

Table S3. List of 54 variants that cause de novo 5' or 3' ectopic splice site activation. Variants were obtained from DBASS5 and DBASS3. References can be found in the main text of the paper.

SNP No.	Chr	Position	Gene	Potential Coding effect	Splice site activation	Variant (DNA)	max(Ass)	Reference
1	3	15651990	BTB	Missense	3'	G->A	7.96	[106]
2	4	155708309	FGB	Missense	3'	T->A	8.37	[107]
3	6	31432182	HLA-B	Missense	3'	G->C	9.95	[108]
4	11	107511124	ACAT1	Missense	3'	C->T	2.11	[109]
5	15	41282727	EPB42	Missense	3'	G->T	13.31	[110]
6	19	17860206	SLC5A5	Missense	3'	C->G	8.06	[111]
7	X	32238252	DMD	Nonsense	3'	T->G	8.59	[112]
8	11	67567654	TCIRG1	Synonymous	3'	G->A	7.95	[113]
9	19	11084983	LDLR	Synonymous	3'	C->A	8.04	[114]
10	X	153838609	F8	Synonymous	3'	G->T	2.51	[115]
11	1	154375026	LMNA	Missense	5'	G->A	1.53	[4]
12	4	89215079	PKD2	Nonsense	5'	C->T	0	[116]
13	5	70980772	MCCB	Missense	5'	A->G	8.18	[117]
14	7	91702781	KRIT1	Missense	5'	C->G	1.81	[118]
15	7	91703738	KRIT1	Missense	5'	A->G	8.18	[118]
16	10	89682830	PTEN	Missense	5'	C->G	8.27	[119]
17	11	107708775	ATM	Missense	5'	C->T	7.75	[85]
18	16	67413594	CDH1	Missense	5'	C->T	7.75	[120]
19	17	26551666	NF1	Missense	5'	C->T	7.75	[121]
20	17	26580523	NF1	Missense	5'	G->A	3.81	[122]
21	17	26565668	NF1	Nonsense	5'	A->G	8.18	[122]
22	17	26583297	NF1	Missense	5'	T->A	5.33	[121]
23	17	38512021	BRCA1	Missense	5'	T->G	8.6	[123]
24	17	45619024	COL1A1	Missense	5'	A->G	8.18	[124]
25	X	100516810	BTK	Nonsense	5'	A->G	8.18	[125]
26	X	133436951	HPRT1	Missense	5'	G->T	7.65	[126]
27	X	135560190	CD40L	Nonsense	5'	G->T	4.23	[127]
28	X	148392770	IDS	Missense	5'	C->G	0	[128]
29	X	153235823	FLNA	Missense	5'	C->T	0	[129]
30	1	154375028	LMNA	Synonymous	5'	C->T	0.49	[130]
31	1	153474560	GBA	Synonymous	5'	A->G	8.18	[131]
32	1	156912581	SPTA1	Synonymous	5'	A->G	8.18	[132]
33	2	219382723	CYP27A1	Synonymous	5'	G->T	7.65	[133]
34	3	48593334	COL7A1	Synonymous	5'	C->T	7.75	[134]
35	3	143757971	ATR	Synonymous	5'	A->G	0	[37]
36	3	185449258	ALG3	Synonymous	5'	C->T	0.75	[135]
37	5	42747170	GHR	Synonymous	5'	C->T	7.76	[136]
38	5	42735837	GHR	Synonymous	5'	A->G	4.8	[137]
39	7	65070189	GUSB	Synonymous	5'	C->T	7.76	[138]

40	11	5204749	HBB	Synonymous	5'	G->A	2.24	[139]
41	11	5204753	HBB	Synonymous	5'	T->A	2.28	[140]
42	12	108497085	MVK	Synonymous	5'	C->T	7.75	[141]
43	12	46535733	VDR	Synonymous	5'	C->G	3.5	[142]
44	15	70428488	HEXA	Synonymous	5'	T->A	4.52	[143]
45	16	162980	HBA2	Synonymous	5'	C->T	7.76	[144]
46	16	66027759	HSD11B2	Synonymous	5'	C->G	2.51	[145]
47	17	42731797	ITGB3	Synonymous	5'	C->T	7.76	[146]
48	17	26580468	NF1	Synonymous	5'	G->A	3.51	[121]
49	17	26586877	NF1	Synonymous	5'	C->T	7.75	[121]
50	19	17808957	JAK3	Synonymous	5'	C->T	7.75	[147]
51	19	806755	ELA2	Synonymous	5'	C->A	7.03	[148]
52	X	100500293	BTK	Synonymous	5'	A->G	0	[149]
53	X	153301140	TAZ	Synonymous	5'	G->A	0	[150]
54	X	148376419	IDS	Synonymous	5'	C->T	7.75	[151]