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Supplemental Data

Mutations in *SPATA7* Cause Leber Congenital Amaurosis and Juvenile Retinitis Pigmentosa

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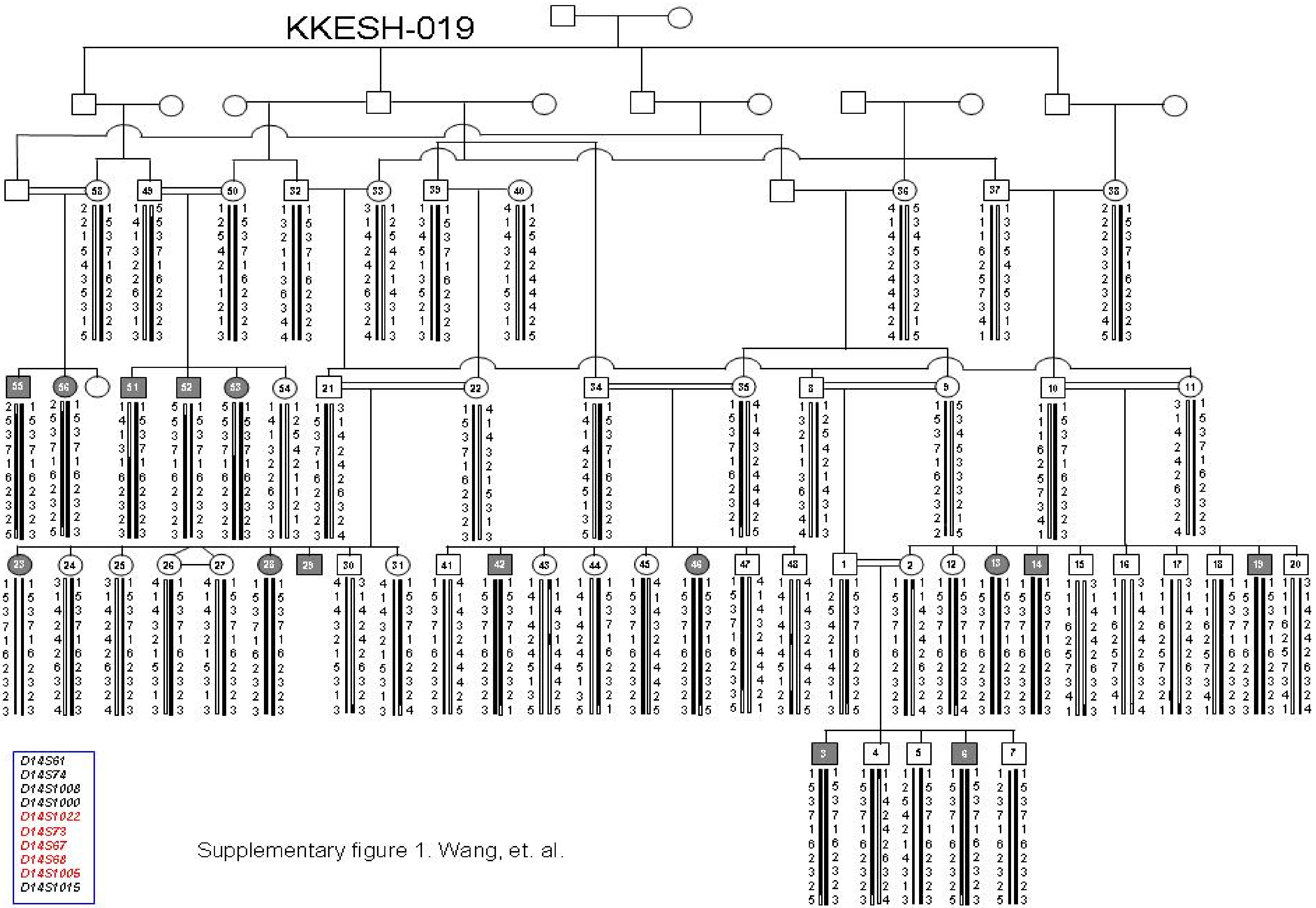
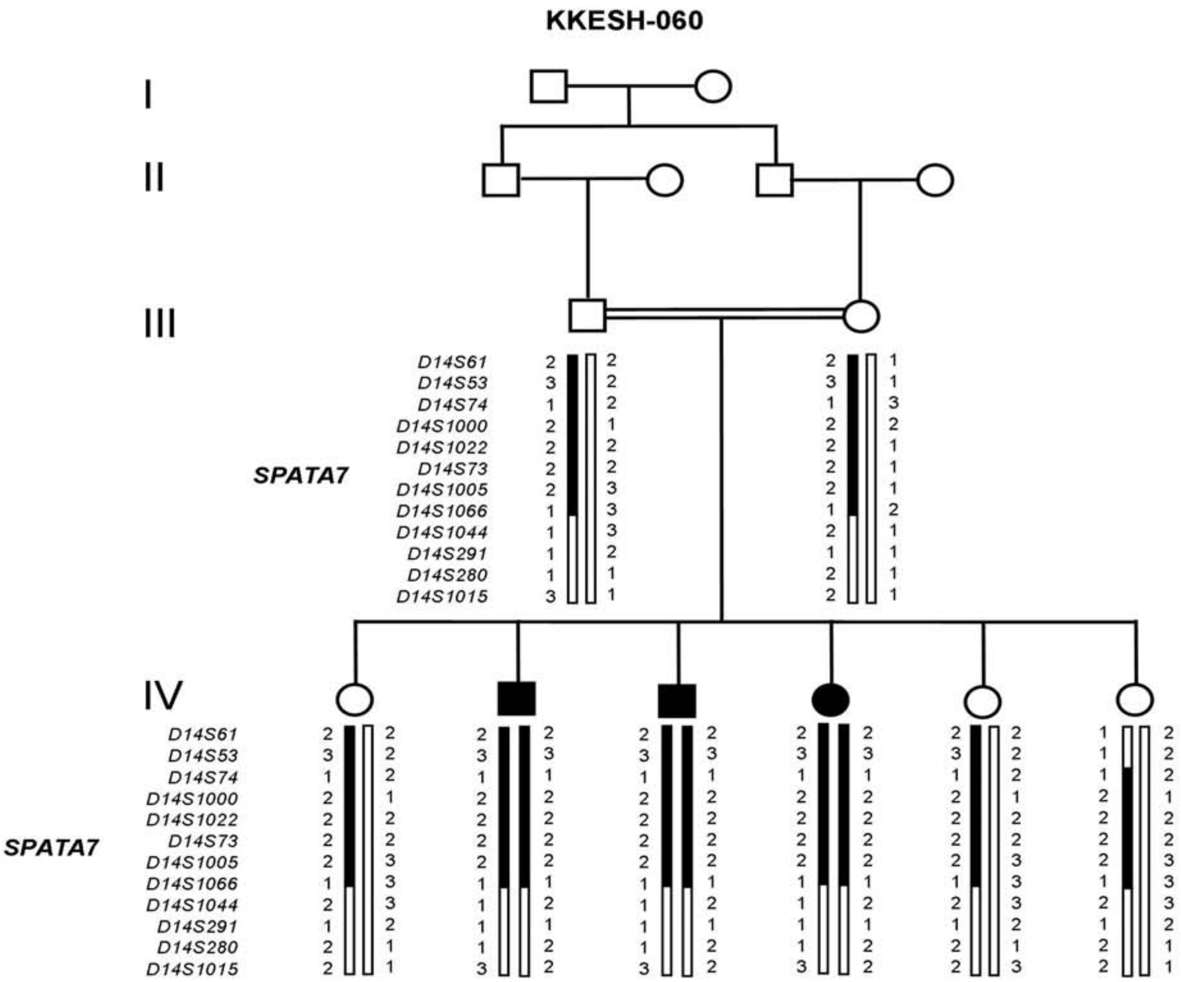


Supplementary figure 1. Wang, et. al.

Figure S1. Pedigree and Haplotypes of KKESH-019

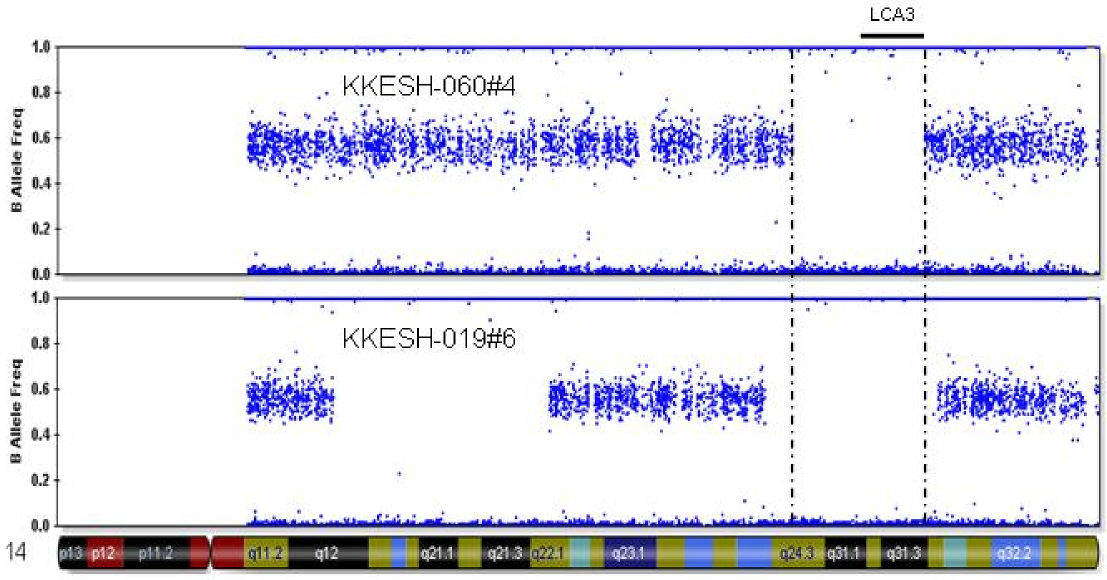
Genotyped markers between *D14S61* and *D14S1015* are shown in the blue box, and each individual's allele numbers are listed. The disease allele is indicated in black. The markers that show homozygous in all affected members are in red.



Supplementary Figure 2. Wang, et. al.

Figure S2. Homozygosity Mapping of KKESH-060

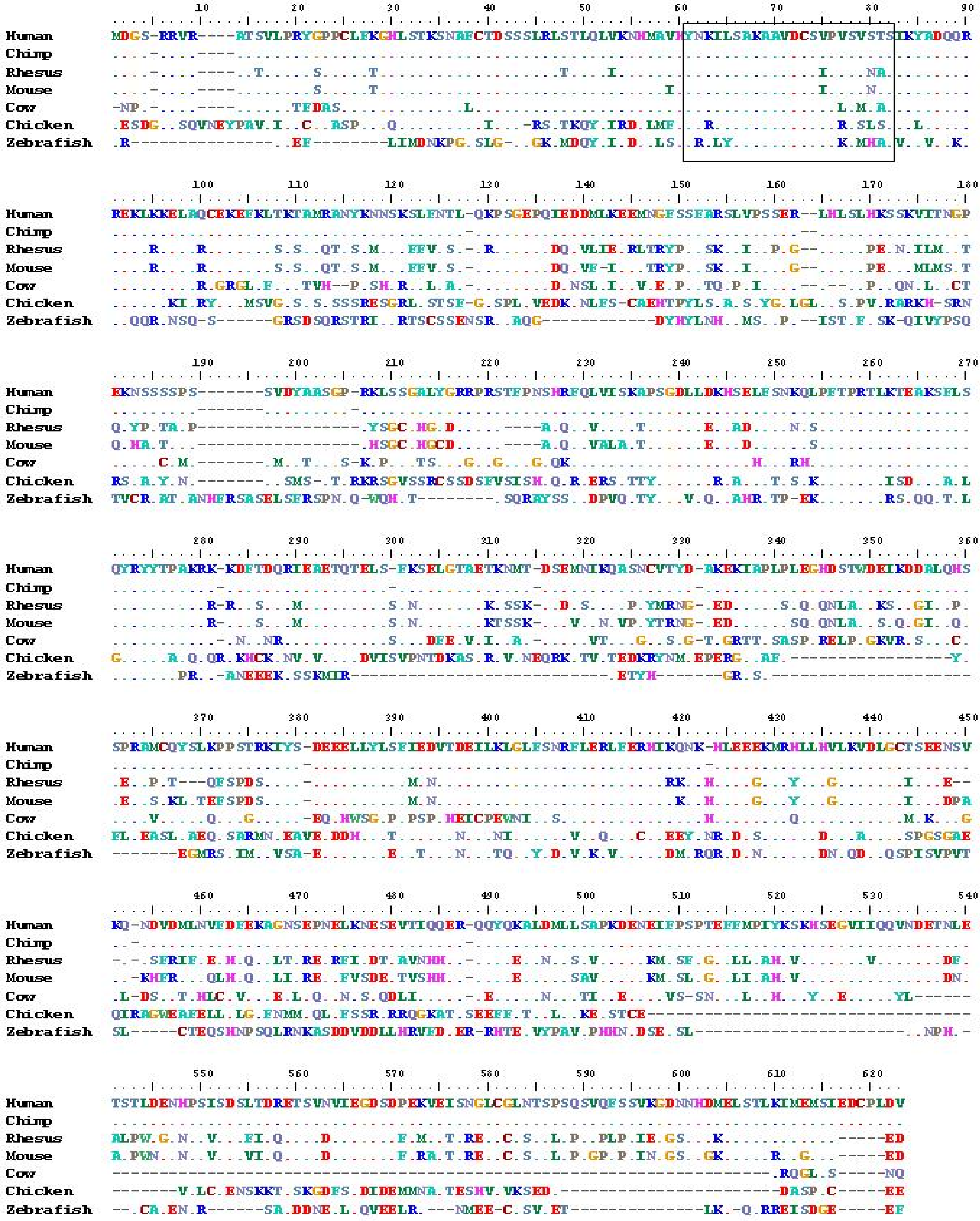
Genotyped markers between *D14S61* and *D14S1015* are shown, and each individual's allele numbers are listed. The homozygous region shared by three affected members is indicated in black. The *SPATA7* gene is located between markers *D14S73* and *D14S1005*.



Supplementary Figure 3. Wang, et. al.

Figure S3. SNP Mapping Is Consistent with STR Results

One patient from each LCA3 family (KKESH-060#4 and KKESH-019#6) was chosen to do the SNP genotyping. The homozygous block of both KKESH-019#6 and KKESH-060#4 on chromosome 14 are generated by Illumina Beadsstudio software. The shared the region, which is from 14q24.3 to 14q31.3, is indicated by a black dash. The bar indicates the homozygous region generated by the STR genotyping based on all the members from KKESH-060 and KKESH-019.



Supplementary Figure 4. Wang, et. al.

Figure S4. Amino Acid Alignments of SPATA7 Proteins from Different Species Reveal a Highly Conserved Protein

Amino acid sequences were aligned using Tcoffee (http://www.tcoffee.org/Projects_home_page/t_coffee_home_page.html). The dots below each line represent the same amino acid as in the human sequence. A predicted transmembrane domain is highlighted.

Table S1. List of Microsatellite Markers

Marker	Size	Location	Max Heterozygosity
D14S61	197-227	chr14:75404993	0.81
D14S53	135-155	chr14:75992089	NA
D14S42	115-133	chr14:76200151	0.68
D14S59	99-109	chr14:77142282	0.88
D14S74	281-313	chr14:77728132	0.8
D14S1008	112-126	chr14:78971571	0.66
D14S1000	107-133	chr14:81175672	0.7
D14S1022	236-246	chr14:84845288	0.73
D14S73	99-119	chr14:86152265	0.67
D14S67	133-167	chr14:87457553	0.88
D14S68	148-172	chr14:87697387	0.89
D14S1005	204-230	chr14:88447727	0.72
D14S1066	288-298	chr14:88872000	0.66
D14s1044	219-261	chr14:89140146	0.65
D14S291	210-216	chr14:90339518	0.46
D14S280	229-241	chr14:91252620	0.69
D14S1015	252-264	chr14:91806196	0.76

A total of 17 STR markers that span the *LCA3* locus are used to genotype all members of the potential patient families. The identification and chromosomal locations of these markers are listed.

Table S2. Primers of *SPATA7* Exons

Primer name	Primer sequence(5'-3')	size	temp
SPATA7-1-F	CTCGTGTAAAACGACGGCCAGTCGCAACTGTCCTCCTAGTACC	486	55
SPATA7-1-R	CTGCTCAGGAAACAGCTATGACACAAATTCAGGGCAAAGAAGC		
SPATA7-2-F	CTCGTGTAAAACGACGGCCAGTTTTAATGCTGTAACTCAGACTTCCT	369	55
SPATA7-2-R	CTGCTCAGGAAACAGCTATGACTGAAGTTCAAATATTCGTCAAATG		
SPATA7-3-F	CTCGTGTAAAACGACGGCCAGTAAGGTTTGAACCCAAATGGTC	475	55
SPATA7-3-R	CTGCTCAGGAAACAGCTATGACCAAAAATGGGTATGAATTTGCT		
SPATA7-4-F	CTCGTGTAAAACGACGGCCAGTCAAGGTCTGGAACATTTTGTGA	334	55
SPATA7-4-R	CTGCTCAGGAAACAGCTATGACTGTTTATGTGGCACAGGAATTT		
SPATA7-5-F	CTCGTGTAAAACGACGGCCAGTATCTAGAGGCACATGTGAAATAAA	398	55
SPATA7-5-R	CTGCTCAGGAAACAGCTATGACCAAAGTCAGATTGTACCACTAAAGAA		
SPATA7-6.1-F	CTCGTGTAAAACGACGGCCAGTTTTTGTAAACCCTTGAGGCTATC	458	55
SPATA7-6.1-R	CTGCTCAGGAAACAGCTATGACGGAGTGAATGGCAATTGTTTGT		
SPATA7-6.2-F	CTCGTGTAAAACGACGGCCAGTAGTCATCACAAATGGTCCTGAG	467	55
SPATA7-6.2-R	CTGCTCAGGAAACAGCTATGACTTCCAATCAAAAGGGCACTATC		
SPATA7-7-F	CTCGTGTAAAACGACGGCCAGTTCTGGCAGTAGGTTTTAGTTGTTTT	449	55
SPATA7-7-R	CTGCTCAGGAAACAGCTATGACTGTATGATAAGTGCCACCAACAG		
SPATA7-8-F	CTCGTGTAAAACGACGGCCAGTTGCTGTGTTATATTCTGCTTTCG	296	55
SPATA7-8-R	CTGCTCAGGAAACAGCTATGACTAGATTGGAGCATGCAATTAAA		
SPATA7-9-F	CTCGTGTAAAACGACGGCCAGTCATTAACCTTAGTCAAATTGTCATTG	688	55
SPATA7-9-R	CTGCTCAGGAAACAGCTATGACTGGTTTCTTTGATTCTTAATCCTTG		
SPATA7-10-F	CTCGTGTAAAACGACGGCCAGTCCCAGTGGATTGCATTTGA	600	55
SPATA7-10-R	CTGCTCAGGAAACAGCTATGACGGTGAACTTCCCCTAGAGTATGA		
SPATA7-11-F	CTCGTGTAAAACGACGGCCAGTTTTTCAACCTTTGTAGTTTCAGTG	300	55
SPATA7-11-R	CTGCTCAGGAAACAGCTATGACTTCCTTTCACTTCTCCCACCAC		
SPATA7-12.1-F	CTCGTGTAAAACGACGGCCAGTAATCCTGTGAGATTTTCAGCAC	483	55
SPATA7-12.1-R	CTGCTCAGGAAACAGCTATGACTCACAGAAGTTTCCCGATCTGT		
SPATA7-12.2-F	CTCGTGTAAAACGACGGCCAGTGAAGTAACAATTCAGCAGGAACG	469	55
SPATA7-12.2-R	CTGCTCAGGAAACAGCTATGACTGAGTTACTGGCCATTTGAGGT		