

Validation

varSEAK Online

Splice Site Prediction

Version 2.1

developed by

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1 Validation

For the validation of the varSEAK Splice Site Prediction the data set of Leman *et al.* (2018) was used.

The data set contains 395 variants with known splice effects.

The overall splice effect is indicated by YES/NO.

Additionally, the detailed effect on splicing is given in the column "Observation".

Of the 395 variants, 5 are GC 5' splice sites. As these do not behave like normal GT splice sites, we did not analyze them (yet).

Hence, the number of splice sites actually evaluated with the varSEAK algorithm is 390.

GC 5' splice sites possibly influenced by variants were generally rated as class 3.

For evaluation, we defined the following rules:

Splice Effect YES: Classes 4 and 5 are correctly predicted, all other classes are wrong.

Splice Effect NO: Classes 1 and 2 are correctly predicted, all other classes are wrong.

This results in the following statistics:

Total:

Analyzed Variants	:	390 / 395
Correct Prediction	:	376 / 390 (96.41 %)
False Prediction	:	14 / 390 (3.59 %)
GC at 5' SS	:	5

Positive:

Positives	:	299 / 390 (76.67 %)
True positives	:	292 / 299 (97.66 %)
False negatives	:	7 / 299 (2.34 %)

Negative:

Negatives	:	91 / 390 (23.33 %)
True negatives	:	84 / 91 (92.31 %)
False positives	:	7 / 91 (7.69 %)

2 List of variants in the data set

The following table lists the 395 tested variants.

For each, gene, transcript, and variant are given.

The column "JSI Class" presents the class calculated by the varSEAK algorithm:

- 1 = no splicing effect
- 2 = likely no splicing effect
- 3 = unknown splicing effect
- 4 = likely splicing effect
- 5 = splicing effect

A '*' (asterisk) in front of the class marks wrong evaluations.

A '-' (minus) in front of the class marks unevaluated 5' GC sites.

The column "Effect" contains the overall splicing effect (YES/NO) as specified by Leman *et al.*

The column "Observation" contains the detailed splicing effect of the variant as specified by Leman *et al.*

The variants are linked to varSEAK.bio.

To see details of the analysis please click on the respective link.

	Gene	Transcript	Variant	JSI Class	Effect	Observation
1	BRCA1	NM_007294.3	c.133A>C	2	NO	No change
2	BRCA1	NM_007294.3	c.134+1G>A	5	YES	Exon 3 skipped
3	BRCA1	NM_007294.3	c.134+3A>C	5	YES	Exon 3 skipped
4	BRCA1	NM_007294.3	c.211A>G	5	YES	Exon 5 skipped and use of a cryptic site 22 nt upstream from 5' ss
5	BRCA1	NM_007294.3	c.211dup	1	NO	No change
6	BRCA1	NM_007294.3	c.212+1G>A	5	YES	Use of a cryptic site 22 nt upstream from 5' ss
7	BRCA1	NM_007294.3	c.212+1G>T	5	YES	Use of a cryptic site 22 nt upstream from 5' ss
8	BRCA1	NM_007294.3	c.212+2T>C	5	YES	Use of a cryptic site 22 nt upstream from 5' ss
9	BRCA1	NM_007294.3	c.212+3A>G	5	YES	Use of a cryptic site 22 nt upstream from 5' ss
10	BRCA1	NM_007294.3	c.212G>A	5	YES	Use of a cryptic site 22 nt upstream from 5' ss
11	BRCA1	NM_007294.3	c.213-11T>G	5	YES	Use of a cryptic site 59 nt upstream from 3' ss
12	BRCA1	NM_007294.3	c.213-2A>C	5	YES	Use of a cryptic site 59 nt upstream from 3' ss
13	BRCA1	NM_007294.3	c.213-2A>G	5	YES	Use of a cryptic site 59 nt upstream from 3' ss
14	BRCA1	NM_007294.3	c.301+1G>C	5	YES	Use of a cryptic site 9 nt upstream from 5' ss
15	BRCA1	NM_007294.3	c.301+7G>A	1	NO	No change
16	BRCA1	NM_007294.3	c.302-5T>C	1	NO	No change
17	BRCA1	NM_007294.3	c.4097-11T>C	1	NO	No change
18	BRCA1	NM_007294.3	c.4097-2A>G	5	YES	Use of a cryptic site 1 nt upstream from 3' ss
19	BRCA1	NM_007294.3	c.4185+3A>T	5	YES	Exon 12 skipped
20	BRCA1	NM_007294.3	c.4185G>C	5	YES	Exon 12 skipped
21	BRCA1	NM_007294.3	c.4357+1dup	5	YES	Use of a cryptic site 1 nt downstream from 3' ss
22	BRCA1	NM_007294.3	c.4358-10C>T	1	NO	No change
23	BRCA1	NM_007294.3	c.442-3T>G	4	YES	Use of a cryptic site 3 nt downstream from 5' ss
24	BRCA1	NM_007294.3	c.4484+1G>A	5	YES	Exon 14 skipped
25	BRCA1	NM_007294.3	c.4484G>A	4	YES	Exon 14 skipped
26	BRCA1	NM_007294.3	c.4484G>T	5	YES	Exon 14 skipped
27	BRCA1	NM_007294.3	c.4485-1G>C	5	YES	Exon 15 skipped
28	BRCA1	NM_007294.3	c.4675G>C	5	YES	Exon 15 skipped
29	BRCA1	NM_007294.3	c.4986+3G>C	5	YES	Use of a cryptic site 65 nt downstream from 5' ss
30	BRCA1	NM_007294.3	c.4986+6T>C	4	YES	Use of a cryptic site 65 nt downstream from 5' ss
31	BRCA1	NM_007294.3	c.4987-4del	5	YES	Exon 17 skipped
32	BRCA1	NM_007294.3	c.4987-5T>A	5	YES	Exon 17 skipped
33	BRCA1	NM_007294.3	c.4987-6T>C	5	YES	Exon 17 skipped
34	BRCA1	NM_007294.3	c.5072C>G	1	NO	No change
35	BRCA1	NM_007294.3	c.5074+1G>A	5	YES	Exon 17 skipped and use of a cryptic site 153 nt downstream from 5' ss
36	BRCA1	NM_007294.3	c.5074+5A>T	1	NO	No change
37	BRCA1	NM_007294.3	c.5074+6C>G	1	NO	No change
38	BRCA1	NM_007294.3	c.5074G>C	5	YES	Exon 17 skipped and use of a cryptic site 153 nt downstream from 5' ss
39	BRCA1	NM_007294.3	c.5075-2A>G	5	YES	Exon 18 skipped
40	BRCA1	NM_007294.3	c.5075-6C>A	2	NO	No change
41	BRCA1	NM_007294.3	c.5075-8T>G	5	YES	Use of a cryptic site 7 nt upstream from 3' ss
42	BRCA1	NM_007294.3	c.5075-9A>T	1	NO	No change
43	BRCA1	NM_007294.3	c.5153-6C>A	2	NO	No change
44	BRCA1	NM_007294.3	c.5193+2del	5	YES	Exon 19 skipped and Exon 18 and 19 skipped
45	BRCA1	NM_007294.3	c.5194-6T>C	1	NO	No change
46	BRCA1	NM_007294.3	c.5278-1G>C	5	YES	Exon 21 skipped
47	BRCA1	NM_007294.3	c.5278-1G>T	5	YES	Exon 21 skipped and use of a cryptic site 8 nt downstream from 3' ss
48	BRCA1	NM_007294.3	c.5331_5332+1 delinsCAACAT	5	YES	Exon 21 skipped
49	BRCA1	NM_007294.3	c.5332+2T>A	5	YES	Exon 21 skipped
50	BRCA1	NM_007294.3	c.5332G>A	4	YES	Exon 21 skipped
51	BRCA1	NM_007294.3	c.5333-3T>G	5	YES	Exon 22 skipped
52	BRCA1	NM_007294.3	c.5334T>C	2	NO	No change
53	BRCA1	NM_007294.3	c.5406+1G>A	5	YES	Exon 22 skipped
54	BRCA1	NM_007294.3	c.5406+2T>G	5	YES	Exon 22 skipped
55	BRCA1	NM_007294.3	c.5406+5G>C	5	YES	Exon 22 skipped
56	BRCA1	NM_007294.3	c.5406+8T>C	1	NO	No change

57	BRCA1	NM_007294.3	c.5467+1G>A	5	YES	Exon 23 skipped
58	BRCA1	NM_007294.3	c.5467+2T>C	5	YES	Exon 23 skipped
59	BRCA1	NM_007294.3	c.5468-10C>A	2	NO	No change
60	BRCA1	NM_007294.3	c.5468-2A>G	5	YES	Use of a cryptic site 11 nt downstream from 3' ss
61	BRCA1	NM_007294.3	c.5468-5T>A	2	NO	No change
62	BRCA1	NM_007294.3	c.5468-8G>A	1	NO	No change
63	BRCA1	NM_007294.3	c.547+2T>A	5	YES	Exon 8 skipped
64	BRCA1	NM_007294.3	c.547+3A>T	5	YES	Exon 8 skipped
65	BRCA1	NM_007294.3	c.547+7G>A	1	NO	No change
66	BRCA1	NM_007294.3	c.591C>T	4	YES	Exon 9 and exon 9 and 10 skipped
67	BRCA1	NM_007294.3	c.593+3G>A	1	NO	No change
68	BRCA1	NM_007294.3	c.670+8C>T	2	NO	No change
69	BRCA1	NM_007294.3	c.81-12C>G	2	NO	No change
70	BRCA1	NM_007294.3	c.81-12del	1	NO	No change
71	BRCA1	NM_007294.3	c.81-1G>C	5	YES	Use of a cryptic site 7 nt downstream from 3' ss
72	BRCA1	NM_007294.3	c.81-6T>C	1	NO	No change
73	BRCA2	NM_000059.3	c.316+5G>C	5	YES	Exon 3 skipped
74	BRCA2	NM_000059.3	c.316+6T>C	* 2	YES	Exon 3 skipped
75	BRCA2	NM_000059.3	c.317-12G>A	1	NO	No change
76	BRCA2	NM_000059.3	c.-39-1G>A	5	YES	Exon 2 and exon 2and 3 skipped
77	BRCA2	NM_000059.3	c.-39-5del	1	NO	No change
78	BRCA2	NM_000059.3	c.426-2A>T	5	YES	Exon 5 skipped
79	BRCA2	NM_000059.3	c.475+3A>G	5	YES	Exon 5 skipped
80	BRCA2	NM_000059.3	c.475+3A>T	5	YES	Exon 5 skipped
81	BRCA2	NM_000059.3	c.475+8dup	1	NO	No change
82	BRCA2	NM_000059.3	c.476-3C>A	4	YES	Exon 5 and 6 skipped
83	BRCA2	NM_000059.3	c.476-9dup	1	NO	No change
84	BRCA2	NM_000059.3	c.516+2T>C	5	YES	Exon 6 skipped
85	BRCA2	NM_000059.3	c.516+4_516+5del	4	YES	Exon 6 and exon 5 and 6 skipped
86	BRCA2	NM_000059.3	c.517-2A>G	5	YES	Exon 7 skipped
87	BRCA2	NM_000059.3	c.631+3A>G	4	YES	Exon 7 skipped
88	BRCA2	NM_000059.3	c.631G>A	5	YES	Exon 7 skipped
89	BRCA2	NM_000059.3	c.632-3C>G	5	YES	Use of a cryptic site 2 nt upstream from 3' ss
90	BRCA2	NM_000059.3	c.632-9A>G	1	NO	No change
91	BRCA2	NM_000059.3	c.633C>T	1	NO	No change
92	BRCA2	NM_000059.3	c.67+1G>T	5	YES	Exon 2 skipped
93	BRCA2	NM_000059.3	c.67+2T>C	5	YES	Exon 2 skipped
94	BRCA2	NM_000059.3	c.67+3A>G	5	YES	Exon 2 skipped and exon 2 and 3 skipped
95	BRCA2	NM_000059.3	c.67+4T>C	1	NO	No change
96	BRCA2	NM_000059.3	c.67G>A	5	YES	Use of a cryptic site 4 nt upstream from 5' ss
97	BRCA2	NM_000059.3	c.681+4A>G	5	YES	Use of a cryptic site 4 nt downstream from 5' ss
98	BRCA2	NM_000059.3	c.681+7A>T	1	NO	No change
99	BRCA2	NM_000059.3	c.682-12del	1	NO	No change
100	BRCA2	NM_000059.3	c.682-4G>C	2	NO	No change
101	BRCA2	NM_000059.3	c.682-9T>C	1	NO	No change
102	BRCA2	NM_000059.3	c.6841+1del	5	YES	Exon 11 skipped
103	BRCA2	NM_000059.3	c.68-7del	1	NO	No change
104	BRCA2	NM_000059.3	c.68-7T>A	5	YES	Exon 3 skipped
105	BRCA2	NM_000059.3	c.6935A>T	* 4	NO	No change
106	BRCA2	NM_000059.3	c.7006C>T	2	NO	No change
107	BRCA2	NM_000059.3	c.7007+1G>C	5	YES	Exon 13 and exon 12 and 13 skipped
108	BRCA2	NM_000059.3	c.7007G>A	5	YES	Exon 13 and exon 12 and 13 skipped
109	BRCA2	NM_000059.3	c.7007G>C	5	YES	Exon 12 and 13 skipped
110	BRCA2	NM_000059.3	c.7008-2A>T	5	YES	Use of a cryptic site 10 nt upstream from 3' ss
111	BRCA2	NM_000059.3	c.7435+6G>A	1	NO	No change
112	BRCA2	NM_000059.3	c.7436-2A>G	5	YES	Use of a cryptic site 13 nt downstream from 3' ss
113	BRCA2	NM_000059.3	c.7436-2A>T	5	YES	Use of a cryptic site 13 nt downstream from 3' ss
114	BRCA2	NM_000059.3	c.7436-4A>T	1	NO	No change

115	BRCA2	NM_000059.3	c.7617+1G>T	5	YES	Exon 15 skipped
116	BRCA2	NM_000059.3	c.7617G>A	* 4	NO	No change
117	BRCA2	NM_000059.3	c.7805+6C>G	1	NO	No change
118	BRCA2	NM_000059.3	c.7805G>C	4	YES	Exon 16 skipped and use of a cryptic site 10 nt upstream from 3' ss
119	BRCA2	NM_000059.3	c.7806-2A>G	5	YES	Use of a cryptic site 20 nt downstream from 3' ss
120	BRCA2	NM_000059.3	c.7807G>C	1	NO	No change
121	BRCA2	NM_000059.3	c.793+1G>C	5	YES	Exon 9 skipped
122	BRCA2	NM_000059.3	c.794-11T>C	1	NO	No change
123	BRCA2	NM_000059.3	c.7975A>G	- 3	YES	Exon 17 skipped and exon 17 and 18 skipped
124	BRCA2	NM_000059.3	c.7976G>C	- 3	YES	Exon 17 skipped
125	BRCA2	NM_000059.3	c.7977-6T>G	* 2	YES	Exon 18 skipped
126	BRCA2	NM_000059.3	c.7977-7C>G	5	YES	Exon 18 skipped and use of cryptic site 7 nt upstream from 3' ss
127	BRCA2	NM_000059.3	c.8486A>G	4	YES	Exon 19 skipped
128	BRCA2	NM_000059.3	c.8487+2T>C	5	YES	Exon 19 skipped
129	BRCA2	NM_000059.3	c.8487+3A>G	5	YES	Exon 19 skipped
130	BRCA2	NM_000059.3	c.8487G>C	5	YES	Exon 19 skipped
131	BRCA2	NM_000059.3	c.8754+4A>G	4	YES	Use of a cryptic site 47 nt downstream from 5' ss
132	BRCA2	NM_000059.3	c.8754G>C	5	YES	Use of a cryptic site 46 nt downstream from 5' ss
133	BRCA2	NM_000059.3	c.8953+2T>G	5	YES	Exon 22 skipped
134	BRCA2	NM_000059.3	c.8954-5A>G	5	YES	Use of a cryptic site 4 nt upstream from 3' ss
135	BRCA2	NM_000059.3	c.9116C>T	1	NO	No change
136	BRCA2	NM_000059.3	c.9117+2T>C	5	YES	Exon 23 skipped
137	BRCA2	NM_000059.3	c.9117G>A	5	YES	Exon 23 skipped
138	BRCA2	NM_000059.3	c.9118-4_9118-2_delinsTTT	5	YES	Use of a cryptic site 7 nt downstream from 3' ss
139	BRCA2	NM_000059.3	c.9257-10dup	1	NO	No change
140	BRCA2	NM_000059.3	c.9501+3A>T	5	YES	Exon 25 skipped
141	BRCA2	NM_000059.3	c.9501+4A>G	* 5	NO	No change
142	BRCA2	NM_000059.3	c.9502-12T>G	* 2	YES	Exon 26 skipped
143	BRCA1	NM_007294.3	c.132C>T	5	YES	Use of a cryptic site 4 nt upstream from 5' ss
144	BRCA1	NM_007294.3	c.133_133+2del	5	YES	exon 3 skipped
145	BRCA1	NM_007294.3	c.134+3_134+6del	5	YES	exon 3 skipped
146	BRCA1	NM_007294.3	c.135-15_135-12del	1	NO	No change
147	BRCA1	NM_007294.3	c.135-1G>C	5	YES	exon 5 skipped
148	BRCA1	NM_007294.3	c.135-6T>G	5	YES	exon 5 skipped
149	BRCA1	NM_007294.3	c.-19-10T>C	* 5	NO	No change
150	BRCA1	NM_007294.3	c.212+1G>C	5	YES	Use of a cryptic site 22 nt upstream from 5' ss
151	BRCA1	NM_007294.3	c.213-12A>G	5	YES	Use of a cryptic site 11 nt upstream from 3' ss
152	BRCA1	NM_007294.3	c.213-1G>A	5	YES	Use of a cryptic site 59 nt upstream from 3' ss
153	BRCA1	NM_007294.3	c.301+6T>C	4	YES	Use of a cryptic site 9 nt upstream from 5' ss
154	BRCA1	NM_007294.3	c.302-1G>A	5	YES	exon 7 skipped
155	BRCA1	NM_007294.3	c.302-2del	5	YES	Use of a cryptic site 10 nt downstream from 3' ss
156	BRCA1	NM_007294.3	c.302-3C>G	5	YES	Use of a cryptic site 2 nt upstream from 3' ss
157	BRCA1	NM_007294.3	c.4096+1G>A	5	YES	Use of a cryptic site 3309 nt upstream from 5' ss
158	BRCA1	NM_007294.3	c.4096+3A>G	4	YES	Use of a cryptic site 3309 nt upstream from 5' ss
159	BRCA1	NM_007294.3	c.4097G>A	1	NO	No change
160	BRCA1	NM_007294.3	c.4185_4185+3del	5	YES	exon 12 skipped
161	BRCA1	NM_007294.3	c.4185+2_4185+22delinsA	5	YES	exon 12 skipped
162	BRCA1	NM_007294.3	c.4185G>A	5	YES	exon 12 skipped
163	BRCA1	NM_007294.3	c.4357+1del	5	YES	exon 13 skipped
164	BRCA1	NM_007294.3	c.4357+1G>A	5	YES	exon 13 skipped
165	BRCA1	NM_007294.3	c.4357+1G>T	5	YES	exon 13 skipped
166	BRCA1	NM_007294.3	c.4357+6T>C	* 1	YES	exon 13 skipped
167	BRCA1	NM_007294.3	c.4358-2del	1	NO	No change
168	BRCA1	NM_007294.3	c.441+1G>A	5	YES	Use of a cryptic site 62 nt upstream from 5' ss and exon 7 skipped
169	BRCA1	NM_007294.3	c.441+2T>A	5	YES	Use of a cryptic site 62 nt upstream from 5' ss
170	BRCA1	NM_007294.3	c.441+2T>G	5	YES	Use of a cryptic site 62 nt upstream from 5' ss
171	BRCA1	NM_007294.3	c.4484+1G>T	5	YES	exon 14 skipped
172	BRCA1	NM_007294.3	c.4484G>C	5	YES	exon 14 skipped and exon 14, 15 skipped

173	BRCA1	NM_007294.3	c.4675+1G>A	5	YES	Use of a cryptic site 11 nt upstream from 5' ss and exon 15 skipped
174	BRCA1	NM_007294.3	c.4675G>A	5	YES	Use of a cryptic site 11 nt upstream from 5' ss
175	BRCA1	NM_007294.3	c.4985T>C	1	NO	No change
176	BRCA1	NM_007294.3	c.4986+1G>T	5	YES	Use of a cryptic site 65 nt downstream from 5' ss
177	BRCA1	NM_007294.3	c.4986+4A>G	5	YES	Use of a cryptic site 65 nt downstream from 5' ss
178	BRCA1	NM_007294.3	c.4986+5G>A	5	YES	Use of a cryptic site 65 nt downstream from 5' ss
179	BRCA1	NM_007294.3	c.4986+5G>T	5	YES	Use of a cryptic site 65 nt downstream from 5' ss
180	BRCA1	NM_007294.3	c.4986+6T>G	5	YES	Use of a cryptic site 65 nt downstream from 5' ss
181	BRCA1	NM_007294.3	c.4987-1G>A	5	YES	exon 17 skipped
182	BRCA1	NM_007294.3	c.4987-2A>G	5	YES	exon 17 skipped
183	BRCA1	NM_007294.3	c.4987-3C>G	5	YES	exon 17 skipped
184	BRCA1	NM_007294.3	c.4987A>T	5	YES	exon 17 skipped
185	BRCA1	NM_007294.3	c.4988T>A	5	YES	exon 17 skipping
186	BRCA1	NM_007294.3	c.5072C>A	1	NO	No change
187	BRCA1	NM_007294.3	c.5072C>T	5	YES	exon 17 skipping
188	BRCA1	NM_007294.3	c.5074+1G>T	5	YES	Use of a cryptic site 153 nt downstream from 5' ss
189	BRCA1	NM_007294.3	c.5074+3A>G	5	YES	Use of a cryptic site 153 nt downstream from 5' ss and exon 17 skipped
190	BRCA1	NM_007294.3	c.5074G>A	5	YES	Use of a cryptic site 153 nt downstream from 5' ss and exon 17 skipped
191	BRCA1	NM_007294.3	c.5074G>T	5	YES	Use of a cryptic site 153 nt downstream from 5' ss and exon 17 skipped
192	BRCA1	NM_007294.3	c.5152+1G>C	5	YES	exon 18 skipped
193	BRCA1	NM_007294.3	c.5152+5G>A	5	YES	exon 18 skipped
194	BRCA1	NM_007294.3	c.5153-1G>A	5	YES	Use of a cryptic site 1 nt downstream from 3' ss
195	BRCA1	NM_007294.3	c.5153-2del	5	YES	exon 19 skipped
196	BRCA1	NM_007294.3	c.5153G>C	1	NO	No change
197	BRCA1	NM_007294.3	c.5154G>T	1	NO	No change
198	BRCA1	NM_007294.3	c.5193+1del	5	YES	Use of a cryptic site 1 nt upstream from 5' ss
199	BRCA1	NM_007294.3	c.5193+2T>G	5	YES	exon 19 skipped
200	BRCA1	NM_007294.3	c.5194-12G>A	5	YES	Use of a cryptic site 10 nt upstream from 3' ss
201	BRCA1	NM_007294.3	c.5194-1G>T	5	YES	Use of a cryptic site 13 nt downstream from 3' ss and exon 20 skipped
202	BRCA1	NM_007294.3	c.5277+1G>A	5	YES	Use of a cryptic site 87 nt upstream from 3' ss and exon 20 skipped
203	BRCA1	NM_007294.3	c.5277+6G>C	2	NO	No change
204	BRCA1	NM_007294.3	c.5278-1G>A	5	YES	Use of a cryptic site 8 nt downstream from 3' ss and exon 21 skipped
205	BRCA1	NM_007294.3	c.5278-2del	5	YES	Use of a cryptic site 8 nt downstream from 3' ss and exon 21 skipped
206	BRCA1	NM_007294.3	c.5278-6_5278-4del	1	NO	No change
207	BRCA1	NM_007294.3	c.5332+1G>A	5	YES	exon 21 skipped
208	BRCA1	NM_007294.3	c.5332G>T	5	YES	exon 21 skipped
209	BRCA1	NM_007294.3	c.5333-1G>T	5	YES	exon 22 skipped
210	BRCA1	NM_007294.3	c.5333-6T>G	5	YES	exon 22 skipped
211	BRCA1	NM_007294.3	c.5333A>G	1	NO	No change
212	BRCA1	NM_007294.3	c.5406+2del	5	YES	exon 22 skipped
213	BRCA1	NM_007294.3	c.5406+3A>T	5	YES	exon 22 skipped
214	BRCA1	NM_007294.3	c.5406+4A>G	5	YES	exon 22 skipped
215	BRCA1	NM_007294.3	c.5406+5G>A	5	YES	exon 22 skipped
216	BRCA1	NM_007294.3	c.5407-2A>G	5	YES	exon 23 skipped
217	BRCA1	NM_007294.3	c.5408G>C	4	YES	exon 23 skipped
218	BRCA1	NM_007294.3	c.5467+5G>C	5	YES	exon 23 skipped
219	BRCA1	NM_007294.3	c.5467G>A	5	YES	exon 23 skipped
220	BRCA1	NM_007294.3	c.547+2T>C	5	YES	exon 8 skipped
221	BRCA1	NM_007294.3	c.547+6G>A	1	NO	No change
222	BRCA1	NM_007294.3	c.548-3del	2	NO	No change
223	BRCA1	NM_007294.3	c.593+4A>G	4	YES	exon 9 skipped
224	BRCA1	NM_007294.3	c.593+8A>G	1	NO	No change
225	BRCA1	NM_007294.3	c.594-2A>C	5	YES	exon 10 skipped
226	BRCA1	NM_007294.3	c.594-4A>G	1	NO	No change
227	BRCA1	NM_007294.3	c.668A>G	4	YES	exon 10 skipped
228	BRCA1	NM_007294.3	c.670+1delG	5	YES	Use of a cryptic site 1 nt upstream from 5' ss
229	BRCA1	NM_007294.3	c.671-2A>C	5	YES	exons 10, 11 skipped and exons 9_11 skipped
230	BRCA1	NM_007294.3	c.80+1G>A	5	YES	exon 2 skipped

231	BRCA1	NM_007294.3	c.80+3_80+6del	5	YES	exon 2 skipped
232	BRCA1	NM_007294.3	c.81-11del	1	NO	No change
233	BRCA1	NM_007294.3	c.81-6T>A	5	YES	Use of a cryptic site 2 nt upstream from 3' ss
234	BRCA1	NM_007294.3	c.81-9C>G	5	YES	Use of a cryptic site 8 nt upstream from 3' ss
235	BRCA2	NM_000059.3	c.316+5G>A	4	YES	exon 3 skipped
236	BRCA2	NM_000059.3	c.-39-9_-39-7del	1	NO	No change
237	BRCA2	NM_000059.3	c.-40+1G>A	5	YES	Use of a cryptic site 99 nt upstream from 5' ss
238	BRCA2	NM_000059.3	c.425G>T	5	YES	exon 4 skipped
239	BRCA2	NM_000059.3	c.426-12_426-8del	5	YES	exon 5 skipped
240	BRCA2	NM_000059.3	c.470_474del	4	YES	exon 5 skipped
241	BRCA2	NM_000059.3	c.473C>T	4	YES	exon 5 skipped
242	BRCA2	NM_000059.3	c.475+1G>A	5	YES	exon 5 skipped
243	BRCA2	NM_000059.3	c.475G>A	5	YES	exon 5 skipped
244	BRCA2	NM_000059.3	c.476-2A>G	5	YES	exon 5 skipped and exons 5, 6 skipped
245	BRCA2	NM_000059.3	c.516+1G>A	5	YES	exon 6 skipped and exons 5, 6 skipped
246	BRCA2	NM_000059.3	c.516+1G>T	5	YES	Use of a cryptic site 39 nt upstream from 5' ss and exon 6 skipped and exons ..
247	BRCA2	NM_000059.3	c.516G>A	5	YES	exon 6 skipped and exons 5, 6 skipped
248	BRCA2	NM_000059.3	c.517-1G>A	5	YES	exon 7 skipped
249	BRCA2	NM_000059.3	c.517G>T	* 1	YES	exon 7 skipped
250	BRCA2	NM_000059.3	c.518del	1	NO	No change
251	BRCA2	NM_000059.3	c.518G>T	2	NO	No change
252	BRCA2	NM_000059.3	c.631+2T>G	5	YES	exon 7 skipped
253	BRCA2	NM_000059.3	c.631+4A>G	4	YES	exon 7 skipped
254	BRCA2	NM_000059.3	c.632-1G>A	5	YES	exon 8 skipped
255	BRCA2	NM_000059.3	c.67+1G>A	5	YES	exon 2 skipped
256	BRCA2	NM_000059.3	c.680C>T	1	NO	No change
257	BRCA2	NM_000059.3	c.682-12_682-11del	1	NO	No change
258	BRCA2	NM_000059.3	c.682-2A>C	5	YES	Use of a cryptic site 73 nt downstream of 3' ss and exon 9 skipped
259	BRCA2	NM_000059.3	c.682-2A>G	5	YES	Use of a cryptic site 73 nt downstream of 3' ss and exon 9 skipped
260	BRCA2	NM_000059.3	c.6938-3T>C	2	NO	No change
261	BRCA2	NM_000059.3	c.6938-9_6938-6del	* 4	NO	No change
262	BRCA2	NM_000059.3	c.7008-2A>G	5	YES	exon 14 skipped
263	BRCA2	NM_000059.3	c.7435+8A>G	1	NO	No change
264	BRCA2	NM_000059.3	c.7617+1G>A	5	YES	exon 15 skipped
265	BRCA2	NM_000059.3	c.7617+2T>G	5	YES	exon 15 skipped
266	BRCA2	NM_000059.3	c.7618-1G>A	5	YES	Use of a cryptic site 45 nt downstream from 3' ss and exon 16 skipped
267	BRCA2	NM_000059.3	c.7806-9T>G	5	YES	Use of a cryptic site 69 nt downstream from 3' ss and exon 17 skipped
268	BRCA2	NM_000059.3	c.794-5_794-4insT	1	NO	No change
269	BRCA2	NM_000059.3	c.7976+3_7976+4del	- 3	YES	exon 17 skipped
270	BRCA2	NM_000059.3	c.7976+5G>T	- 3	NO	No change
271	BRCA2	NM_000059.3	c.7976G>A	- 3	YES	exon 17 skipped
272	BRCA2	NM_000059.3	c.7977-1G>C	5	YES	exon 18 skipped and exons 17, 18 skipped
273	BRCA2	NM_000059.3	c.7977-4_7977-3del	* 4	NO	No change
274	BRCA2	NM_000059.3	c.8331G>A	5	YES	exon 18 skipped
275	BRCA2	NM_000059.3	c.8332-1G>A	5	YES	Use of a cryptic site 14 nt downstream from 3' ss
276	BRCA2	NM_000059.3	c.8486A>T	4	YES	exon 19 skipped
277	BRCA2	NM_000059.3	c.8487+1G>A	5	YES	exon 19 skipped
278	BRCA2	NM_000059.3	c.8487+8G>A	1	NO	No change
279	BRCA2	NM_000059.3	c.8487G>A	5	YES	exon 19 skipped
280	BRCA2	NM_000059.3	c.8488-1G>A	5	YES	Use of a cryptic site 12 nt downstream from 3' ss and exon 20 skipped
281	BRCA2	NM_000059.3	c.8488-2A>G	5	YES	Use of a cryptic site 12 nt downstream from 3' ss and a cryptic site 1053 up
282	BRCA2	NM_000059.3	c.8632+1G>A	5	YES	exon 20 skipped
283	BRCA2	NM_000059.3	c.8633-24_8634del	5	YES	exon 21 skipped
284	BRCA2	NM_000059.3	c.8633-2A>G	5	YES	Use of a cryptic site 43 nt downstream from 3' ss and a cryptic site 93 nt ..
285	BRCA2	NM_000059.3	c.8754+1G>A	5	YES	Use of a cryptic site 46 nt downstream from 5' ss
286	BRCA2	NM_000059.3	c.8754+1G>C	5	YES	Use of a cryptic site 46 nt downstream from 5' ss
287	BRCA2	NM_000059.3	c.8754+3G>C	4	YES	Use of a cryptic site 46 nt downstream from 5' ss
288	BRCA2	NM_000059.3	c.8754+5G>A	5	YES	Use of a cryptic site 46 nt downstream from 5' ss

289	BRCA2	NM_000059.3	c.8754+5G>T	5	YES	Use of a cryptic site 46 nt downstream from 5' ss
290	BRCA2	NM_000059.3	c.8754G>A	5	YES	Use of a cryptic site 46 nt downstream from 5' ss
291	BRCA2	NM_000059.3	c.8755-1G>A	5	YES	exon 22 skipped
292	BRCA2	NM_000059.3	c.8948_8953+5del	5	YES	exon 22 skipped
293	BRCA2	NM_000059.3	c.8953+1G>T	5	YES	Use of a cryptic site 31 nt upstream from 5' ss and exon 22 skipped
294	BRCA2	NM_000059.3	c.8954-1_8955delinsAA	5	YES	Use of a cryptic site 51 nt downstream from 3' ss and exon 23 skipped
295	BRCA2	NM_000059.3	c.8954-3C>G	5	YES	Use of a cryptic site 2 nt upstream from 3' ss
296	BRCA2	NM_000059.3	c.9117+1G>A	5	YES	exon 23 skipped
297	BRCA2	NM_000059.3	c.9117+1G>T	5	YES	exon 23 skipped
298	BRCA2	NM_000059.3	c.9117+2T>A	5	YES	exon 23 skipped
299	BRCA2	NM_000059.3	c.9118-2A>G	5	YES	Use of a cryptic site 7 nt downstream from 3' ss
300	BRCA2	NM_000059.3	c.9249_9256+8del	5	YES	Use of a cryptic site 43 nt upstream from 5' ss and exon 24 skipped
301	BRCA2	NM_000059.3	c.9256+1G>A	5	YES	exon 24 skipped
302	BRCA2	NM_000059.3	c.9256G>T	5	YES	Use of a cryptic site 43 nt upstream from 5' ss and exon 24 skipped
303	BRCA2	NM_000059.3	c.9257-1G>C	5	YES	Use of a cryptic site 27 nt downstream from 3' ss and exon 25 skipped
304	BRCA2	NM_000059.3	c.9502-2A>C	5	YES	Use of a cryptic site 70 nt upstream from 3' ss and exon 26 skipped
305	BRCA2	NM_000059.3	c.9649-2A>G	5	YES	Use of a cryptic site 62 nt downstream from 3' ss
306	CFTR	NM_000492.3	c.1116+1G>A	5	YES	use of a cryptic site 34 nt upstream from 5' ss frameshift
307	CFTR	NM_000492.3	c.1209+2T>G	5	YES	exon 9 skipped
308	CFTR	NM_000492.3	c.1209G>A	5	YES	exon 9 skipped
309	CFTR	NM_000492.3	c.1393-2A>G	5	YES	use of a cryptic site 87 nt downstream from 3' ss frameshift
310	CFTR	NM_000492.3	c.1584G>A	4	YES	exon 10 skipping of low intensity
311	CFTR	NM_000492.3	c.1585-1G>A	5	YES	exon 12 skipped and use of a de novo 1 nt downstream from 3' ss
312	CFTR	NM_000492.3	c.1585-3T>G	5	YES	exon 12 skipping and use of a de novo site 2 nt upstream from 3' ss
313	CFTR	NM_000492.3	c.1585-8G>A	5	YES	use of a de novo site 7 nt upstream from 3' ss
314	CFTR	NM_000492.3	c.165-3C>T	* 1	YES	exon 3 skipping of low intensity
315	CFTR	NM_000492.3	c.1679+1G>C	5	YES	exon 12 skipped
316	CFTR	NM_000492.3	c.1766+1G>A	5	YES	exon 13 skipped
317	CFTR	NM_000492.3	c.1766+3A>G	4	YES	exon 13 skipped
318	CFTR	NM_000492.3	c.2490+1G>A	5	YES	exon 14 skipped
319	CFTR	NM_000492.3	c.2491G>T	5	YES	exon 15 skipping of strong intensity
320	CFTR	NM_000492.3	c.2620-6T>C	1	NO	no change
321	CFTR	NM_000492.3	c.2657+5G>A	5	YES	exon 16 skipped
322	CFTR	NM_000492.3	c.2658-1G>A	5	YES	exon 17 skipped
323	CFTR	NM_000492.3	c.274-6T>C	1	NO	no change
324	CFTR	NM_000492.3	c.2909-4A>G	1	NO	no change
325	CFTR	NM_000492.3	c.2988+1G>A	5	YES	exon 18 skipped
326	CFTR	NM_000492.3	c.2988G>A	4	YES	exon 18 skipped
327	CFTR	NM_000492.3	c.2989-1G>A	5	YES	exon 19 skipped
328	CFTR	NM_000492.3	c.2989-1G>T	5	YES	exon 19 skipped
329	CFTR	NM_000492.3	c.3139+3A>G	4	YES	exon 19 skipping of strong intensity
330	CFTR	NM_000492.3	c.3468+1G>A	5	YES	exon 21 skipped
331	CFTR	NM_000492.3	c.3468+2dup	5	YES	exon 21 skipped
332	CFTR	NM_000492.3	c.3718-1G>A	5	YES	exon 23 skipping and use of a de novo site 1 nt downstream from 3' ss
333	CFTR	NM_000492.3	c.3873+1G>A	5	YES	use of a cryptic site 29 nt downstream from 5' ss
334	CFTR	NM_000492.3	c.4242_4242+1delinsTT	5	YES	exon 26 skipped
335	CFTR	NM_000492.3	c.4242+1G>T	5	YES	exon 26 skipped
336	CFTR	NM_000492.3	c.489+1G>T	5	YES	exon 4 skipped and use of a cryptic site 93 nt upstream from 5' ss
337	CFTR	NM_000492.3	c.489+3A>G	4	YES	exon 4 skipped and use of a cryptic site 93 nt upstream from 5' ss
338	CFTR	NM_000492.3	c.489+4A>G	4	YES	exon 4 skipped and use of a cryptic site 93 nt upstream from 5' ss
339	CFTR	NM_000492.3	c.489G>A	5	YES	exon 4 skipped and use of a cryptic site 93 nt upstream from 5' ss
340	CFTR	NM_000492.3	c.490-1G>C	5	YES	exon 5 skipped and use of a cryptic site 9 nt downstream from 3' ss
341	CFTR	NM_000492.3	c.579+1G>T	5	YES	exon 5 skipped
342	CFTR	NM_000492.3	c.580-1G>T	5	YES	exon 6 skipped
343	CFTR	NM_000492.3	c.743+1G>A	5	YES	exon 6 skipped
344	CFTR	NM_000492.3	c.744-10_744-3del	4	YES	use of a cryptic site 32 nt upstream from 3' ss
345	CFTR	NM_000492.3	c.744-14_744-3del	4	YES	use of a cryptic site 32 nt upstream from 3' ss
346	CFTR	NM_000492.3	c.744-9_744-6del	2	NO	no change

347	CFTR	NM_000492.3	c.744-9_744-6dup	1	NO	no change
348	CFTR	NM_000492.3	c.870-12T>A	5	YES	use of a cryptic site 71 nt downstream from 3' ss frameshift
349	CFTR	NM_000492.3	c.870-4A>G	1	NO	no change
350	CTRC	NM_007272.2	c.133-3C>A	5	YES	exon 3 skipping of strong intensity
351	CTRC	NM_007272.2	c.640-12G>A	5	YES	use of a de novo splice site 11 nt upstream from 3' ss
352	HFE	NM_000410.3	c.76+2T>C	5	YES	Intron 1 retention
353	HJV	NM_213653.3	c.98-6C>G	1	NO	no change
354	LRP5	NM_002335.3	c.1585G>T	2	NO	no change
355	PKD1	NM_001009944.2	c.2853+2_2853+3del	5	YES	exon 11 skipping of strong intensity
356	RHD	NM_016124.4	c.1073+2T>C	5	YES	exon 7 skipping of strong intensity
357	RHD	NM_016124.4	c.1073T>C	1	NO	no change
358	RHD	NM_016124.4	c.1074-2A>C	5	YES	exon 8 skipping of strong intensity and use of cryptic splice site at 13 ..
359	RHD	NM_016124.4	c.1152A>C	4	YES	exon 8 skipping of strong intensity
360	RHD	NM_016124.4	c.1153+1G>A	5	YES	exon 8 skipped
361	RHD	NM_016124.4	c.1154-8T>A	4	YES	exon 9 skipping of low intensity
362	RHD	NM_016124.4	c.1154G>C	4	YES	exon 9 skipping of low intensity
363	RHD	NM_016124.4	c.1226A>T	4	YES	exon 9 skipping of low intensity
364	RHD	NM_016124.4	c.1227G>A	4	YES	exon 8 skipping of strong intensity
365	RHD	NM_016124.4	c.150T>C	2	NO	no change
366	RHD	NM_016124.4	c.334A>G	5	YES	exon 2 skipping of low intensity and use of cryptic splice site at 20 nt ups..
367	RHD	NM_016124.4	c.335+1G>A	5	YES	exon 2 skipping of strong intensity and use of cryptic splice site at 20 nt ..
368	RHD	NM_016124.4	c.335G>C	5	YES	exon 2 skipping and use of cryptic splice site at 20 nt upstream from 5' ss
369	RHD	NM_016124.4	c.335G>T	5	YES	exon 2 skipping of strong intensity and use of cryptic splice site at 20 nt u
370	RHD	NM_016124.4	c.336-1G>A	5	YES	exon 2 skipping of low intensity and use of two cryptic splice site at 28 nt..
371	RHD	NM_016124.4	c.336-2A>G	5	YES	exon 2 skipping of low intensity and use of two cryptic splice site at 28 nt..
372	RHD	NM_016124.4	c.336-2delA	5	YES	exon 2 skipping of low intensity and use of cryptic splice site at 28 nt up..
373	RHD	NM_016124.4	c.485A>G	1	NO	no change
374	RHD	NM_016124.4	c.486+1G>A	5	YES	exon 3 skipping of strong intensity and use of cryptic splice site 48 nt up..
375	RHD	NM_016124.4	c.486+2T>A	5	YES	exon 3 skipping of strong intensity and use of cryptic splice site 48 nt up..
376	RHD	NM_016124.4	c.486+3G>C	5	YES	exon 3 skipping of low intensity
377	RHD	NM_016124.4	c.486+5G>A	5	YES	exon 3 skipping of strong intensity
378	RHD	NM_016124.4	c.634+1G>A	5	YES	exon 4 skipping of low intensity and use of two cryptic splice site at 11- ..
379	RHD	NM_016124.4	c.634+2T>A	5	YES	exon 4 skipping of low intensity and use of two cryptic splice site at 11- ..
380	RHD	NM_016124.4	c.634+5G>A	5	YES	use of two cryptic splice site at 11- and 16 nt downstream from 5' ss
381	RHD	NM_016124.4	c.634+5G>T	5	YES	use of two cryptic splice site at 11- and 16 nt downstream from 5' ss
382	RHD	NM_016124.4	c.634G>C	5	YES	use of two cryptic splice site at 11- and 16 nt downstream from 5' ss
383	RHD	NM_016124.4	c.634G>T	5	YES	use of two cryptic splice site at 11- and 16 nt downstream from 5' ss
384	RHD	NM_016124.4	c.635-2A>G	5	YES	exon 5 skipped
385	RHD	NM_016124.4	c.635G>T	* 1	YES	exon 5 skipping of low intensity
386	RHD	NM_016124.4	c.636C>T	1	NO	no change
387	RHD	NM_016124.4	c.800A>T	* 4	NO	no change
388	RHD	NM_016124.4	c.801+1G>A	5	YES	exon 5 skipped
389	RHD	NM_016124.4	c.938C>T	2	NO	no change
390	RHD	NM_016124.4	c.939+2T>A	5	YES	exon 6 skipping of strong intensity and use of two ..
391	RHD	NM_016124.4	c.939G>A	5	YES	exon 6 skipping of strong intensity
392	RHD	NM_016124.4	c.940-4A>C	1	NO	no change
393	RHD	NM_016124.4	c.941G>T	2	NO	no change
394	SLC40A1	NM_014585.5	c.387C>T	2	NO	no change
395	TFR2	NM_003227.3	c.1473G>A	5	YES	use of cryptic site 43 nt downstream from 5' ss

3 References

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