

Skippy reference list: Published exon-skipping variants (hg18)

1	1	196932540	PTPRC	Synonymous	P->P	C->G	(1)
2	2	211181402	CPS1	Synonymous	S->S	C->A	(2)
3	3	37017458	MLH1	Synonymous	D->D	T->C	(3)
4	3	143757998	ATR	Synonymous	E->E	A->G	(4)
5	4	185806724	CASP3	Synonymous	(5' UTR)	G->A	(5)
6	5	69408148	SMN1	Synonymous	L->L	A->G	(6)
7	5	70283529	SMN2	Synonymous	F->F	C->T	(7)
8	5	112198672	APC	Synonymous	R->R	G->T	(8)
9	7	117017661	CFTR	Synonymous	A->A	T->A	(9)
10	7	117017667	CFTR	Synonymous	L->L	G->A	(9)
11	7	117017682	CFTR	Synonymous	S->S	T->C	(9)
12	7	117017691	CFTR	Synonymous	G->G	A->G	(9)
13	7	117017691	CFTR	Synonymous	G->G	A->T	(9)
14	7	117017694	CFTR	Synonymous	Y->Y	C->T	(9)
15	10	42929995	RET	Synonymous	I->I	C->T	(10)
16	10	114190345	ZDHC6	Synonymous	T->T	C->T	(5)
17	11	118464207	HMBS	Synonymous	R->R	C->G	(11)
18	14	22444702	RBM23	Synonymous	R->R	G->A	(5)
19	15	46516836	FBN1	Synonymous	I->I	C->T	(12)
20	15	78251635	FAH	Synonymous	N->N	C->T	(13)
21	17	26551622	NF1	Synonymous	Q->Q	G->A	(14)
22	17	26689883	NF1	Synonymous	Y->Y	C->T	(15)
23	17	59349223	GH1	Synonymous	E->E	A->G	(16)
24	19	11088602	LDLR	Synonymous	N->N	C->T	(17)
25	20	44185176	CD40	Synonymous	T->T	A->T	(18)
26	X	19281185	PDHA	Synonymous	Y->Y	C->T	(19)
27	X	19281200	PDHA	Synonymous	I->I	C->T	(19)
28	X	19282574	PDHA	Synonymous	G->G	A->G	(20)
29	X	32276456	DMD	Synonymous	F->F	C->T	(21)
30	X	40341465	XMRE	Synonymous	D->D	C->T	(22)
31	X	53475492	HADH2	Synonymous	R->R	C->A	(23)
32	X	133460368	HPRT1	Synonymous	F->F	C->T	(24)
33	1	75971876	ACADM	Missense	T->I	C->T	(25)
34	2	48774922	LHCGR	Missense	E->K	G->A	(26)
35	3	37017456	MLH1	Missense	N->H	G->C	(3)
36	3	37017456	MLH1	Missense	N->Y	G->T	(3)
37	3	37065091	MLH1	Missense	R->P	G->C	(27)
38	3	37065091	MLH1	Missense	R->L	G->T	(27)
39	4	104025813	CISD2	Missense	E->G	G->C	(28)
40	5	69408148	SMN1	Missense	L->F	A->C	(6)
41	5	69408149	SMN1	Missense	N->H	A->C	(6)
42	5	69408152	SMN1	Missense	X->E	T->G	(6)
43	5	69408154	SMN1	Missense	X->Y	A->T	(6)
44	5	69408154	SMN1	Missense	X->Y	A->C	(6)
45	5	74052072	HEXB	Missense	D->G	A->G	(29)
46	7	117017657	CFTR	Missense	D->G	A->G	(30)
47	10	124802603	ACADSB	Missense	M->V	A->G	(31)
48	11	107692006	ATM	Missense	E->K	G->A	(32)
49	13	31835504	BRCA2	Missense	T->R	C->G	(33)
50	13	47851743	RB1	Missense	G->R	G->A	(34)
51	15	38487132	IVD	Missense	R->C	C->T	(35)
52	15	38487133	IVD	Missense	R->P	G->T	(35)
53	15	38487189	IVD	Missense	D->N	G->A	(35)
54	16	8812504	PMM2	Missense	E->K	G->A	(36)
55	17	4745668	CHRNE	Missense	F->V	T->G	(37)
56	17	26521112	NF1	Missense	D->V	A->T	(38)
57	17	26551623	NF1	Missense	L->M	C->A	(14)
58	17	38469489	BRCA1	Missense	E->K	G->A	(39)
59	17	59349200	GH1	Missense	K->R	A->G	(40)
60	17	59349202	GH1	Missense	Q->H	G->T	(40)
61	17	59349205	GH1	Missense	E->D	A->T	(40)
62	17	59349206	GH1	Missense	E->V	A->T	(40)
63	17	59349223	GH1	Missense	E->D	A->C	(16)

64	17	59349224	GHI	Missense	E->G	A->G	(16)
65	17	59349225	GHI	Missense	E->Q	G->C	(16)
66	20	42685097	ADA	Missense	A->T	G->A	(41)
67	X	19282542	PDHA1	Missense	A->T	G->A	(42)
68	X	19282611	PDHA1	Missense	A->T	G->A	(43)
69	X	32276455	DMD	Missense	N->Y	A->T	(21)
70	X	32276456	DMD	Missense	F->L	C->G	(21)
71	X	32276456	DMD	Missense	F->L	C->A	(21)
72	X	32276457	DMD	Missense	F->Y	T->A	(21)
73	X	32276458	DMD	Missense	F->I	T->A	(21)
74	X	77184841	ATP7A	Missense	G->R	G->A	(44)
75	X	102928267	PLP1	Missense	R->W	C->T	(45)
76	X	102928267	PLP1	Missense	R->G	C->G	(45)
77	X	133435146	HPRT1	Missense	G->V	G->T	(46)
78	X	133436885	HPRT1	Missense	R->H	G->A	(46)
79	X	133455283	HPRT1	Missense	A->E	C->A	(46)
80	X	133460309	HPRT1	Missense	G->R	G->A	(47)
81	X	133460310	HPRT1	Missense	G->V	G->T	(47)
82	X	133460315	HPRT1	Missense	E->K	G->A	(47)
83	X	133460322	HPRT1	Missense	P->L	C->T	(24)
84	X	133460351	HPRT1	Missense	D->Y	G->T	(46)
85	X	133460361	HPRT1	Missense	E->V	A->T	(46)
86	X	133460373	HPRT1	Missense	D->V	A->T	(48)
87	X	153783589	F8	Missense	R->W	C->T	(49)

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