGTCTGGG	CTCATTGCAA	CCTCTGCTCC	CCAGGCTCAA	GTGACCCCTTC	CACCTCAGCC	TCCCAGGTAG	
human-S	1001	CCTGTCCACC	CAGGCTGGA	GTGAGTGGT	ACAGTCTGGG	CTCATTGCAA	CCTCTGCTCC	CCAGGCTCAA	GTGACCCCTTC	CACCTCAGCC	TCCCAGGTAG	
chimp	1001	CCTGTCCACC	CAGGCTGGA	GTGAGTGGT	ACAGTCTGGG	CTCATTGCAA	CCTCTGCTCC	CCAGGCTCAA	GTGACCCCTTC	CACCTCAGCC	TCCCAGGTAG	
gorilla-L	1001	CCTGTCCACC	CAGGCTGGA	GTGAGTGGT	ACAGTCTGGG	CTCATTGCAA	CCTCTGCTCC	CCAGGCTCAA	GTGACCCCTTC	CACCTCAGCC	TCCCAGGTAG	
gorilla-S	1001	CCTGTCCACC	CAGGCTGGA	GTGAGTGGT	ACAGTCTGGG	CTCATTGCAA	CCTCTGCTCC	CCAGGCTCAA	GTGACCCCTTC	CACCTCAGCC	TCCCAGGTAG	
rhesus	1001	CCTGTCCACC	CAGGCTGGA	GTGAGTGGT	ACAGTCTGGG	CTCATTGCAA	CCTCTGCTCC	CCAGGCTCAA	GTGACCCCTTC	CACCTCAGCC	TCCCAGGTAG	
green	1001	CCTGTCCACC	CAGGCTGGA	GTGAGTGGT	ACAGTCTGGG	CTCATTGCAA	CCTCTGCTCC	CCAGGCTCAA	GTGACCCCTTC	CACCTCAGCC	TCCCAGGTAG	
tamarin	1001	CCTGTCCACC	CAGGCTGGA	GTGAGTGGT	ACAGTCTGGG	CTCATTGCAA	CCTCTGCTCC	CCAGGCTCAA	GTGACCCCTTC	CACCTCAGCC	TCCCAGGTAG	
owl	1001	CCTGTCCACC	CAGGCTGGA	GTGAGTGGT	ACAGTCTGGG	CTCATTGCAA	CCTCTGCTCC	CCAGGCTCAA	GTGACCCCTTC	CACCTCAGCC	TCCCAGGTAG	
		1110	1120	1130	1140	1150	1160	1170	1180	1190	1200	
human-L	1101	CTGGGCTCAG	AGGTGCACAC	CAACACACAC	GGCTAAATTTT	TGTATTTTTTT	TTTTTTTTTTT	TTTTTTT	AGATA	GATAGGGTATT	TGCCATGTTC	CCCAGGCTGG
human-S	1101	CTGGGCTCAG	AGGTGCACAC	CAACACACAC	GGCTAAATTTT	TGTATTTTTTT	TTTTTTTTTTT	TTTTTTT	TAGATA	GATAGGGTATT	TGCCATGTTC	CCCAGGCTGG
chimp	1101	CTGGGCTCAG	AGGTGCACAC	CAACACACAC	GGCTAAATTTT	TGTATTTTTTT	TTTTTTTTTTT	TTTTTTT	TAGATA	GATAGGGTATT	TGCCATGTTC	CCCAGGCTGG
gorilla-L	1101	CTGGGCTCAG	AGGTGCACAC	CAACACACAC	GGCTAAATTTT	TGTATTTTTTT	TTTTTTTTTTT	TTTTTTT	AGATA	GATAGGGTATT	TGCCATGTTC	CCCAGGCTGG
gorilla-S	1101	CTGGGCTCAG	AGGTGCACAC	CAACACACAC	GGCTAAATTTT	TGTATTTTTTT	TTTTTTTTTTT	TTTTTTT	AGATA	GATAGGGTATT	TGCCATGTTC	CCCAGGCTGG
rhesus	1101	CTGGGCTCAG	AGGTGCACAC	CAACACACAC	GGCTAAATTTT	TGTATTTTTTT	TTTTTTTTTTT	TTTTTTT	AGATA	GATAGGGTATT	TGCCATGTTC	CCCAGGCTGG
green	1101	CTGGGCTCAG	AGGTGCACAC	CAACACACAC	GGCTAAATTTT	TGTATTTTTTT	TTTTTTTTTTT	TTTTTTT	AGATA	GATAGGGTATT	TGCCATGTTC	CCCAGGCTGG
tamarin	1101	CTGGGCTCAG	AGGTGCACAC	CAACACACAC	GGCTAAATTTT	TGTATTTTTTT	TTTTTTTTTTT	TTTTTTT	AGATA	GATAGGGTATT	TGCCATGTTC	CCCAGGCTGG
owl	1101	CTGGGCTCAG	AGGTGCACAC	CAACACACAC	GGCTAAATTTT	TGTATTTTTTT	TTTTTTTTTTT	TTTTTTT	AGATA	GATAGGGTATT	TGCCATGTTC	CCCAGGCTGG
		1210	1220	1230	1240	1250	1260	1270	1280	1290	1300	
human-L	1201	CTTGAAGCTC	CTGGCCCTCAA	GCAATCCGGG	GGCTCAGGCC	TCCCACAGTG	CTGGGATTAC	AAGCCGTGAGC	CACCTGACACC	AGCTCTGTAT	TTTCAAAATTA	
human-S	1201	CTTGAAGCTC	CTGGCCCTCAA	GCAATCCGGG	GGCTCAGGCC	TCCCACAGTG	CTGGGATTAC	AAGCCGTGAGC	CACCTGACACC	AGCTCTGTAT	TTTCAAAATTA	
chimp	1201	CTTGAAGCTC	CTGGCCCTCAA	GCAATCCGGG	GGCTCAGGCC	TCCCACAGTG	CTGGGATTAC	AAGCCGTGAGC	CACCTGACACC	AGCTCTGTAT	TTTCAAAATTA	
gorilla-L	1201	CTTGAAGCTC	CTGGCCCTCAA	GCAATCCGGG	GGCTCAGGCC	TCCCACAGTG	CTGGGATTAC	AAGCCGTGAGC	CACCTGACACC	AGCTCTGTAT	TTTCAAAATTA	
gorilla-S	1201	CTTGAAGCTC	CTGGCCCTCAA	GCAATCCGGG	GGCTCAGGCC	TCCCACAGTG	CTGGGATTAC	AAGCCGTGAGC	CACCTGACACC	AGCTCTGTAT	TTTCAAAATTA	
rhesus	1201	CTTGAAGCTC	CTGGCCCTCAA	GCAATCCGGG	GGCTCAGGCC	TCCCACAGTG	CTGGGATTAC	AAGCCGTGAGC	CACCTGACACC	AGCTCTGTAT	TTTCAAAATTA	
green	1201	CTTGAAGCTC	CTGGCCCTCAA	GCAATCCGGG	GGCTCAGGCC	TCCCACAGTG	CTGGGATTAC	AAGCCGTGAGC	CACCTGACACC	AGCTCTGTAT	TTTCAAAATTA	
tamarin	1201	CTTGAAGCTC	CTGGCCCTCAA	GCAATCCGGG	GGCTCAGGCC	TCCCACAGTG	CTGGGATTAC	AAGCCGTGAGC	CACCTGACACC	AGCTCTGTAT	TTTCAAAATTA	
owl	1201	CTTGAAGCTC	CTGGCCCTCAA	GCAATCCGGG	GGCTCAGGCC	TCCCACAGTG	CTGGGATTAC	AAGCCGTGAGC	CACCTGACACC	AGCTCTGTAT	TTTCAAAATTA	
		1310	1320	1330	1340	1350	1360	1370	1380	1390	1400	
human-L	1301	CTATTTTATA	TGATGCTAC	ATTACTGTCT	TAGAAAAGCA	TAGAGACTCT	TTGCAAGTGA	CTGAAGATAG	PATTGATAGA	GATTTT	-----	
human-S	1301	CTATTTTATA	TGATGCTAC	ATTACTGTCT	TAGAAAAGCA	TAGAGACTCT	TTGCAAGTGA	CTGAAGATAG	PATTGATAGA	GATTTT	-----	
chimp	1301	CTATTTTATA	TGATGCTAC	ATTACTGTCT	TAGAAAAGCA	TAGAGACTCT	TTGCAAGTGA	CTGAAGATAG	PATTGATAGA	GATTTT	-----	
gorilla-L	1301	CTATTTTATA	TGATGCTAC	ATTACTGTCT	TAGAAAAGCA	TAGAGACTCT	TTGCAAGTGA	CTGAAGATAG	PATTGATAGA	GATTTT	-----	
gorilla-S	1301	CTATTTTATA	TGATGCTAC	ATTACTGTCT	TAGAAAAGCA	TAGAGACTCT	TTGCAAGTGA	CTGAAGATAG	PATTGATAGA	GATTTT	-----	
rhesus	1301	CTATTTTATA	TGATGCTAC	ATTACTGTCT	TAGAAAAGCA	TAGAGACTCT	TTGCAAGTGA	CTGAAGATAG	PATTGATAGA	GATTTT	-----	
green	1301	CTATTTTATA	TGATGCTAC	ATTACTGTCT	TAGAAAAGCA	TAGAGACTCT	TTGCAAGTGA	CTGAAGATAG	PATTGATAGA	GATTTT	-----	
tamarin	1301	CTATTTTATA	TGATGCTAC	ATTACTGTCT	TAGAAAAGCA	TAGAGACTCT	TTGCAAGTGA	CTGAAGATAG	PATTGATAGA	GATTTT	-----	
owl	1301	CTATTTTATA	TGATGCTAC	ATTACTGTCT	TAGAAAAGCA	TAGAGACTCT	TTGCAAGTGA	CTGAAGATAG	PATTGATAGA	GATTTT	-----	
		1410	1420	1430	1440	1450	1460	1470	1480	1490	1500	
human-L	1401	-----	-----	-----	-----	-----	-----	-----	-----	-----	-----	
human-S	1401	-----	-----	-----	-----	-----	-----	-----	-----	-----	-----	
chimp	1401	-----	-----	-----	-----	-----	-----	-----	-----	-----	-----	
gorilla-L	1401	-----	-----	-----	-----	-----	-----	-----	-----	-----	-----	
gorilla-S	1401	-----	-----	-----	-----	-----	-----	-----	-----	-----	-----	
rhesus	1401	-----	-----	-----	-----	-----	-----	-----	-----	-----	-----	
green	1401	-----	-----	-----	-----	-----	-----	-----	-----	-----	-----	
tamarin	1401	TTTGTAGAGC	GAGTTTCACT	CTTGTACC	AGACTGGAGT	GCAATGGCGC	AGTCTCAGCT	CACCCAAATC	TCCGCTTCT	GGGTTCAGGC	AATTCCTCTG	
owl	1401	TTTGTAGAGC	GAGTTTCACT	CTTGTACC	AGACTGGAGT	GCAATGGCGC	AGTCTCAGCT	CACCCAAATC	TCCGCTTCT	GGGTTCAGGC	AATTCCTCTG	
		1510	1520	1530	1540	1550	1560	1570	1580	1590	1600	
human-L	1501	-----	-----	-----	-----	-----	-----	-----	-----	-----	-----	
human-S	1501	-----	-----	-----	-----	-----	-----	-----	-----	-----	-----	
chimp	1501	-----	-----	-----	-----	-----	-----	-----	-----	-----	-----	
gorilla-L	1501	-----	-----	-----	-----	-----	-----	-----	-----	-----	-----	
gorilla-S	1501	-----	-----	-----	-----	-----	-----	-----	-----	-----	-----	
rhesus	1501	-----	-----	-----	-----	-----	-----	-----	-----	-----	-----	
green	1501	-----	-----	-----	-----	-----	-----	-----	-----	-----	-----	
tamarin	1501	CCTCAGGCTC	CTGAGTACCT	GGGATPACAG	GCACGCCGCA	CCATGCCOCAG	CTAATTTTTTT	ATATTTTTTAG	TAGAGACGGA	GTTCACCAT	GTTCACCAGG	
owl	1501	CCTCAGGCTC	CTGAGTACCT	GGGATPACAG	GCACGCCGCA	CCATGCCOCAG	CTAATTTTTTT	ATATTTTTTAG	TAGAGACGGA	GTTCACCAT	GTTCACCAGG	
		1610	1620	1630	1640	1650	1660	1670	1680	1690	1700	
human-L	1601	-----	-----	-----	-----	-----	-----	-----	-----	-----	AA	
human-S	1601	-----	-----	-----	-----	-----	-----	-----	-----	-----	AA	
chimp	1601	-----	-----	-----	-----	-----	-----	-----	-----	-----	AA	
gorilla-L	1601	-----	-----	-----	-----	-----	-----	-----	-----	-----	AA	
gorilla-S	1601	-----	-----	-----	-----	-----	-----	-----	-----	-----	AA	
rhesus	1601	-----	-----	-----	-----	-----	-----	-----	-----	-----	AA	
green	1601	-----	-----	-----	-----	-----	-----	-----	-----	-----	AA	
tamarin	1601	ATGGTCTCAA	TCTCTTGACC	TCGTGATCCA	CCCGCTCCGG	CCTCCCAAAG	TGCTGGGAT	ACAGGCTTGA	GCCACCGCGC	CGGCGCCGAT	AGAGATTTTTA	
owl	1601	ATGGTCTCAA	TCTCTTGACC	TCGTGATCCA	CCCGCTCCGG	CCTCCCAAAG	TGCTGGGAT	ACAGGCTTGA	GCCACCGCGC	CGGCGCCGAT	AGAGATTTTTA	
		1710	1720	1730	1740	1750	1760	1770	1780	1790	1800	
human-L	1701	AATTTTTGAA	TACCAAATCC	TAAATCTCTG	AAGGAGTCAA	ATGAATATAC	TCATCCTTTC	CTGGATATTT	TATAATTAAT	TGTAATTTACT	GACAGGCTG	
human-S	1701	AATTTTTGAA	TACCAAATCC	TAAATCTCTG	AAGGAGTCAA	ATGAATATAC	TCATCCTTTC	CTGGATATTT	TATAATTAAT	TGTAATTTACT	GACAGGCTG	
chimp	1701	AATTTTTGAA	TACCAAATCC	TAAATCTCTG	AAGGAGTCAA	ATGAATATAC	TCATCCTTTC	CTGGATATTT	TATAATTAAT	TGTAATTTACT	GACAGGCTG	
gorilla-L	1701	AATTTTTGAA	TACCAAATCC	TAAATCTCTG	AAGGAGTCAA	ATGAATATAC	TCATCCTTTC	CTGGATATTT	TATAATTAAT	TGTAATTTACT	GACAGGCTG	
gorilla-S	1701	AATTTTTGAA	TACCAAATCC	TAAATCTCTG	AAGGAGTCAA	ATGAATATAC	TCATCCTTTC	CTGGATATTT	TATAATTAAT	TGTAATTTACT	GACAGGCTG	
rhesus	1701	AATTTTTGAA	TACCAAATCC	TAAATCTCTG	AAGGAGTCAA	ATGAATATAC	TCATCCTTTC	CTGGATATTT	TATAATTAAT	TGTAATTTACT	GACAGGCTG	
green	1701	AATTTTTGAA	TACCAAATCC	TAAATCTCTG	AAGGAGTCAA	ATGAATATAC	TCATCCTTTC	CTGGATATTT	TATAATTAAT	TGTAATTTACT	GACAGGCTG	
tamarin	1701	AATTTTTGAA	TACCAAATCC	TAAATCTCTG	AAGGAGTCAA	ATGAATATAC	TCATCCTTTC	CTGGATATTT	TATAATTAAT	TGTAATTTACT	GACAGGCTG	
owl	1701	AATTTTTGAA	TACCAAATCC	TAAATCTCTG	AAGGAGTCAA	ATGAATATAC	TCATCCTTTC	CTGGATATTT	TATAATTAAT	TGTAATTTACT	GACAGGCTG	
		1810	1820	1830	1840	1850	1860	1870	1880	1890	1900	
human-L	1801	TAATAAAAA	CTAGTATTTT	TCAGGCCTCA	GSTAAAATAG	AATTTTCATA	TTGATTAGGC	TGTTCCAARG	AATATTTTTT	AATTAATAAT	TAATAATTTA	
human-S	1801	TAATAAAAA	CTAGTATTTT	TCAGGCCTCA	GSTAAAATAG	AATTTTCATA	TTGATTAGGC	TGTTCCAARG	AATATTTTTT	AATTAATAAT	TAATAATTTA	
chimp	1801	TAATAAAAA	CTAGTATTTT	TCAGGCCTCA	GSTAAAATAG	AATTTTCATA	TTGATTAGGC	TGTTCCAARG	AATATTTTTT	AATTAATAAT	TAATAATTTA	
gorilla-L	1801	TAATAAAAA	CTAGTATTTT	TCAGGCCTCA	GSTAAAATAG	AATTTTCATA	TTGATTAGGC	TGTTCCAARG	AATATTTTTT	AATTAATAAT	TAATAATTTA	
gorilla-S	1801	TAATAAAAA	CTAGTATTTT	TCAGGCCTCA	GSTAAAATAG	AATTTTCATA	TTGATTAGGC	TGTTCCAARG	AATATTTTTT	AATTAATAAT	TAATAATTTA	
rhesus	1801	TAATAAAAA	CTAGTATTTT	TCAGGCCTCA	GSTAAAATAG	AATTTTCATA	TTGATTAGGC	TGTTCCAARG	AATATTTTTT	AATTAATAAT	TAATAATTTA	
green	1801	TAATAAAAA	CTAGTATTTT	TCAGGCCTCA	GSTAAAATAG	AATTTTCATA	TTGATTAGGC	TGTTCCAARG	AATATTTTTT	AATTAATAAT	TAATAATTTA	
tamarin	1801	TAATAAAAA	CTAGTATTTT	TCAGGCCTCA	GSTAAAATAG	AATTTTCATA	TTGATTAGGC	TGTTCCAARG	AATATTTTTT	AATTAATAAT	TAATAATTTA	
owl	1801	TAATAAAAA	CTAGTATTTT	TCAGGCCTCA	GSTAAAATAG	AATTTTCATA	TTGATTAGGC	TGTTCCAARG	AATATTTTTT	AATTAATAAT	TAATAATTTA	
		1910	1920	1930	1940	1950	1960					
human-L	1901	GAGTGATTAA	AAACATGTTA	TTTTTCCTTC	TCACACTAGAT	TACAGATCTG	CTTGATGTTG	TACTA				
human-S	1901	GAGTGATTAA	AAACATGTTA	TTTTTCCTTC	TCACACTAGAT	TACAGATCTG	CTTGATGTTG	TACTA				
chimp	1901	GAGTGATTAA	AAACATGTTA	TTTTTCCTTC	TCACACTAGAT	TACAGATCTG	CTTGATGTTG	TACTA				
gorilla-L	1901	GAGTGATTAA	AAACATGTTA	TTTTTCCTTC	TCACACTAGAT	TACAGATCTG	CTTGATGTTG	TACTA				
gorilla-S	1901	GAGTGATTAA	AAACATGTTA	TTTTTCCTTC	TCACACTAGAT	TACAGATCTG	CTTGATGTTG	TACTA				
rhesus	1901	GAGTGATTAA	AAACATGTTA	TTTTTCCTTC	TCACACTAGAT	TACAGATCTG	CTTGATGTTG	TACTA				
green	1901	GAGTGATTAA	AAACATGTTA	TTTTTCCTTC	TCACACTAGAT	TACAGATCTG	CTTGATGTTG	TACTA				
tamarin	1901	GAGTGATTAA	AAACATGTTA	TTTTTCCTTC	TCACACTAGAT	TACAGATCTG	CTTGATGTTG	TACTA				
owl	1901	GAGTGATTAA	AAACATGTTA	TTTTTCCTTC	TCACACTAGAT	TACAGATCTG	CTTGATGTTG	TACTA			</	