Gene	Exon	Phenotype	Mutation	Reference	HOT-SKIP rank	HOT-SKIP percentile	ID[1]	15-mer included	15-mer excluded	Exon length
RET	11	Hirschsprung disease	E11+62C>T	Auricchio, A. et al.(1999) Am. J. Hum. Genet. 64, 1216-1221.	9	0.89	S1	CCTTCATCGTCTCGG	CCTTCATTGTCTCGG	257
TNFRSF5	5	Hyper IgM syndrome	E5+5A>T	Ferrari, S. et al. (2001) Proc. Natl. Acad. Sci. USA 98, 12614-12619.	80	22.22	S2	cagCTACAGGGGTTT	cagCTACTGGGGTTT	94
PTPRC	4	T cell activation	E4+76C>G	Thude, H. et al. (1995) Eur. J. Immunol. 25, 2101-2106.	560	72.16	S3	TCTCACCCGCAAGCA	TCTCACCGGCAAGCA	198
HPRT1	3	HPRT deficiency	E3+29A>T	Steingrimsdottir, H. et al. (1992) Nucleic Acids Res. 20, 1201-1208.	129	17.92	S4	TGTGATGAAGGAGAT	TGTGATGTAGGAGAT	184
FBN1	52	Marfan syndrome	E52+41C>T	Liu, W.G.et al. (1997) Nat. Genet. 16, 328-329.	33	13.31	S5	GGATCATCGTGGGAC	GGATCATTGTGGGAC	66
HMBS	3	Acute intermittend porphyria	E3+33C>G	Llewellyn, D.H.et al (1996) J. Med. Genet. 33, 437-438.	3	1.50	S6	TGATTCGCGTGGGTA	TGATTCGGGTGGGTA	54
GLA	Cryptic	Fabry disease	Ecr+54A>G	Ishii, S. et al. Am J Hum Genet, 70, 994-1002.	111	52.36	S7	CCCACTAAAGTgtaa	CCCACTAGAGTgtaa	57
FGB	Cryptic	Afibrinogenemia	Ecr+23G>A	Dear, A. et al. J. Thromb. Haemost, 4, 471-472.	109	59.24	S8	CACAGATGAAGCAAC	CACAGATAAAGCAAC	50
MLH1	3	Hereditary non-polyposis colorectal cancer	E3+7G>T	McVety, S. et al. (2006) J. Med. Genet. 43, 153-156.	233	61.32	S9	gAAAGAAGATCTGGA	gAAAGAATATCTGGA	99

Gene	Exon	Phenotype	Mutation	Reference	HOT-SKIP rank	HOT-SKIP percentile	ID[1]	15-mer included	15-mer excluded	Exon length
DMD	27	Muscular dystrophy	E27+27G>T	Shiga, N. et al. (1997) J. Clin. Invest. 100, 2204-2210.	89	12.43	S11	ACAAAAAGAAGCGAA	ACAAAAATAAGCGAA	183
COL4A5	Cryp-tic	Alport syndrome	Ecr+45A>G	King, K. et al. (2002) Hum. Genet, 111, 548-554.	277	48.43	S12	TAACAAGATGGAGAG	TAACAAGGTGGAGAG	147
GH1	3	Isolated growth hormone Deficiency, type II	E3+5A>G	Ryther, R.C. et al. (2003) Hum. Genet. 113, 140-148.	413	89.01	S13	tagGAAGAAGCCTAT	tagGAAGGAGCCTAT	120
NF1	30	Neurofibro-matosis, type I	E30+174G>T	Zatkova, A. et al. (2004) Hum. Mutat. 24, 491-501.	104	13.07	S14	ATTTTTGGAAGAGTG	ATTTTTGTAAGAGTG	203
ADA	7	Adenosine deminase deficiency	E7+37G>A	Ozsahin, H. et al. (1997) Blood 89, 2849-2855.	190	69.85	S15	TGTCCACGCCGGGGA	TGTCCACACCGGGGA	72
ATM	44	Ataxia telangiectasia	E44+58G>A	Teraoka, S.N. et al.(1999) Am. J. Hum. Genet. 64, 1617-1631.	293	73.99	S16	TGACCTCGAAACAGC	TGACCTCAAAACAGC	103
ATP7A	4	Menkes disease	E4+103G>A	Vulpe, C. et al.(1993) Nat. Genet. 3, 7-13.	288	36.00	S17	GGGAGATGGAATCAA	GGGAGATAGAATCAA	204
BRCA1	18	Breast and ovarian cancer	E18+18G>T	Mazoyer, S. et al.(1998) Am. J. Hum. Genet. 62, 713-715.	50	16.89	S18	TGTGTGTGAACGGAC	TGTGTGTTAACGGAC	78
F8	19	Haemophilia	E19+47C>T	Theophilus, B.D. et al. (2001) Haemophilia 7, 381-391.	3	0.66	S19	AATTTGGCGGGTGGA	AATTTGGTGGGTGGA	117
HEXB	11	Sandhoff disease	E11+8C>T	Wakamatsu, N. et al.(1992) J. Biol. Chem. 267, 2406-2413.	164	23.98	S20	CTTGCGCCGGGCACA	CTTGCGCTGGGCACA	175
IVD	2	Isovaleri-acidemia	E2+4C>T	Vockley, J., et al.	9	2.62	S21	ttagCTTCGTCAG	ttagCTTTGTCAGAC	90

Gene	Exon	Phenotype	Mutation	Reference	HOT-SKIP rank	HOT-SKIP percentile	ID[1]	15-mer included	15-mer excluded	Exon length
				(2000) Am. J. Hum. Genet. 66, 356-367.				AC		
MAPT	10	Frontotemporal dementia	E10+15G>T	Jiang, Z et al. (2000) Mol. Cell. Biol. 20, 4036-4048.	87	24.44	S23	TAATTAAGAAGAAGC	TAATTAATAAGAAGC	93
MAPT	10	Frontotemporal dementia	E10+29C>T	Jiang, Z et al. (2000) Mol. Cell. Biol. 20, 4036-4048.	91	25.56	S24	CTGGATCCTAGCAAC	CTGGATCTTAGCAAC	93
MAPT	10	Frontotemporal dementia	E10+66C>T	Jiang, Z et al. (2000) Mol. Cell. Biol. 20, 4036-4048.	65	18.26	S25	AGGATAACATCAAAC	AGGATAATATCAAAC	93
MLH1	17	Hereditary non-polyposis colorectal cancer	E17+79C>T	Nystrom-Lahti, M., et al. (1999) Genes Chromos. Cancer 26, 372-375.	68	19.10	S26	CATTCTTCGACTAGC	CATTCTTCACTAGCC	93
MLH1	17	Hereditary non-polyposis colorectal cancer	E17+90G>C	Nystrom-Lahti, M., et al. (1999) Genes Chromos. Cancer 26, 372-375.	69	19.38	S27	ATTCTTCGACTAGCC	ATTCTTCCACTAGCC	93
PMM2	5	Carbohydrate-deficient glycoprotein syndrome	E5+68G>A	Vuillaumier-Barrot, S. et al.(1999) Hum. Mutat. 14, 543-544.	332	86.46	S28	CAGCCAAGAAGAACG	CAGCCAAAAAGAACG	100
SMN1	7	Spinal muscular atrophy	E7+25G>T	Lorson, C.L. & Androphy, E.J. (2000) Hum. Mol. Genet. 9, 259-265.	184	92.00	S29	AAAAGAAGGAAGGTG	AAAAGAATGAAGGTG	54
APC	14	Familial adenomatous polyposis	E14+175C>G	Goncalves, V., et al. (2008) Mutat. Res. 662, 33-36.	63	7.46	S30	GATATTACGGAATGT	GATATTAGGGAATGT	215
MCAD	5	Medium-chain acyl-CoA dehydrogenas	E5+76C>T	Nielsen, K.B. et al.(2007) Am. J.	167	43.04	S31	GTTCAGACTGCTATT	GTTCAGATTGCTATT	101

Gene	Exon	Phenotype	Mutation	Reference	HOT-SKIP rank	HOT-SKIP percentile	ID[1]	15-mer included	15-mer excluded	Exon length
		e deficiency		Hum. Genet. 80, 416-432. Suphapeeti						
PTEN	6	Bannayan-Riley Ruvalcaba syndrome	E6+19C>T	porn, K. et al. (2006) Jpn. J. Clin. Oncol. 36, 814-821.	90	16.30	S32	TCCCAG TCAGA GGCG	TCCCAGT TAGAGG CG	142
PDHA 1	6	Pyruvate dehydrogenase complex deficiency	E6+82G>A	Okajima, K. et al. (2006) Mol. Genet. Metab. 87, 162-168.	159	44.66	S33	CGATG GTGCTG CTAA	CGATGGT ACTGCTA A	93
DMD	38	Muscular dystrophy	E38+108 del4	Tran, V.K.et al. (2006) J. Med. Genet. 43, 924-930.	120	25.21	S34	AAGAC TTCAAT AAAG	AAGACTT GAATAA AG	123
BRCA2	18	Breast cancer	E18+189 C>G	Fackenthal, J.D. et al.(2002) Am. J. Hum. Genet. 71, 625-631.	1194	85.04	S37	GAACTT ACAGA TGGG	GAACTTA GAGATG GG	355
DMD	29	Muscular dystrophy	E29+19C>T	Ginjaar, I.B. et al.(2000) Eur. J. Hum. Genet. 8, 793-796.	116	19.86	S38	TTTGAT GCGAC ATTC	TTTGATG TGACATT C	150
SMN1	7	Spinal muscular atrophy	E7+6C>T	Kashima, T. et al.(2007) Hum. Mol. Genet. 16, 3149-3159.	6	3.00	S39	agGGTT TCAGAC AAA	agGGTTT TAGACA AA	54
BEST1	6	Vitreoretino-choroidopathy	E6+68T>C	Burgess, R. et al.(2008) J. Med. Genet. 46, 620-625.	104	35.14	S40	CCACTG GTGTAT ACA	CCACTG GCGTATA CA	78
BEST1	6	Vitreoretino-choroidopathy	E6+71A>G	Burgess, R. et al.(2008) J. Med. Genet. 46, 620-625.	27	9.12	S41	CTGGTG TATACA CAG	CTGGTGT GTACAC AG	78
HRPT2	2	No exon skipping	E2+34C>G	Hahn, M. A. et al. (2009) J Endocrinol. 201(3):387-	227	55.64	N1	AGTACT ACACAT TGG	AGTACTA GACATTG G	106

Gene	Exon	Phenotype	Mutation	Reference	HOT-SKIP rank	HOT-SKIP percentile	ID[1]	15-mer included	15-mer excluded	Exon length
				96.						
HRPT2	5	No exon skipping	E5+36C>T	Hahn, M. A. et al. (2009) J Endocrinol. 201(3):387-96.	93	47.45	N2	AGAAG CACAG AAACC	AGAAGC ATAGAA ACC	53
HRPT2	7	No exon skipping	E7+152C>T	Hahn, M. A. et al. (2009) J Endocrinol. 201(3):387-96.	435	51.06	N3	TGTGAC CCGAG ATAT	TGTGACC TGAGAT AT	217
NF1	4a	No exon skipping	E4a+62T>G	Ars, E. et al. (2000)Hum Mol Genet. 22;9(2):237-47.	673	89.97	N4	CCAGA AATCTG CCAT	CCAGAA AGCTGC CAT	191
NF1	12a	No exon skipping	E12a+76 G>A	Ars, E. et al. (2000)Hum Mol Genet. 22;9(2):237-47.	189	39.38	N5	TCAAGT GGTTGC GGG	TCAAGTG ATTGCGG G	124
NF1	13	No exon skipping	E13+40C>T	Ars, E. et al. (2000)Hum Mol Genet. 22;9(2):237-47.	419	42.58	N6	GATTTG CCGAC AAGC	AATTTGC TGACAA GC	250
NF1	20	No exon skipping	E20+105 C>G	Ars, E. et al. (2000)Hum Mol Genet. 22;9(2):237-47.	285	40.03	N7	CTGGCA TCACTG AGG	CTGGCAT GACTGA GG	182
NF1	21	No exon skipping	E21+114 C>T	Ars, E. et al. (2000)Hum Mol Genet. 22;9(2):237-47.	154	18.51	N8	GGCTG ATCGGT TTGA	GGCTGAT TGGTTTG A	212
NF1	25	No exon skipping	E25+5T>C	Ars, E. et al. (2000)Hum Mol Genet. 22;9(2):237-47.	310	82.45	N9	tagATAC TTCAGA GT	tagATACC TCAGAGT	98
NF1	27	No exon skipping	E27a+23 C>T	Ars, E. et al. (2000)Hum Mol Genet. 22;9(2):237-47.	370	64.69	N10	TGGAA GACGA CCTTT	TGGAAG ATGACCT TT	147
NF1	29	No exon skipping	E29+183 T>A	Raponi, M. et al. (2009)	1297	96.22	N11	AGGAG TGTGAA GCCA	AGGAGT GAGAAG CCA	341

Gene	Exon	Phenotype	Mutation	Reference	HOT-SKIP rank	HOT-SKIP percentile	ID[1]	15-mer included	15-mer excluded	Exon length
				FEBS J. 276(7):2060-73. Baralle, M. et al.				TGACAC	TGACACT	
NF1	37	No exon skipping	E37+34T>A	(2006) FEBS Lett. 7;580(18):4449-56.	266	67.86	N12	TTACAACAG	AACAACAG	102
BRCA2	18	No exon skipping	E18+186T>G	Bonnet, C. et al. (2008) J Med Genet. 45(7):438-46.	1039	74.00	N13	ATTGAACTTACAGAT	ATTGAACGTACAGAT	355
BRCA2	23	No exon skipping	E23+19G>A	Whiley, P. J. et al. (2010) BMC Med Genet. 28;11:80.	341	53.28	N14	ATTTGGCGTCCATCA	ATTTGGCATCCATCA	164
BRCA2	24	No exon skipping	E24+55A>G	Whiley, P. J. et al. (2010) BMC Med Genet. 28;11:80.	418	77.41	N15	TCACTTCAGCAAATT	TCACTTCGGCAAATT	139
CFTR	9	No exon skipping	E9+118G>T	Pagani, F. et al. (2003) J Biol Chem. 18;278(29):26580-8.	136	18.99	N16	CCTGAAAGATATTAA	CCTGAAATATATTAA	183
F8	9	No exon skipping	E9+126G>A	Theophilus, B. D. et al. (2001) Haemophilia. 7(4):381-91.	663	98.66	N17	GAATCAGGAATCTTG	GAATCAGAAATCTTG	172
F8	12	No exon skipping	E12+52C>G	Theophilus, B. D. et al. (2001) Haemophilia. 7(4):381-91.	533	90.65	N18	TGAGAACCGAAGCTG	TGAGAACGGAAGCTG	151
F8	16	No exon skipping	E16+184G>A	Theophilus, B. D. et al. (2001) Haemophilia. 7(4):381-91.	803	96.05	N19	CTGCAAAGCCTGGGC	CTGCAAAACCTGGGC	213
F8	18	No exon skipping	E18+139G>A	Theophilus, B. D. et al. (2001) Haemophilia. 7(4):381-91.	255	35.61	N20	ACTGTACGAAAAAAA	ACTGTACAAAAAAAA	183
F8	19	No exon skipping	E19+13C>G	Theophilus, B. D. et al.	289	63.94	N21	TTTGAGACAGT	TTTGAGAGAGTGG	117

Gene	Exon	Phenotype	Mutation	Reference	HOT-SKIP rank	HOT-SKIP percentile	ID[1]	15-mer included	15-mer excluded	Exon length
				(2001) Haemophilia. 7(4):381-91.				GGAA	AA	
F8	22	No exon skipping	E22+4G>T	Theophilus, B. D. et al. (2001) Haemophilia. 7(4):381-91.	104	17.11	N22	gtagGTGGATCTGTT	gtagGTGTATCTGTT	156
F8	23	No exon skipping	E23+77G>A	Theophilus, B. D. et al. (2001) Haemophilia. 7(4):381-91.	183	32.45	N23	TACATCCGTTTGCAC	TACATCCATTTGCAC	145
F8	24	No exon skipping	E24+109G>T	Theophilus, B. D. et al. (2001) Haemophilia. 7(4):381-91.	444	76.55	N24	AAAGCTCGACTTCAC	AAAGCTCTACTTCAC	149
MSH2	2	No exon skipping	E2+128G>A	Tournier, I. et al. (2008) Hum. Mutat. 29(12):1412-24.	583	96.52	N25	CATCCAAGGAGAATG	CATCCAAAGAGAATG	155
MSH2	3	No exon skipping	E3+229T>C	Tournier, I. et al. (2008) Hum. Mutat. 29(12):1412-24.	727	66.09	N26	AAAGGAATGTGTTTT	AAAGGAACGTGTTTT	279
MSH2	6	No exon skipping	E6+49A>G	Tournier, I. et al. (2008) Hum. Mutat. 29(12):1412-24.	231	44.42	N27	CTTGCTGAATAAGTG	CTTGCTGGATAAGTG	134
MSH2	9	No exon skipping	E9+102A>G	Tournier, I. et al. (2008) Hum. Mutat. 29(12):1412-24.	145	30.21	N28	CAACATTAATAAGTG	CAACATTGATAAGTG	124
MSH2	10	No exon skipping	E10+92T>A	Tournier, I. et al. (2008) Hum. Mutat. 29(12):1412-24.	541	92.01	N29	TCCTTCGTAACAATA	TCCTTCGAAACAATA	151
MSH2	11	No exon skipping	E11+76A>G	Tournier, I. et al.	312	82.98	N30	TTGTTAAAGAA	TTGTTAAGGAAAT	98

Gene	Exon	Phenotype	Mutation	Reference	HOT-SKIP rank	HOT-SKIP percentile	ID[1]	15-mer included	15-mer excluded	Exon length
				(2008) Hum. Mutat. 29(12):1412-24.				ATTG	TG	
MSH2	12	No exon skipping	E12+69C>A	Tournier, I. et al. (2008) Hum. Mutat. 29(12):1412-24.	510	52.69	N31	CTTTGCTCACGTGTC	CTTTGCTAACGTGTC	246
MSH2	13	No exon skipping	E13+82C>T	Tournier, I. et al. (2008) Hum. Mutat. 29(12):1412-24.	359	44.65	N32	TTTGTGCCATGTGAG	TTTGTGCTATGTGAG	205
MSH2	14	No exon skipping	E14+32G>T	Tournier, I. et al. (2008) Hum. Mutat. 29(12):1412-24.	390	39.96	N33	AATCATAGATGAATT	AATCATATATGAATT	248
MSH2	15	No exon skipping	E15+59T>A	Tournier, I. et al. (2008) Hum. Mutat. 29(12):1412-24.	557	80.96	N34	CTAAGCATGTAATAG	CTAAGCAAGTAATAG	176
MLH1	2	No exon skipping	E2+83G>A	Tournier, I. et al. (2008) Hum. Mutat. 29(12):1412-24.	276	79.31	N35	TGGCACCGGGATCAG	TGGCACCAGGATCAG	91
MLH1	3	No exon skipping	E3+85G>A	Tournier, I. et al. (2008) Hum. Mutat. 29(12):1412-24.	146	38.42	N36	TACCTATGGCTTTCG	TACCTATAGCTTTCG	99
MLH1	4	No exon skipping	E4+44C>T	Tournier, I. et al. (2008) Hum. Mutat. 29(12):1412-24.	250	89.29	N37	ATTACAACGAAAACA	ATTACAATGAAAACA	74
MLH1	8	No exon skipping	E8+59T>G	Tournier, I. et al. (2008) Hum. Mutat.	180	52.94	N38	GACAATATTCGCTCC	GACAATAGTCGCTCC	89

Gene	Exon	Phenotype	Mutation	Reference	HOT-SKIP rank	HOT-SKIP percentile	ID[1]	15-mer included	15-mer excluded	Exon length
				29(12):1412-24. Tournier, I. et al.						
MLH1	9	No exon skipping	E9+101C>T	(2008) Hum. Mutat. 29(12):1412-24. Tournier, I. et al.	12	2.75	N39	CTTCTTACTCTTCAT	CTTCTTATTCTTCAT	113
MLH1	11	No exon skipping	E11+76G>C	(2008) Hum. Mutat. 29(12):1412-24. Lastella, P	529	88.17	N40	ACGAGGAGAGCATCC	ACGAGGACAGCATCC	154
MLH1	12	No exon skipping	E12+345G>T	et al. (2004)J Med Genet. 41(6):e72. Tournier, I. et al.	979	66.69	N41	CAGAGAAGAGAGGAC	CAGAGAATAGAGGAC	371
MLH1	13	No exon skipping	E13+12G>A	(2008) Hum. Mutat. 29(12):1412-24. Tournier, I. et al.	540	93.10	N42	AGACATCGGGAAGAT	AGACATCAGGAAGAT	149
MLH1	14	No exon skipping	E14+58C>A	(2008) Hum. Mutat. 29(12):1412-24. Tournier, I. et al.	112	26.67	N43	CAGTGGGCCTTGGCA	CAGTGGGACTTGGCA	109
MLH1	16	No exon skipping	E16+89T>A	(2008) Hum. Mutat. 29(12):1412-24. Tournier, I. et al.	553	85.87	N44	GAAGGACTTGCTGAA	GAAGGACATGCTGAA	165
MLH1	17	No exon skipping	E17+63G>T	(2008) Hum. Mutat. 29(12):1412-24. Tournier, I. et al.	121	33.99	N45	AGGGACTGCCTATCT	AGGGACTTCCTATCT	93
MLH1	18	No exon skipping	E18+52G>A	(2008) Hum. Mutat. 29(12):1412-24.	163	37.05	N46	AGAATGCGCTATGTT	AGAATGCACTATGTT	1