

Supplementary table 1

Index no	Systematic name	Mutation name	Position	Type	Match	Probe Length	Sequence	Calculated Tm	DTM	Calc DG	DDG	Stringency
1	c.47_48delCT	L15/S16fsdelC orp.S16>XfsX1	Central	Wt Mt	Mt-2	15 17	AACTCTCTGACTTTG GAAACTCTGACTTTGGA	32.4 35.2	-2.8	-12 -14	2.7	0.35 X SSC
2	c.60+5G>T	IVS1+5G>T	Proximal	Wt Mt	Mt+2	19 18	GACAGgtgagccacggcag GGACAGgtgatccacggc	55.1 53.3	1.8	-25 -22	-2.3	4 X SSC
3	c.115_117del TTC	deltaF39/ F39del	Central	Wt Mt	Mt+2	15 13	CTGATCTTCTCACTC ACTGATCTCACTC	39.0 31.9	7.1	-16 -14	-2.0	0.35 X SSC
4	c.117C>G	F39L	Proximal	Wt Mt	Tm	19 19	ACTGATCTTCTCACTCAAA ACTGATCTTGTCACTCAAA	39.6 39.3	0.3	-17 -16	-1.3	1 X SSC
5	c.136G>A	G46S	Distal	Wt Mt	Tm	19 19	GAAGAAGTTGGTGCATTGG AAGAAGTTAGTGCATTGGC	41.8 41.4	0.4	-17 -17	0.2	0.35 X SSC
6	c.140C>T	A47V	Proximal	Wt Mt	Tm	25 24	AAGAAGTTGGTGCATTGGCCAAAGT AGAAGTTGGTGTATTGGCCAAAGT	49.4 48.7	0.7	-24 -21	-2.6	0.35 X SSC
7	c.143T>C	L48S	Distal	Wt Mt	Mt-2	17 18	TGGTGCATTGGCCAAAG TTGGTGCATCGGCCAAAG	43.8 46.3	-2.5	-19 -21	1.8	0.35 X SSC
8	[c.187A>C;c.1 90C>A]	T63P/H64N	Central	Wt Mt	Mt+2	19 17	TAAACCTGACCACATTGA AAACCTGCCCAACATTG	41.3 39.9	1.4	-21 -19	-1.9	1 M Urea
9	c.194T>C	I65T	Proximal	Wt Mt	Mt+2	19 16	TGACCCACATTGAATCTAG ACCCACACTGAATCTA	40.6 39.1	1.5	-19 -18	-0.9	4 X SSC
10	c.204A>T	R68S	Proximal	Wt Mt	Tm	15 14	AATCTAGACCTTCTC AATCTAGTCTTCT	34.9 34.6	0.3	-16 -14	-1.9	1 X SSC
11	c.311C>A	A104D	Proximal	Wt Mt	Mt+2	15 15	ATTGGTGCCACTGTCTC TTGGTGACACTGTCC	46.1 43.3	2.8	-17 -16	-0.5	0.35 X SSC
12	c.329C>T	S110L	Proximal	Wt Mt	Tm	15 17	GAGCTTTCACGAGAT CAGAGCTTTTACGAGAT	37.1 37.3	-0.2	-17 -17	-0.2	4 X SSC
13	c.371C>T	T124I	Distal	Wt Mt	Tm	17 17	CCAAGAACCATTCAA CCAAGAATCATCAAGA	31.4 31.6	-0.2	-15 -14	-1.3	0.35 X SSC
14	c.473G>A	R158Q	Proximal	Wt Mt	Tm	19 21	GTGCAAGACGGAAGCAGTT CGTGCAAGACAGAAGCAGTTT	45.8 45.6	0.2	-19 -20	0.4	0.1 X SSC
15	c.529G>C	V177L	Proximal	Wt Mt	Mt-2	15 18	CCCTCGAGTGGAATA CATCCCTCGACTGGAATA	40.7 44.3	-3.6	-17 -21	3.8	3 M Urea
16	c.663_664del AG	221/222 del AG/ E221fs	Distal	Wt Mt	Tm	15 15	CCATGAAGATAACAT TCCATGAATAACATT	26.3 25.8	0.5	-13 -12	-0.7	4 X SSC
17	c.688G>A	V230I	Central	Wt Mt	Mt-2	17 20	TGGAAGACGTTTCTCAg CTGGAAGACATTTCTCAgTT	38.6 43.1	-4.5	-16 -19	3.1	0.01 X SSC
18	c.727C>T-GG	R243X	Proximal	Wt Mt	Tm	13 13	CGCCTCCGACCTG TCCGCCTCTGACC	48.6 49.4	-0.8	-21 -20	-0.4	0.35 X SSC
19	c.727C>T-AA	R243X	Proximal	Wt Mt	Tm	13 13	CGCCTCCGACCTG TCCGCCTCTGACC	48.6 49.4	-0.8	-21 -20	-0.4	0.01 X SSC
20	c.730C>T-AG	P244S	Distal	Wt Mt	Mt+2	15 13	CCTCCGACCTGTaGC CCTCCGATCTGTa	51.5 40.3	11.2	-21 -15	-6.0	0.35 X SSC
21	c.730C>T-GA	P244S	Proximal	Wt Mt	Tm	21 22	CCGCCTCCGACCTGTgGCTGG CCGCCTCCGATCTGTgGCTGGC	64.8 65.8	-1	-32 -32	0.1	3 M Urea
22	c.734T>A	V245E	Distal	Wt Mt	Tm	19 19	TCCGACCTGTgGCTGGCCT CCGACCTGcGCTGGCCTG	62.6 62.2	0.4	-27 -28	1.6	1 M Urea
23	c.781C>T	R261X	Central	Wt Mt	Mt+2	15 14	GGCCTTCCGAGTCTT GGCCTTCTGAGTCT	52.5 48.8	3.7	-21 -18	-2.6	0.01 X SSC
24	c.782G>A	R261Q	Distal	Wt Mt	Tm	15 16	GCCTTCCGAGTCTTC GGCCTTCCAAGTCTTC	51.3 51.2	0.1	-20 -19	-0.9	1 M Urea
25	c.814G>T	G272X	Central	Wt Mt	Mt-2	23 26	ACATCAGACATGGATCCAAGCCC GTACATCAGACATTGATCCAAGCCCA	53 53.9	-0.9	-28 -28	-0.6	3 M Urea
26	c.842C>T	P281L	Central	Wt Mt	Tm	13 15	CCGAACCGtgagt CCGAACCTgtgagtlac	38.3 36.5	1.8	-15 -15	-0.2	1 M Urea
27	c.842+4A>G	IVS7nt4a>g	Central	Wt Mt	Tm	15 14	AACCgtgagtactgt AACGtgggtactg	37.8 38.4	-0.6	-14 -15	0.6	1 M Urea
28	c.844G>A	D282N	Distal	Wt Mt	Mt+2	21 21	gtctttcagTGACATCTGCCA gtctttcagTAACATCTGCCA	54.5 50.4	4.1	-22 -21	-1.6	0.1 X SSC
29	c.898G>T	A300S	Central	Wt Mt	Tm	13 14	AGCTTTGCCCAGT CAGCTTTTCCAGT	46.5 45.4	1.1	-18 -18	0.0	0.35 X SSC
30	c.916A>G	I306V	Central	Wt Mt	Mt-2	17 17	tacagGAAATTGGCCTT tacagGAAGTTGGCCTT	38.4 44.1	-5.7	-17 -18	1.3	0.35 X SSC
31	c.997C>T	L333F	Proximal	Wt Mt	Tm	15 15	GTTTGGGCTCTGCAA AGTTTGGGTTCTGCA	45.5 44.6	0.9	-17 -16	-1.3	4 X SSC
32	c.1006C>T	Q336X	Distal	Wt Mt	Tm	17 18	TCTGCAAAACAGGAGAC TCTGCAAAATAAGGAGACT	35.0 35.0	0	-16 -16	-0.2	4 X SSC
33	c.1042C>G	L348V	Distal	Wt Mt	Tm	13 15	GGGCTCCTGTCTCAT TGGGCTCGTGTCTATC	48.5 48.3	0.2	-17 -17	-0.4	1 M Urea
34	c.1066- 11G>A	IVS10-11G>A	Central	Wt Mt	Mt+2	15 16	ttcacttggggccta tcacttagggcctaca	47.1 47.0	0.1	-20 -19	-0.6	0.35 X SSC
35	c.1068C>G	Y356X	Proximal	Wt Mt	Mt-2	23 25	ggcctacagTACTGCTTATCAGA ggcctacagTAGTGCTTATCAGAGA	54.4 56.5	-2.1	-26 -27	1.5	1 M Urea
36	c.1139C>T	T380M	Central	Wt Mt	Mt+2	17 17	CACTGTCTACGGAGTTTC CACTGTCTACGGAGTTCC	42.1 48.8	-6.7	-15 -19	4.1	0.1 X SSC
37	c.1157A>G	Y386C	Distal	Wt Mt	Tm	15 14	CCCTcTATTACGTG CCCTcTGTTACGTG	43.7 43	0.7	-18 -16	-2.3	0.1 X SSC
38	c.1160A>G	F390C	Central	Wt	Mt+2	21	TACGTGGCAGAGAGTTTAAAT	42.5	-1.3	-19	-2.2	0.35 X SSC

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38	c.1109A>G	L390G	Central	Mt		17	GTGGCAGGGAGTTTAA	43.8		-17	-2.2	0.35 X SSC
39	c.1208C>T	A403V	Central	Wt	Tm	17	GAACTTTGCTGCCACAA	41.7	-0.4	-18	-0.8	0.35 X SSC
				Mt		18	gGAACTTTGTGCCACAA	42.1		-17		
40	c.1222C>T	R408W	Central	Wt	Mt-2	13	ATACCTCGGCCCT	47.8	1.9	-20	-0.1	4 X SSC
				Mt		16	CAATACCTTGGCCCTT	45.9		-20		
41	c.1223G>A	R408Q	Distal	Wt	Tm	13	TACCTCGGCCCTT	48.3	-0.1	-20	0.3	0.1 X SSC
				Mt		14	TACCTCAGCCCTTC	48.4		-20		
42	c.1241A>G	Y414C	Distal	Wt	Tm	21	TCAGTTCGCTACGACCCATAC	50.9	-1.3	-23	0.3	1 M Urea
				Mt		19	AGTTCGCTGCGACCCATAC	52.2		-23		
43	c.1243G>A	D415N	Distal	Wt	Tm	15	TCGCTACGACCCATA	42.3	0	-20	-0.3	0.35 X SSC
				Mt		17	TCGCTACAACCCATACA	42.3		-19		
44	c.1315+1G>A	IVS12+1G>A	Central	Wt	Tm	19	CCATTAACAgtaagtaatt	31.9	0.4	-13	0.5	1 X SSC
				Mt		22	TCCATTAACAataagtaattta	31.5		-14		

References

1. Nowacki et al. Nucleic Acid Res. 1997, 25(1) 139-142
2. Hoang et al. Nucleic Acid Res. 1996, 24(1) 127-131
3. Guldberg et al. Am J Hum Genet. 1998, 63(1) 71-79