

Text S2: Tables of mutagenic TE insertions in introns

Table 1. Mutagenic intronic Alu insertions in humans

Gene with mutation	Alu subfamily	Orientation [#]	Nearest splice-site	Distance to exon (bp)	Related diseases	References
FAS	Sb1	-	SA	50	Autoimmune lymphoproliferative syndrome (ALPS)	[1-2]
FGFR2	Yb8	-	SA	10	Apert Syndrome	[3]
APC	Yb9	-	SD	20	Familial Adenomatous Polyposis	[4]
GK	Ya5	-	SA	30	Glycerol kinase deficiency	[5]
F8	Yb9	-	SA	10	Hemophilia A	[1, 6]
OPA1	Yb8	-	SA	21	Autosomal dominant optic atrophy	[7]

[#] The "-" sign represents antisense orientation of the TE with respect to the enclosing gene

Table 2. Mutagenic intronic L1 insertions in humans

Gene with mutation	Orientation	Nearest splice-site	Distance to exon (bp)	Related diseases	References
HBB	+	SA	100	Beta-thalassemia	[8]
FKTN	+	SA	24	Fukuyama-type congenital muscular dystrophy (FCMD)	[9]
RPS7KA3	-	SA	10	Coffin-Lowry syndrome (CLS)	[10]
CYBB	+	SA	265	Chronic granulomatous disease	[1, 11]
RP2	+	SD	653	X-linked retinitis pigmentosa (XLRP)	[12-13]

[#] The "+"/"-" sign represents sense/antisense orientation of the TE with respect to the enclosing gene

Table 3. Mutagenic intronic ERV insertions in mice

Mutation	ERV family	Orientation [#]	Nearest splice-site*	Distance to exon (bp)	References
<i>Ap3d1</i> ^{mh2j}	IAP	+	SD	6	[14]
<i>Atrn</i> ^{mg}	IAP	-	SD	136	[15]
<i>Atrn</i> ^{mg-L}	IAP	+	SD	~420	[15]
<i>Eya1</i> ^{bor}	IAP	+	SA	1575	[16]
<i>Gus</i> ^{mps2j}	IAP	+	SA	btw. 850-1100	[17]
<i>Lama2</i> ^{pas}	IAP	+	SD	~300	[18]
<i>Lamb3</i> ^{IAP}	IAP	-	SA	1	[19]
<i>Mgrn1</i> ^{md-2j}	IAP	+	SD	616	Ref 20
<i>Mgrn1</i> ^{md}	IAP	+	SA	~942	[20-21]
<i>Pitpna</i> ^{vb}	IAP	+	SA	~1126	[22]
<i>Spna1</i> ^{Dem}	IAP	?	SA	1	[23]
<i>Pofut1</i> ^{cox}	IAP	-	SA	24	[24]
<i>Pmca2</i> ^{loggle}	IAP	+	SD	~15	[25]
<i>Gria4</i> ^{spkw1}	IAP	+	SD	~720	[26]
<i>Zfp69</i> ^{S/L}	IAP	?	SA	965	[27]
<i>Adcy1</i> ^{brl}	ETn ^a	+	SD	~1700	[28-29]
<i>Cacng2</i> ^{stg}	ETn	+	SD	btw. 1500-2100	[30-31]
<i>Cacng2</i> ^{stg-3j}	ETn	+	SD	btw. 2500-4100	[30-31]
<i>Clcn1</i> ^{odr}	ETn	+	SD	1033	[32]
<i>Fas</i> ^{lpr}	ETn	+	SA	3500	[33-35]
<i>Fbxw4</i> ^{Dac-2j}	ETn	+	SD	~14000	[36]
<i>Fign</i> ^{fi}	ETn	+	SD	~60000	[37]
<i>Foxn1</i> ^{nu-Bc}	ETn	-	SA	~5200	[38]
<i>Gli3</i> ^{pdn}	ETn	+	SA	24514	[39]
<i>Hk1</i> ^{dea}	ETn	?	SA	901	[40]
<i>Lep</i> ^{Ob-2j}	ETn	+	SD	~3200	[41]
<i>Mip</i> ^{Cat-Fr}	ETn	+	SA	~800	[42-43]
<i>Muted</i> ^{mu}	ETn	+	SD	2362	[44]
<i>Ttc7</i> ^{sn}	ETn	+	SA	57	[45]
<i>Hsf4</i> ^{lop11}	ETn	+	SA	61	[46]
<i>Fig4</i> ^{paletremor}	ETn	+	SA	384	[47]
<i>Dysf</i> ^{prmd}	ETn	+	SD	495	[48]
<i>Zhx2</i> ^{Afr1}	ETn	+	SA	~20600	[49-50]
<i>a</i>	VL30 ^b	-	SD	~1200	[51]
<i>Abcb1a</i> ^{mds}	MuLV ^c	-	SA	4	[52]
<i>Myo5a</i> ^d	MuLV	+	SD	~500	[53]
<i>Nox3</i> ^{het}	unk ^d	?	SD	~4000	[54]
<i>Pdcd8</i> ^{Hq}	MuLV	+	SD	3432	[55]
<i>Pde6b</i> ^{rd1}	MuLV	-	SD	1511	[56]
<i>Lmf1</i> ^{cid}	MuERV	+	?	<250	[57]

[#] The "+"/"-/?" indicates orientation of the TE with respect to the enclosing gene; "?" indicates orientation is unknown;

*SD-splice donor sites; SA-splice acceptor sites; "?" indicates that the nearest splice site was not given;

^aETn represents the ETn/MusD family; ^bVirus-like 30 element; ^c Murine leukemia virus; ^d"unk" indicates the ERV type was not given.

References for Supporting Tables

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