Skippy reference: Neutral variants (hg18)

SNP No.	Chr	Position	Gene	Potential Coding Effect	Mutation (Protein)	Mutation (DNA)	Reference
1	21	21668058	NCAM2	Missense	L->P	T->C	(1)
2	5	13882799	DNAH5	Synonymous	A->A	C->T	(1)
3	12	103233689	TXNRD1	Synonymous	L->L	C->T	(1)
4	1	181451239	LAMC2	Synonymous	S->S	C->T	(1)
5	1	27552384	SYTL1	Missense	Q->R	A->G	(1)
6	2	98516378	INPP4A	Synonymous	T->T	G->A	(1)
7	2	24378462	ITSN2	Missense	V->I	G->A	(1)
8	12	27758994	MRPS35	Missense	G->R	G->A	(1)
9	4	9531265	SLC2A9	Missense	V->I	G->A	(1)
10	4	96325345	UNC5C	Missense	M->T	T->C	(1)
11	15	84607033	AGBL1	Synonymous	D->D	C->T	(1)
12	1	17174367	MFAP2	Synonymous	H->H	T->C	(1)
13	2	218976025	CTDSP1	Synonymous	H->H	C->T	(1)
14	16	83480360	CRISPLD2	Synonymous	A->A	C->T	(1)
15	18	75574115	CTDP1	Missense	T->M	C->T	(1)
16	5	38406294	EGFLAM	Missense	W->R	T->C	(1)
17	12	108102248	ACACB	Synonymous	D->D	T->C	(1)
18	22	29187448	SEC14L3	Synonymous	D->D	T->C	(1)
19	12	116154297	NOS1	Synonymous	D->D	C->T	(1)
20	11	68462250	IGHMBP2	Missense	T->K	C->A	(1)
21	2	160516321	PLA2R1	Missense	G->S	G->A	(1)
22	7	158520087	VIPR2	Synonymous	N->N	C->T	(1)
23	7	21565758	DNAH11	Synonymous	N->N	C->T	(1)
24	17	68922486	SDK2	Synonymous	T->T	G->A	(1)
25	10	102255805	SEC31B	Synonymous	H->H	T->C	(1)
26	17	70121684	CD300E	Missense	G->R	G->A	(1)
27	15	39953792	SPTBN5	Missense	R->H	G->A	(1)
28	4	154733172	KIAA0922	Synonymous	H->H	C->T	(1)
29	3	133701313	DNAJC13	Missense	A->S	G->T	(1)
30	2	200962201	LOC26010	Synonymous	(5' UTR)	A->G	(1)
31	5	41036100	AC114967.2	Synonymous	Y->Y	C->T	(1)
32	12	47351994	C12orf41	Synonymous	T->T	A->G	(1)
33	10	113925369	GPAM	Missense	E->G	A->G	(1)
34	3	113403806	SLC9A10	Missense	S->I	G->T	(1)
35	3	155501581	DHX36	Synonymous	S->S	G->C	(1)
36	4	87925530	PTPN13	Missense	Y->D	T->G	(1)
37	15	38443137	DISP2	Missense	P->A	C->G	(1)
38	9	83456967	TLE1	Synonymous	E->E	A->G	(1)
39	X	14797068	FANCB	Synonymous	(5' UTR)	C->G	(1)
40	6	41106119	UNC5CL	Synonymous	C->C	C->T	(1)
41	11	60422917	PRPF19	Synonymous	S->S	C->T	(1)
42	11	85646271	EED	Synonymous	L->L	C->T	(1)
43	3	37042476	MLH1	Missense	K->N	G->T	(2)
44	3	37042191	MLH1	Synonymous	L->L	G->T	(2)
45	3	37042232	MLH1	Synonymous	A->A	C->T	(2)
46	3	37042239	MLH1	Missense	Q->H	G->C	(2)
47	3	37042240	MLH1	Missense	M->L	A->T	(2)
48	3	37042297	MLH1	Missense	K->Q	A->C	(2)
49	3	37042335	MLH1	Missense	E->D	G->C	(2)
50	3	37042363	MLH1	Missense	A->S	G->C	(2)
51	3	37042376	MLH1	Missense	D->V	A->T	(2)
52	3	37042406	MLH1	Missense	A->V	C->T	(2)
53	3	37042432	MLH1	Synonymous	L->L	T->C	(2)
54	3	37042454	MLH1	Missense	G->G	G->C	(2)
55	7	116975945	CFTR	Missense	L->F	A->C	(3)
56	7	116975973	CFTR	Missense	N->D	A->G	(3)
57	7	116976002	CFTR	Missense	S->R	C->A	(3)

58	7	116976048	CFTR	Missense	D->N	G->A	(3)
59	7	116976081	CFTR	Missense	L->M	T->A	(3)
60	7	116976081	CFTR	Synonymous	L->L	T->C	(3)
61	7	116976083	CFTR	Missense	L->F	G->C	(3)
62	7	116976087	CFTR	Missense	V->F	G->T	(3)
63	17	26689881	NF1	Missense	Y->N	T->A	(4)
64	17	26689881	NF1	Missense	Y->H	T->C	(4)
65	17	26689881	NF1	Missense	Y->D	T->G	(4)
66	17	26689882	NF1	Missense	Y->S	A->C	(4)
67	17	26689882	NF1	Missense	Y->C	A->G	(4)
68	17	26689882	NF1	Missense	Y->F	A->T	(4)
69	17	26689884	NF1	Missense	N->H	A->C	(4)
70	17	26689884	NF1	Missense	N->D	A->G	(4)
71	17	26689884	NF1	Missense	N->Y	A->T	(4)
72	17	26689885	NF1	Missense	N->T	A->C	(4)
73	17	26689885	NF1	Missense	N->S	A->G	(4)
74	17	26689885	NF1	Missense	N->I	A->T	(4)
75	17	26689887	NF1	Missense	S->R	A->C	(4)
76	17	26689887	NF1	Missense	S->G	A->G	(4)
77	17	26689887	NF1	Missense	S->C	A->T	(4)
78	17	26689888	NF1	Missense	S->N	G->A	(4)
79	17	26689888	NF1	Missense	S->I	G->T	(4)
80	17	26689888	NF1	Missense	S->T	G->C	(4)

1. Elsharawy, A., Hundrieser, B., Brosch, M., Wittig, M., Huse, K., Platzer, M., Becker, A., Simon, M., Rosenstiel, P., Schreiber, S. *et al.* (2009). Systematic evaluation of the effect of common SNPs on pre-mRNA splicing. Hum Mutat, *30*, 625 - 632.
2. Lastella, P., Resta, N., Miccolis, I., Quagliarella, A., Guanti, G. and Stella, A. (2004). Site directed mutagenesis of hMLH1 exonic splicing enhancers does not correlate with splicing disruption. J Med Genet, *41*, e72.
3. Pagani, F., Buratti, E., Stuani, C. and Baralle, F. (2003). Missense, nonsense, and neutral mutations define juxtaposed regulatory elements of splicing in cystic fibrosis transmembrane regulator exon 9. J Biol Chem, *278*, 26580 - 26588.
4. Baralle, M., Skoko, N., Knezevich, A., De Conti, L., Motti, D., Bhuvanagiri, M., Baralle, D., Buratti, E. and Baralle, F. (2006). NF1 mRNA biogenesis: effect of the genomic milieu in splicing regulation of the NF1 exon 37 region. FEBS Lett, *580*, 4449 - 4456.