

Table S1 – List of 87 Synonymous and Missense variants that cause exon skipping used for analysis in this study. The variants are located in 40 genes and 47 individual exons. Variant locations are from human genome assembly hg18. References can be found in the main text of the paper.

SNP No.	Chr	Position	Gene	Potential Coding effect	Variant (Protein)	Variant (DNA)	Reference
1	1	196932540	PTPRC	Synonymous	P->P	C->G	[55]
2	2	211181402	CPS1	Synonymous	S->S	C->A	[64]
3	3	37017458	MLH1	Synonymous	D->D	T->C	[65]
4	3	143757998	ATR	Synonymous	E->E	A->G	[37]
5	4	185806724	CASP3	Synonymous (5' UTR)	-	G->A	[53]
6	5	69408148	SMN1	Synonymous	L->L	A->G	[43]
7	5	70283529	SMN2	Synonymous	F->F	C->T	[66]
8	5	112198672	APC	Synonymous	R->R	G->T	[67]
9	7	117017661	CFTR	Synonymous	A->A	T->A	[46]
10	7	117017667	CFTR	Synonymous	L->L	G->A	[46]
11	7	117017682	CFTR	Synonymous	S->S	T->C	[46]
12	7	117017691	CFTR	Synonymous	G->G	A->G	[46]
13	7	117017691	CFTR	Synonymous	G->G	A->T	[46]
14	7	117017694	CFTR	Synonymous	Y->Y	C->T	[46]
15	10	42929995	RET	Synonymous	I->I	C->T	[68]
16	10	114190345	ZDHHC6	Synonymous	T->T	C->T	[53]
17	11	118464207	HMBS	Synonymous	R->R	C->G	[69]
18	14	22444702	RBM23	Synonymous	R->R	G->A	[53]
19	15	46516836	FBN1	Synonymous	I->I	C->T	[70]
20	15	78251635	FAH	Synonymous	N->N	C->T	[71]
21	17	26551622	NF1	Synonymous	Q->Q	G->A	[72]
22	17	26689883	NF1	Synonymous	Y->Y	C->T	[73]
23	17	59349223	GH1	Synonymous	E->E	A->G	[13]
24	19	11088602	LDLR	Synonymous	N->N	C->T	[54]
25	20	44185176	CD40	Synonymous	T->T	A->T	[74]
26	X	19281185	PDHA	Synonymous	Y->Y	C->T	[12]
27	X	19281200	PDHA	Synonymous	I->I	C->T	[12]
28	X	19282574	PDHA	Synonymous	G->G	A->G	[75]
29	X	32276456	DMD	Synonymous	F->F	C->T	[76]
30	X	40341465	XMRE	Synonymous	D->D	C->T	[77]
31	X	53475492	HADH2	Synonymous	R->R	C->A	[78]
32	X	133460368	HPRT1	Synonymous	F->F	C->T	[79]
33	1	75971876	ACADM	Missense	T->I	C->T	[80]
34	2	48774922	LHCGR	Missense	E->K	G->A	[81]
35	3	37017456	MLH1	Missense	N->H	G->C	[65]
36	3	37017456	MLH1	Missense	N->Y	G->T	[65]
37	3	37065091	MLH1	Missense	R->P	G->C	[82]

38	3	37065091	MLH1	Missense	R->L	G->T	[82]
39	4	104025813	CISD2	Missense	E->G	G->C	[83]
40	5	69408148	SMN1	Missense	L->F	A->C	[43]
41	5	69408149	SMN1	Missense	N->H	A->C	[43]
42	5	69408152	SMN1	Missense	X->E	T->G	[43]
43	5	69408154	SMN1	Missense	X->Y	A->T	[43]
44	5	69408154	SMN1	Missense	X->Y	A->C	[43]
45	5	74052072	HEXB	Missense	D->G	A->G	[40]
46	7	117017657	CFTR	Missense	D->G	A->G	[47]
47	10	124802603	ACADSB	Missense	M->V	A->G	[84]
48	11	107692006	ATM	Missense	E->K	G->A	[85]
49	13	31835504	BRCA2	Missense	T->R	C->G	[86]
50	13	47851743	RB1	Missense	G->R	G->A	[87]
51	15	38487132	IVD	Missense	R->C	C->T	[88]
52	15	38487133	IVD	Missense	R->P	G->T	[88]
53	15	38487189	IVD	Missense	D->N	G->A	[88]
54	16	8812504	PMM2	Missense	E->K	G->A	[89]
55	17	4745668	CHRNE	Missense	F->V	T->G	[90]
56	17	26521112	NF1	Missense	D->V	A->T	[91]
57	17	26551623	NF1	Missense	L->M	C->A	[72]
58	17	38469489	BRCA1	Missense	E->K	G->A	[92]
59	17	59349200	GH1	Missense	K->R	A->G	[93]
60	17	59349202	GH1	Missense	Q->H	G->T	[93]
61	17	59349205	GH1	Missense	E->D	A->T	[93]
62	17	59349206	GH1	Missense	E->V	A->T	[93]
63	17	59349223	GH1	Missense	E->D	A->C	[13]
64	17	59349224	GH1	Missense	E->G	A->G	[13]
65	17	59349225	GH1	Missense	E->Q	G->C	[13]
66	20	42685097	ADA	Missense	A->T	G->A	[94]
67	X	19282542	PDHA1	Missense	A->T	G->A	[95]
68	X	19282611	PDHA1	Missense	A->T	G->A	[96]
69	X	32276455	DMD	Missense	N->Y	A->T	[76]
70	X	32276456	DMD	Missense	F->L	C->G	[76]
71	X	32276456	DMD	Missense	F->L	C->A	[76]
72	X	32276457	DMD	Missense	F->Y	T->A	[76]
73	X	32276458	DMD	Missense	F->I	T->A	[76]
74	X	77184841	ATP7A	Missense	G->R	G->A	[97]
75	X	102928267	PLP1	Missense	R->W	C->T	[98]
76	X	102928267	PLP1	Missense	R->G	C->G	[98]
77	X	133435146	HPRT1	Missense	G->V	G->T	[99]
78	X	133436885	HPRT1	Missense	R->H	G->A	[99]
79	X	133455283	HPRT1	Missense	A->E	C->A	[99]
80	X	133460309	HPRT1	Missense	G->R	G->A	[100]
81	X	133460310	HPRT1	Missense	G->V	G->T	[100]
82	X	133460315	HPRT1	Missense	E->K	G->A	[99]

83	X	133460322	HPRT1	Missense	P->L	C->T	[79]
84	X	133460351	HPRT1	Missense	D->Y	G->T	[99]
85	X	133460361	HPRT1	Missense	E->V	A->T	[99]
86	X	133460373	HPRT1	Missense	D->V	A->T	[101]
87	X	153783589	F8	Missense	R->W	C->T	[102]