Supplementary Material for the article:

Inagaki et al., ***Human Mutation***

A Palindromic AT-rich Repeat in the *NF1* Gene is Hypervariable in Humans and Evolutionarily Conserved Among Primates

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

				SNPs (distance from 17PATRR)			
No. or name	ethnicity	PATRR type		dbSNP2854306* (-14 kb)	2854308* (+4 kb)	(+17 kb)	964288
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	Japaense	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	G		A
	Rhesus monkey			A/A	G/G	A/A	
	African green monkey			A/A	G/G		A/A
	Tamarin			n.d	n.d.	A/A	
	Owl monkey			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 foward 5'- GGTATAGTTTGCTTTTGTTCCAGG -3'
PCR reverse 5'- GACAACTAGTCTAGCCAGAATGG -3'
Sequencing 5'- GCTTGGACATTACAAATTCTACTG -3'

dbSNP2854308: PCR forward 5'- AGTTGCTTAAAAGGACCTGACACTTA -3'
PCR reverse 5'- GTGTATTCACTTCAAATTTGTCACA -3'

Sequencing 5’- ATGGCATGGTATTACCATCCAG -3’

Figure S1

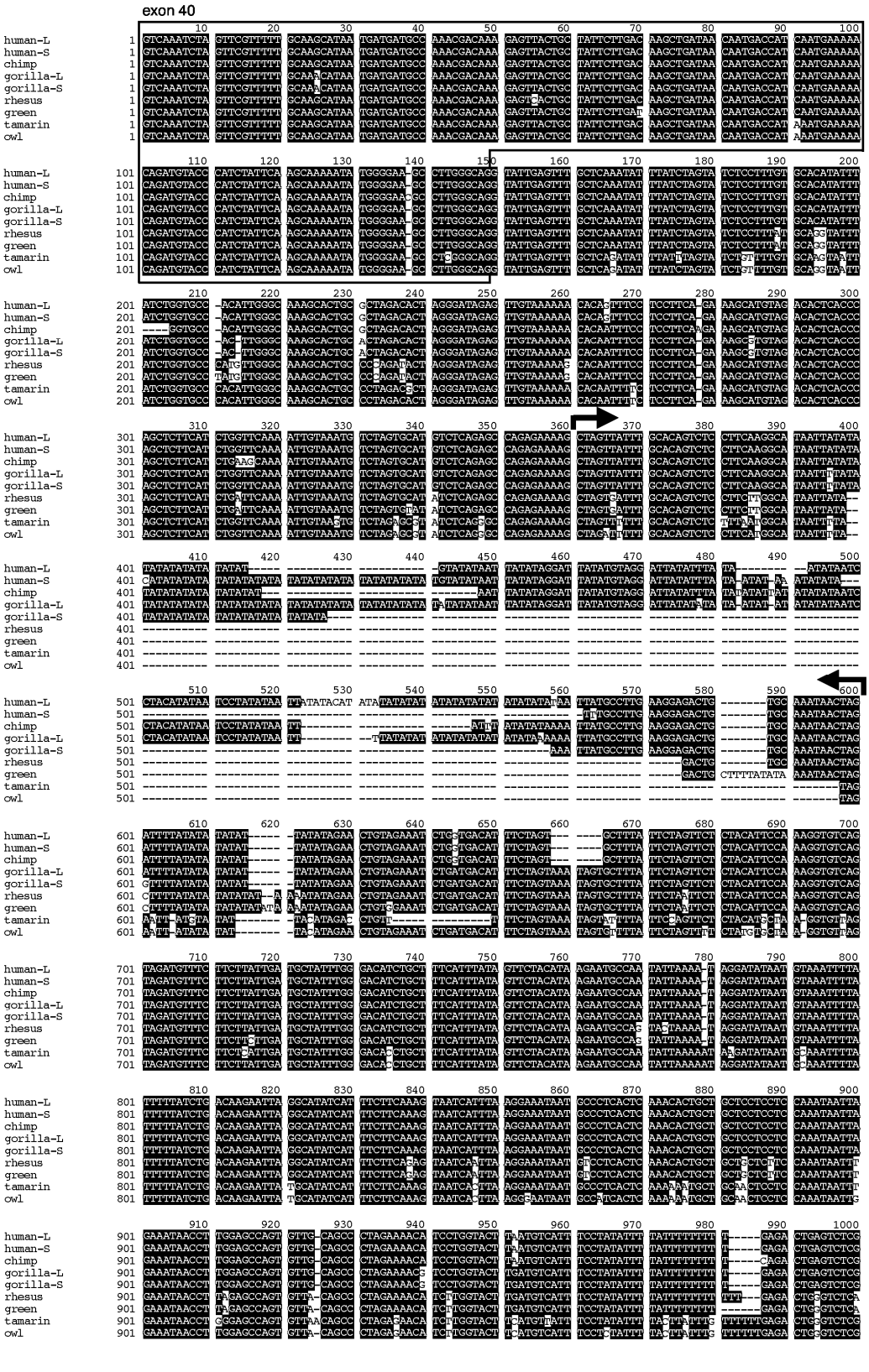
exon 40
human-L
human-S
chimp
gorilla-L
gorilla-S
rhesus
green
tamarin
owl

Figure S1 (continued)

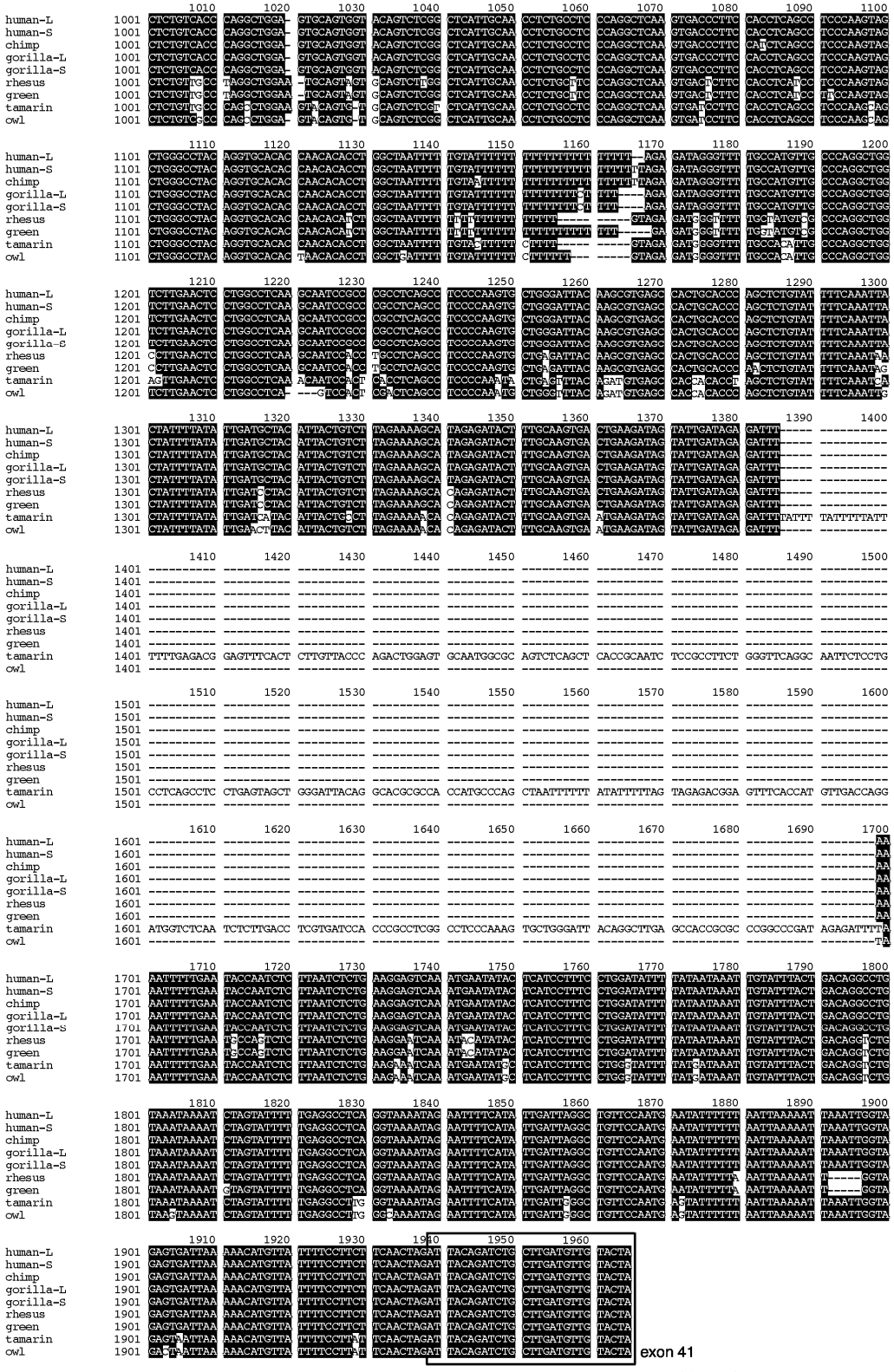

Figure S1 Alignment of nucleotide sequences of human and primates between exon 40 to 41. Boxes and arrows indicate exons and PATRR region, respectively. Other abbreviations are the same in Figure 4A.