Skippy Reference: Variants causing exon inclusion (hg18)

SNP	Chr	Position	Gene	Coding effect	Variant (Protein)	Variant (DNA)	Reference
1	5	70283543	**SMN1**	Missense	K->I	A->T	**(1)**
2	5	70283560	**SMN1**	Missense	H->T	C->T	**(1)**
3	5	70283570	**SMN1**	Missense	N->T	A->C	**(1)**
4	7	116975948	**CFTR**	Missense	F->L	T->G	**(2)**
5	7	116976075	**CFTR**	Missense	Q->G	C->G	**(2)**
6	7	116976075	**CFTR**	Missense	Q->L	C->A	**(2)**
7	7	116976076	**CFTR**	Missense	Q->R	A->G	**(2)**
8	7	116976076	**CFTR**	Missense	Q->L	A->T	**(2)**
9	7	116976076	**CFTR**	Missense	Q->P	A->C	**(2)**
10	7	116976077	**CFTR**	Missense	Q->H	G->T	**(2)**
11	7	116976077	**CFTR**	Missense	Q->H	G->C	**(2)**
12	7	116976078	**CFTR**	Synonymous	L->L	T->C	**(2)**
13	7	116976078	**CFTR**	Missense	L->M	T->A	**(2)**
14	7	117017676	**CFTR**	Synonymous	L->L	A->G	**(3)**
15	7	117017679	**CFTR**	Synonymous	N->N	C->T	**(3)**
16	7	117017682	**CFTR**	Synonymous	S->S	T->G	**(3)**
17	7	117017685	**CFTR**	Synonymous	P->P	T->A	**(3)**
18	17	41443542	**MAPT**	Synonymous	L->L	T->C	**(4)**
19	17	41443527	**MAPT**	Missense	N->K	T->G	**(4)**
20	17	41443578	**MAPT**	Synonymous	N->N	T->C	**(5)**

1. Singh, N., Androphy, E. and Singh, R. (2004). In vivo selection reveals combinatorial controls that define a critical exon in the spinal muscular atrophy genes. Rna, *10*, 1291 - 1305.
2. 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.
3. Pagani, F., Raponi, M. and Baralle, F. (2005). Synonymous mutations in CFTR exon 12 affect splicing and are not neutral in evolution. Proc Natl Acad Sci USA, *102*, 6368 - 6372.
4. D'Souza, I., Poorkaj, P., Hong, M., Nochlin, D., Lee, V., Bird, T. and Schellenberg, G. (1999). Missense and silent tau gene mutations cause frontotemporal dementia with parkinsonism-chromosome 17 type, by affecting multiple alternative RNA splicing regulatory elements. Proc Natl Acad Sci USA, *96*, 5598 - 5603.
5. Lee, V., Goedert, M. and Trojanowski, J. (2001). Neurodegenerative tauopathies. Annu Rev Neurosci, *24*, 1121 - 1159.