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Supplemental Data

Reccurrent *F8* Intronic Deletion Found in Mild Hemophilia A Causes *Alu* Exonization

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Supplemental data

Table S1: Primers used for *F8* RT-PCR analysis.

Primer pair	Forward primer		Reverse primer		Product size, bp
	Exonic site	Sequence	Exonic site	Sequence	
A	1	CTTTTGCGATTCTGCTTTAG	9	GCTGAGGGCCATTGTTCAAA	1273
A1	1	CTTTTGCGATTCTGCTTTAG	5-6	GAGTGCCAACTTTTCCCTTCAT	650
A2	5	AAGGAAAAGACACAGACCTT	7-8	TCATGTTGGTGGGAAGAGATA	398
B	7	TTACTGCTCAAACACTCTTG	14	TCATATTTGGCTTCTTGGAG	1561
B1	7	TTACTGCTCAAACACTCTTG	11	AGTTGGCCCATCTTCTACAGTCACTGTC	686
B2[a]	13	TGCCTGACCCGCTATTACTC	14	AGAAGCTTCTTGGTTCAATG	645
C	14	GATACCATTTTGTCCCTGAA	22	ACATGATGATAAACTGAGAGA	1528
C1	14	GATACCATTTTGTCCCTGAA	17	CATGGAAGCGATAAT	973
C2	17	AAAATATGGAAAGAAACTGC	22	ACATGATGATAAACTGAGAGA	636
D	19	CCTTATTGGCGAGCATCTACA	3'UTR	TTGCCTAGTTATATTGGAAG	1254
D1	22	TTATTCACGGCATCAAGACCC	23	AAATCACAGCCCATCAACTCC	274
D2	23	CTCCAATTATTGCTCGATACAT	25	CTGGTAAGCAGAGATTTTAC	337
D3[a]	24	GCCATTGGGAATGGAGAGTA	3'UTR	AGTTAATTCAGGAGGCTTCA	606

Reverse primers of pairs A, B, C and D are used for the reverse transcription; Primer pairs A, B, C and D are used for the first PCRs; Primer pairs A1, A2, B1, B2, C1, C2, D1, D2, D3 are used for the second nested PCRs. [a] Primers previously published by el-Maarri *et al.*[22]

Table S2: Primers used for *F8* intron 13 amplification and sequencing and for making *F8* exon 13 minigene construct

Primer pair	Forward primer		Reverse primer		Product size, bp
	site	Sequence	site	Sequence	
E	Intron 12 (-202)	TGGTTTATGACTGTCTCCTCACA	Intron 13 (+770)	GCTGAGGGCCATTGTTCAAA	1182
F	Exon 13	AGCATTGGAGCACAGACTGA	Intron 13 (+675)	AGACCATCCTGGCTAACACG	810
G	Intron 12 (-202)	*CTAAACAGCCACATATG*TGGTTTA TGACTGTCTCCTCACA	Intron 13 (+770)	*CCCCCCTCGACCATATG*CAAAAGAA GACATTCAGGCTGGGC	1182

Primer pair E are used for the amplification reaction; Primer pair F are used for the sequencing; Primer pair G are used for making minigene construct. Sequences in italic correspond to specific tail required for in-fusion reaction.

Table S3. Description of *F8* intragenic markers used in this study

Marker	Genomic position (hg38)	Type of repeat
STR 1	chrX:155,002,613-155,002,646	(CA)n
STR 9	chrX:154,964,082-154,964,109	(CA)n
STR 13	chrX:154,935,982-154,936,024	(AC)n
STR 22	chrX:154,875,737-154,875,774	(GT)n
STR 25 A	chrX:154,850,084-154,850,120	(TG)n
STR 25 B	chrX:154,854,591-154,854,619	(TG)n

Table S4: Primers used for intragenic markers analysis

Primer pair	Sequence of forward primer	Sequence of reverse primer
STR1	TTGCAAATGTACAATAGGGCAGT	TTCTTGGTTCTGCCTTCTGA
STR9	TGGACTTCCAACCCCATAGTC	CACCATGCCTGGCTAATTCA
STR13	TGCATCACTGTACATATGTATCTT	CCAAATTACATATGAATAAGCC
STR22	TTCTAAGAATGTAGTGTGTGTG	TAATGCCCACATTATAGA
STR25A	AGATCGCGCCATCACATTC	AGGGGTAGGCAGGCTTGTTT
STR25B	CAGAACCAATTCCAGAAATCCA	GCTCTCTGGGGTCTCTAGGC
F8_del_IVS13	CTGGAGTGAAATATCTCATTGTGC	AAAATTAGCTGGGTGTGGTGG

Reverse primers are labeled by FAM