

Supplemental Table 1. Summary of Roche 454, Illumina GA, and ABI SOLiD sequence reads. Raw reads were generated, filtered for quality, and then aligned to the reference sequence (Supplemental Methods). All subsequent analyses in this study are based on using the Newbler algorithm to align Roche 454 reads and the MAQ algorithm to align Illumina GA reads.

		NA17156		NA17275		NA17460		NA17773	
Roche 454	No. raw sequence reads	47,968		31,669		71,580		46,244	
	Alignment tool	Newbler	BLAST	Newbler	BLAST	Newbler	BLAST	Newbler	BLAST
	Aligned reads (% of raw)	95.6	83.7	94.7	80.6	95.6	82.2	95.9	82.1

Illumina GA	No. raw sequence reads	6,682,085		5,979,236		5,426,181		5,315,215	
	Reads passing filters (% of raw)	52.26		53.16		57.18		58.36	
	Alignment tool	ELAND	MAQ	ELAND	MAQ	ELAND	MAQ	ELAND	MAQ
	Aligned reads (% of filtered)	84.61	78.72	88.42	82.36	80.95	77.89	71.17	70.16

ABI SOLiD	No. raw sequence reads	20,426,667		15,318,798		16,023,158		26,846,896	
	Reads passing filters (% of raw)	40.39		35.29		28.84		35.63	
	Aligned reads (% of filtered)	96.71		96.51		95.84		96.63	

Supplemental Table 2. Sequence coverage of targeted intervals in the four samples (Supplemental Methods).

	NA17156	NA17275	NA17460	NA17773
Targeted bp	266,201			
Sequenced bp	251,973	255,057	266,201	266,201
Analyzed bp	245,892	248,980	260,036	260,036
Roche 454				
≥10x	242,857 (98.77)	243,085 (97.63)	258,450 (99.39)	257,623 (99.07)
0x	0	0	0	61
Avg. coverage	43	28	61	39
Illumina GA				
≥10x	245,778 (99.95)	248,960 (99.99)	259,626 (99.84)	258,578 (99.82)
0x	0	0	0	5
Avg. coverage	203	263	157	130
ABI SOLiD				
≥10x	245,197 (97.2)	248,112 (99.7)	259,427 (99.8)	259,272 (99.7)
0x	88	153	138	85
Avg. coverage	1036	671	537	1121

Supplemental Table 3. Sequence coverage variability between amplicons. The fold difference and distribution in sequence coverage between amplicons for each NGS technology (Supplemental Methods).

		Max	95th percentile	5th percentile	Min	95th to 5th percentile ratio
Roche 454	NA17156	1.68	1.37	0.63	0.28	2.16
	NA17275	1.43	1.35	0.61	0.33	2.22
	NA17460	2.11	1.52	0.62	0.48	2.46
	NA17773	1.54	1.36	0.72	0.58	1.90
Illumina GA	NA17156	1.55	1.48	0.63	0.42	2.34
	NA17275	1.42	1.34	0.49	0.43	2.74
	NA17460	1.61	1.56	0.60	0.31	2.62
	NA17773	1.53	1.24	0.57	0.34	2.17
ABI SOLiD	NA17156	1.47	1.43	0.48	0.37	2.99
	NA17275	1.41	1.31	0.75	0.73	1.74
	NA17460	3.16	1.53	0.78	0.56	1.97
	NA17773	1.89	1.64	0.60	0.40	2.75

Supplemental Table 4. Sequence coverage of repetitive elements. Relative coverage of repetitive to unique sequences across the 260-kb analyzed interval. Standard deviation corresponds to the 4 samples variation.

	bp per sample	Roche 454	Illumina GA	ABI SOLiD
All repeats	101851	1.13+/-0.03	1.23+/-0.09	0.68+/-0.06
LINE	47030	1.25+/-0.06	0.78+/-0.03	0.56+/-0.03
SINE	33214	1.04+/-0.04	2.08+/-0.21	0.70+/-0.03
Alu	22263	1.02+/-0.06	2.63+/-0.3	0.56+/-0.03
Simple repeats	2462	0.99+/-0.12	2.19+/-0.18	0.65+/-0.08
Low Complexity repeats	1051	0.89+/-0.09	0.84+/-0.08	0.48+/-0.14

Supplemental Table 5A. No Coverage Regions. Regions are counted multiple times if they have 0 coverage in more than one individual.

	No. of Regions	Total Number of base pairs with no coverage	Fraction of Regions that are Repetitive	% AT, averaged across regions
Roche 454	1 ^a	59	0/1	29%
Illumina GA	2	5	2/2	58%
ABI SOLiD	40	464	30/40	90%

Supplemental Table 5B. Low Coverage Regions. For details see Supplemental Table 6.

	No. of Regions	Total Length of Low Coverage Regions (bp)	Fraction of Regions that are Repetitive	% AT, averaged across regions
Roche 454	1	4	0/1	25%
Illumina GA	13	272	11/13	82%
ABI SOLiD	168	3415	128/168	85%

^aRaw 454 reads with low-quality bases aligned to the 59 bp interval, but no high-quality reads aligned to the this interval (see Supplemental Methods).

Supplemental Table 6. Detailed analysis of low and no coverage regions

NGS	coverage type	NCBI36 coordinates	coverage type details	Sequence	repeats	%AT	length	
454	low coverage	chr7:150286181-150286184	≤2reads in ≥2 samples	GAGC		25	4	
	no coverage	chr7:150298287-150298345	no coverage in NA17773	CACACACCCCCAAGCTGGCCCTTCTGCTGCCCATGGCCCCCTCCCCAGAGCCCAAGG		29	59	
Illumina GA	low coverage	chr3:38588463-38588538	≤9 reads in ≥2 samples	TAATATATAAATATGCACT TATATAGGTGCATATTATA TATTATAATATATACTTAT ATTTATATATAGGTTTATA	LINE	87	76	
		chr21:34764577-34764636		GTAATAAATAAATAACTGAACTGATAAA CAATATTAATAATTTAATAGCCATTAAAT T		85	60	
		chr7:150286423-150286446		CGGGGCGCCCCGCGCCGC CCGCGCC	Low complexity	0	24	
		chr3:38588597-38588620		TAAAGATATATTCATATGT GAATA	LINE	83	24	
		chr21:34769880-34769903		TGAAAAAATTTATAAATT ATACT	LINE	92	24	
		chr3:38588657-38588673		AATATATTTTTATATT	LINE	100	17	
		chr3:38585279-38585295		TATCATTTTATATGATT	LINE	88	17	
		chr21:34752251-34752258		TTTATATT	Low complexity, Simple repeat	100	8	
		chr7:150286125-150286130		TGGCCT		33	6	
		chr3:38589382-38589385		TATA	Simple repeat	100	4	
		chr3:38589328-38589331		ATAT	Simple repeat	100	4	
		chr11:73842475-73842478		TATA	Simple repeat	100	4	
		chr11:73839652-73839655		TAAT	Low complexity	100	4	
	no coverage	chr3:38588492-38588494	no coverage in NA17773	CAT	LINE	66	3	
		chr21:34764600-34764601		GA	Low Complexity	50	2	
	SOLiD	no coverage	chr11:73839650-73839651	no coverage in NA17156	AT	Low Complexity	100	2
			chr11:73842034-73842037		TGTT	SINE	75	4
			chr11:73843420-73843423		TGTT		75	4
chr11:73857248-73857256			ATACAAGGT			67	9	
chr2:223621493-223621497			AAAAA			100	5	
chr3:38588555-38588614			TATAAATACATATATGTAT AATTATATAAATATATATTT TTATAAAGATATATTCATA TG		LINE	92	60	
chr3:38589340-38589342			TAT		Simple Repeat	100	3	
chr21:34769887			A		LINE	100	1	
chr11:73839650-73839659			no coverage in NA17275		ATTAATTAAT	Low Complexity	100	10
chr11:73842034-				TGTT	SINE	75	4	

73842037						
chr11:73843420-73843423		TGTT	75	4		
chr2:223621489-223621498		TTAAAAAAAA	100	10		
chr3:38561031-38561035		AAAAA	Low Complexity	100	5	
chr3:38586364-38586366		TTC	LINE	67	3	
chr3:38588485-38588519		ATAGGTGCATATTATATATATAAATATACTTAT	LINE	86	35	
chr3:38588554-38588619		ATATAAATACATATATGTATAATTATATAAATATATATTTTATAAAGATATATTCATATGTGAAT	LINE	91	66	
chr3:38588657-38588668		AATATATTTTTT	LINE	100	12	
chr3:38589933-38589934		AA	LINE	100	2	
chr21:34661459		T	Low Complexity	100	1	
chr3:38589383		A	Low Complexity	100	1	
chr11:73839653-73839654	no coverage in NA17460	AA	Low Complexity	100	2	
chr11:73842034-73842037		TGTT	SINE	75	4	
chr11:73843419-73843423		TTGTT		80	5	
chr11:73844859-73844865		ATATATA	Simple Repeat	100	7	
chr11:73857248-73857256		ATACAAGGT		67	9	
chr2:223621491-223621492		AA		100	2	
chr3:38588493-38588511		ATATTATATATTATAATAT	LINE	100	19	
chr3:38588552-38588613		TTATATAAATACATATATGTATAATTATATAAATATATATTTTTATAAAGATATATTCATAT	LINE	94	62	
chr3:38588656-38588671		AAATATATTTTTTATA	LINE	100	16	
chr3:38589334-38589342		TTTATATAT	Simple Repeat	100	9	
chr3:38589382-38589384		TAT	Simple Repeat	100	3	
chr11:73839649-73839658		no coverage in NA17773	AATTAATTAA	Low Complexity	100	10
chr11:73842034-73842037			TGTT	SINE	75	4
chr11:73843420-73843424			TGTTA		80	5
chr11:73857247-73857255			TATACAAGG		67	9
chr3:38588493-38588496			ATAT	LINE	100	4
chr3:38588552-38588576	TTATATAAATACATATATGTATAAT		LINE	92	25	
chr3:38588611-38588619	TATGTGAAT		LINE	78	9	
chr3:38588661-38588678	TATTTTTTATATTTATAT		LINE	100	18	
chr21:34779357	A		LINE	100	1	

low coverage	chr3:38588449-38588731	≤42 reads in ≥ 2 samples	ATTTATACATATAATAATA TATAAATATGCACTTATAT AGGTGCATATTATATATTA TAATATATACTTATATTTAT ATATAGGTTTATATAAATTG TATATAATTATATAAATAC ATATATGTATAAATTATATA AATATATATTTTTATAAAG ATATATTCATATGTGAATA TATTTATATGGATTATATT TATATATTTAATATACAAA TATATTTTTTATATTTATAT ATTTAATATACAAATACAT TTTATATATTTATATATTTG TACATATATATTTA	LINE	91	283
	chr3:38589292-38589397		TATAATATATACACATGTA ATATATAATATACACACAT ATATTTTATATATATATATA TATTTGTGTGTGTGTGTG TATATATATACATATATAT ATATATATATT	SINE, Simple repeat	87	106
	chr21:34766415-34766484		TCCAAAACTGATTTTAAA AATGAAGCCATGGGTGAA AGAAGTATTTAAAACCTCA AGAAAAATTACTTT	LINE	74	70
	chr21:34764611-34764675		TTAATAATTTAATAGCCA TTAAATTAACAAAATAATA ATAATTAGATTTGGAATTT TTTAAAGA	Low complexity	88	65
	chr3:38609333-38609390		CTTCCAATCCAAGATC TTAAAGCTTTTTTGCTTGT TTTTATTCTAAAACTTTA AA	LINE	72	58
	chr21:34763834-34763891		TATATATACATAAAATAGG AATACAAAACATGCTATTC CTATTATATGTGTAATAGG A	LINE	78	58
	chr3:38585386-38585442		TCTTAAAACAATTTTTGTG GTTTCATTGTTTTACATTA CAATTTTAAAAATTAATA	LINE	84	57
	chr21:34665968-34666016		ATCTTAAAAAGTTCTCATTATTTAAGAAAA AAAAATAAAAAGTTATAAT		88	49
	chr3:38588773-38588820		TATATCTTTTATAGATTTT ATATATTATATTTATGTGT ATGAATATA	LINE	90	48
	chr21:34740239-34740285		CATTCCCTGCTCCGTCGGACGGTGCC CACATTCCCTGCTCCGTCGG		32	47
	chr21:34769787-34769832		ATTTTCAGATTTCCAGAAA AAAAAAGCAGAAAATCA ATGTAACCT	LINE	74	46
	chr3:38584380-38584425		GGAGTTGTTCAAGATAAA AAAAAAGTAGGTGATTG TATTTTTTTC	LINE	74	46
	chr2:223621480-223621521		ACATTTTTTTTTAAAAAAGTTTTATGAA GACTCAAAAGAA		83	42
chr3:38588835-38588875	ACACACACATACACACAC ATTTTTTTTTCTTTTTTGA GAC	SINE	71	41		
chr7:150286114-150286152	CCCGGCGCGCATGGCCTCGATGTCGTC GGCCGACGAGGC		23	39		
chr21:34768590-34768628	AATTAATAAAATTTCTTG GAACTAATAAGTGATTTTA T	LINE	82	39		
chr21:34769868-34769906	AAGCATGATTCATGAAAA AAATTTATAAATTATACTT	LINE	82	39		

			GA			
	chr3:38563947-38563985		ATTCTGAAAATTTATGAAA AAAATTAAGAAAAAATTCT G	LINE	85	39
	chr3:38585314-38585351		TTCATTTAGTAAAATTTTT GATGTTACATTAAGAAAG C	LINE	79	38
	chr3:38586345-38586382		TAAATTAAGAAATATATT TTCTTTCTTTTTTTTTTTT	LINE	92	38
	chr21:34792028-34792064		TGTTCATTTTTAAATTCT TTTTTTTTCTTTTTTTTG	LINE	86	37
	chr11:73857232-73857265		GTATATTGTATACAATATACAAGGTGGGT ATATT		74	34
	chr3:38586704-38586737		AGCACTATTTAGTTTTTAA AAAAATAATTTCTAG	SINE	82	34
	chr7:150288290-150288323		AATAATTTTTCTTTTTTAAA ATTTTTTAACTTTT	SINE	94	34
	chr3:38610256-38610288		ATAATTTTTTCATATGAAA TTTGCATATCTTTT	LINE	85	33
	chr21:34771485-34771517		CTTAATGAATGCAAATTTT GAAAAATTATTTAA	LINE	85	33
	chr3:38589915-38589946		GTTTTTCTTTTTTAAAAA AAATTTCCCTC	LINE	81	32
	chr11:73839639-73839669		TATTTAATTAATTAATTA TTTATTTTTTG	Low complexity	97	31
	chr11:73842457-73842487		TATATAGATATATAGATAT ATAGATATAGAT	Simple repeat	87	31
	chr3:38589536-38589565		ATGCCAGCTAATTTTTAA AAATCTTTTTG	SINE	73	30
	chr11:73850460-73850488		TATGTACACATAGGTTTAAAAA		83	29
	chr21:34760240-34760267		AAAAAAAATGCTTAAGTA TAAAATATT		89	28
	chr21:34766085-34766112		TATATTTTTCTAGAAAA AAAACATGA		86	28
	chr21:34800133-34800160		TTTAACTACAGTATTTTTA AAATAGAAC		82	28
	chr21:34767518-34767545		AAGAAATAAGAAAAATTA TTAAAAATA	LINE	93	28
	chr21:34773303-34773330		TAAAATCTATTTAAAAAA TTAAATAA	LINE	96	28
	chr11:73843778-73843803		TTTATTTTTATTTTTCAAA ATAGAA		92	26
	chr3:38561022-38561047		CTTTTTAAAAAAAAAAAAA CAAAAAA	Low complexity	92	26
	chr21:34765892-34765916		AAATGAATTTTTGAAAAGA AAAATG	LINE	84	25
	chr21:34765980-34766004		CAAAATTTTAAAAACAAA ATTAGC	LINE	84	25
	chr21:34767323-34767347		AAATATTTTGAACAAATG AAAATA	LINE	88	25
	chr11:73844845-73844868		TTAAATATATATATATAT ATAT	Simple repeat	100	24
	chr11:73847952-73847975		ATTTTTTTTTTTTTTTTTT TGAG	SINE	92	24
	chr11:73861890-73861913		TGTCTCAGGGGAAAAAAA AAAAAA	SINE	71	24
	chr21:34779348-34779371		AAAAATTAATTTATTTTT AAAAA	LINE	100	24
	chr21:34661145-34661167		AAAAAAAAAAAAAAAAAAAA AAAAA	SINE	100	23
	chr21:34773600-34773622		AATAAGTATTAATTTATGA AAAT	LINE	91	23
	chr21:34794863-		CATATATAATCTTATTTTT	LINE	87	23

34794885	CAAA			
chr2:223618795-223618817	TTATTTTTTATTTTTTATTT TTT	SINE	100	23
chr3:38656858-38656879	AGTTTAAAAAATGTGGCT GTTA		73	22
chr11:73840545-73840566	TTTTTTTTTTTTTTTTTTTT TA	SINE	100	22
chr21:34752232-34752253	TTAAATTTTATATTATTTAT TT	Simple repeat	100	22
chr21:34785307-34785328	AAAAAAAAAAAAAAAAAAAA AAA	SINE	100	22
chr11:73844432-73844452	TTTTTTTGAGACAGAGCC TTG	SINE	62	21
chr21:34749348-34749368	CTTAAAAAAAAAAAAAAAAAG AA	LINE	90	21
chr21:34754893-34754913	TTTTTTCTTTTTTTCTGA G	SINE	81	21
chr21:34775648-34775668	CAAAAAAAAAAAAAAAAAAA AA	SINE	95	21
chr21:34781297-34781317	ATGTTGGTGTGCTGCACC CAT	LINE	48	21
chr3:38651571-38651591	TATTTTAAAAATTAAAAAA AA	Low complexity	100	21
chr21:34661327-34661346	TCAAAAAAAAAAAAAAAAAA A		95	20
chr21:34764325-34764344	TTCAAGTTGAATTTTTTAT A		85	20
chr11:73839006-73839025	TAGAAAAAAAAAAAAAAAAA A	Simple repeat	95	20
chr11:73839795-73839814	TTTTTTAAAAATTATTTT TA	SINE	100	20
chr11:73840384-73840403	TTTTTTTTTTTTGAGACAG A	SINE	80	20
chr21:34764556-34764575	ATAAAATAAAAATTTTGAA A	Low complexity	95	20
chr21:34752368-34752386	AAAAAAATTTTGCCAGTC A		74	19
chr21:34810583-34810601	GGTTTAAAGCCTTTTAAA		74	19
chr3:38610336-38610354	TGGAACCTTTAAAAATTTA	LINE	84	19
chr21:34661453-34661471	ACTTTTTTATTTTAAAAAA	SINE, Low complexity	95	19
chr21:34662769-34662787	AAAAAACAAAAATTTTTT	SINE	95	19
chr21:34774706-34774724	TTTTTTTTTTTTTTTTGAG	SINE	89	19
chr21:34781207-34781225	TTTTTTTTTTAATATATAT	LINE	100	19
chr21:34748682-34748699	TTTTAAAGAATTTTTTTA		94	18
chr11:73842027-73842044	ATAGGGTTGTTATGAGGG	SINE	56	18
chr21:34740384-34740400	CATTCCCTGCTCCGTCC		35	17
chr21:34758606-34758622	AAGATTTAAAAATTGAG		82	17
chr21:34668223-34668239	AAAAAAAAAAAAAAAAAAAA	SINE	100	17
chr21:34787644-34787660	TTTTTTTATTTTTTAAT	LINE	100	17
chr3:38587375-38587391	TGAATTAATAATTTAT	LINE	94	17
chr3:38608142-	CAAAAAAAAAAAAAATAG	SINE	88	17

38608158
chr21:34738730-34738745
chr21:34789673-34789688
chr21:34802359-34802374
chr3:38601417-38601432
chr11:73843414-73843428
chr21:34810697-34810711
chr7:150283890-150283904
chr11:73841785-73841799
chr3:38575905-38575919
chr3:38613625-38613639
chr7:150286427-150286441
chr21:34778470-34778483
chr11:73859080-73859093
chr21:34768247-34768260
chr21:34770609-34770622
chr21:34776430-34776443
chr21:34779857-34779870
chr21:34803622-34803635
chr21:34813572-34813585
chr3:38636242-38636255
chr2:223625535-223625547
chr11:73845021-73845033
chr21:34771402-34771414
chr21:34790243-34790255
chr7:150295303-150295315
chr21:34669909-34669920
chr21:34816357-34816368
chr11:73844590-73844601
chr3:38619706-38619717
chr7:150277436-150277447
chr3:38609269-38609279
chr21:34788495-

AAAAAAAAAAAAAAAAAAAA	SINE	100	16
CTGGACTTTTTTTTTTTT	LINE	75	16
TTTTTTTTTTTTTTTTTT	SINE	100	16
TCTCAAAAAAAAAAAAAA	SINE	88	16
TAGGGTTGTTATGAG		60	15
TAAAAAAAAATATTTT		100	15
AGGCTCCCCCGCCCC		13	15
TCAAAAAAAAAAAAAAT	SINE	93	15
TTTTTTTTTTTTTTTTTT	SINE	100	15
CAACAAATATTTATT	SINE	87	15
GCGCCCGCGCCGCC	Low complexity	0	15
GGAAAAAAAAAAAAAA		86	14
AAAAAAAAAAAAATTA	SINE	100	14
AAAATCATATGAT	LINE	86	14
AAAATCTGCATGTA	LINE	71	14
AAAAAAAAAAAAAAAAAA	SINE	100	14
AAAAAAAAAAAAAAAAAA	SINE	100	14
TTAACTTTTTTTTTTT	SINE	93	14
TTTTTTTTTTTTTTTTTT	SINE	100	14
TTTTTCTTTTTTCT	SINE	86	14
AAAAAAAAAAAAAAAAAA		100	13
TCAAAAAAAAAAAAAA	SINE	92	13
ATGTAATTTTA	LINE	92	13
ACTTTTTTTTTTTC	SINE, LINE	85	13
AAATTAATTAATT	SINE	100	13
CTGCGGGGCGCC		8	12
AAAAAAAAAAAAAA		100	12
TTTGTATTTTTT	SINE	92	12
TTTTTTTTTTTTTT	SINE	100	12
TTTTTTTTTTTTTT	Simple repeat	100	12
TTTAACAATTT	LINE	91	11
AGGATTTTTTTT	LINE	82	11

34788505				
chr2:223618906-223618916	CATTCTCCTGC	SINE	45	11
chr3:38590562-38590572	TTTTTTTTTTTT	SINE	100	11
chr3:38610782-38610792	TTTTTTTTTTGA	SINE	91	11
chr3:38613419-38613428	GAAAAAAAAA		90	10
chr21:34664204-34664213	TTTAAAAAAC	LINE	90	10
chr21:34783162-34783171	AAAAAAAAAA	SINE	100	10
chr11:73850436-73850444	TTTTTTTTTA		100	9
chr21:34741859-34741867	GGGGCAGCC		11	9
chr11:73859245-73859253	AAAAAAAAAA	SINE	100	9
chr21:34738236-34738244	TAAAAATTA	SINE	100	9
chr21:34771136-34771144	ACCAACATG	LINE	56	9
chr21:34740875-34740882	AATTTTTT		100	8
chr21:34750880-34750887	TTTGAAA		75	8
chr3:38579741-38579748	CAGGCCAA		38	8
chr3:38616065-38616072	TTTTAAAT	LINE	100	8
chr11:73858951-73858958	AAAAAAAA	SINE	100	8
chr21:34658942-34658949	AAAAAAAA	SINE	100	8
chr21:34764767-34764774	GAAATTTT	LINE	88	8
chr3:38589750-38589757	TTTTTTTG	LINE	88	8
chr3:38644238-38644245	AAAAAAAA	SINE	100	8
chr21:34789626-34789632	CAATTCT	LINE	71	7
chr3:38556661-38556667	TTTTTTT	SINE	100	7
chr3:38563736-38563742	AAATAAA	LINE	100	7
chr3:38588256-38588262	TTATATA	Low complexity	100	7
chr7:150289324-150289330	TTTTTTT	SINE	100	7
chr21:34799621-34799626	TTTTTG		83	6
chr21:34774288-34774293	AAAAAA	SINE	100	6
chr21:34665956-34665960	TGCAG		40	5
chr2:223618295-223618299	TTTAA		100	5
chr7:150282296-150282300	AAAAA		100	5
chr21:34764804-34764808	TAAAA	LINE	100	5
chr21:34817959-	TTTTT	SINE	100	5

34817963					
chr7:150291368-150291372		CCAGA	SINE	40	5
chr21:34743183-34743186		TGCT		50	4
chr21:34660979-34660982		TGTA	SINE	75	4
chr21:34739279-34739282		TCTA	SINE	75	4
chr21:34764859-34764862		AACA	LINE	75	4
chr2:223622088-223622091		TAAA	LINE	100	4
chr21:34815282-34815284		TTA		100	3
chr21:34767716-34767718		TTA	LINE	100	3
chr21:34794247-34794249		TAA	Low complexity	100	3
chr7:150275826-150275828		CCG	Low complexity	0	3
chr21:34748441-34748442		AA		100	2
chr21:34815564-34815565		AT		100	2
chr7:150294314-150294315		AA		100	2
chr21:34764814-34764815		TA	LINE	100	2
chr21:34814581		A		100	1
chr2:223625484		T		100	1
chr3:38606725		A		100	1
chr11:73847538		A	SINE	100	1
chr21:34662447		T	Simple repeat	100	1
chr21:34746912		T	SINE	100	1
chr21:34766497		A	LINE	100	1
chr21:34802205		T	SINE	100	1
chr2:223622081		T	LINE	100	1
chr3:38587759		T	SINE	100	1

Supplemental Table 7. Analysis of 23 LR-PCR amplicons for imbalanced allelic amplification. The distribution of the alternate allele read frequency at positions known to be heterozygous based on ABI Sanger data is shown with the data from all three technologies combined. Amplicons that do not follow the expected AARF distribution from Supplemental Figure 1 (with $p < 0.01$) are highlighted in grey and were removed from subsequent analysis.

Amplicon name	Amplicon coordinate	Sample	p-value	# of Sanger Het positions by 3 NGS	Distribution of AARF						
					0-10	10-20	20-40	40-60	60-80	80-90	90-100
LPCR_382423_11004	chr2:223616214-223627218	NA17156		3*2	0	0	1	3	2	0	0
		NA17460		3*6	0	1	5	10	2	0	0
		NA17275		3*8	3	1	7	12	1	0	0
		NA17773		3*3	0	0	4	5	0	0	0
LPCR_721496_11101	chr3:38553978-38565079	NA17773		3*1	0	0	1	2	0	0	0
LPCR_721498_8720	chr3:38575485-38584205	NA17156		3*1	0	0	2	1	0	0	0
LPCR_721500-_11083	chr3:38595323-38606406	NA17275	10 ⁻⁷	3*2	6	0	0	0	0	0	0
		NA17773		3*1	0	0	0	3	0	0	0
LPCR_721501_11969	chr3:38606353-38618322	NA17275		3*1	0	0	0	3	0	0	0
LPCR_721502_11967	chr3:38618395-38630362	NA17156		3*11	0	0	9	22	2	0	0
		NA17460		3*3	0	0	3	6	0	0	0
		NA17275		3*12	1	1	2	31	1	0	0
		NA17773		3*14	2	1	10	27	2	0	0
LPCR_721503_11821	chr3:38630330-38642151	NA17156		3*1	1	0	1	1	0	0	0
		NA17460		3*4	0	0	1	9	2	0	0
		NA17275		3*4	1	0	3	7	1	0	0
		NA17773		3*1	0	0	0	3	0	0	0
LPCR_721504_11891	chr3:38642100-38653991	NA17156		3*3	0	0	2	7	0	0	0
		NA17460		3*7	0	0	7	13	1	0	0
		NA17275		3*3	0	0	6	3	0	0	0
		NA17773		3*2	0	0	2	1	3	0	0
LPCR_721505_11811	chr3:38653478-38665289	NA17156		3*3	0	0	4	2	3	0	0
		NA17460		3*4	0	0	5	7	0	0	0
		NA17275		3*5	0	0	5	10	0	0	0
		NA17773		3*7	0	0	6	13	2	0	0
LPCR_54275_11143	chr7:150269610-150280753	NA17156	0.0001	3*1	3	0	0	0	0	0	0
		NA17460		3*6	5	1	3	8	1	0	0
		NA17773		3*8	3	0	7	12	1	1	0
LPCR_54276_4905	chr7:150280713-150285618	NA17156		3*3	0	0	3	4	2	0	0
		NA17275		3*2	0	0	1	5	0	0	0
		NA17773		3*2	0	0	0	4	2	0	0
LPCR_1527027_3087	chr7:150284619-150287706	NA17156		3*3	0	0	2	7	0	0	0
		NA17460		3*3	0	0	3	5	1	0	0
		NA17275		3*1	0	0	0	2	0	1	0
		NA17773		3*3	0	0	3	5	1	0	0
LPCR_54277_11487	chr7:150286689-150298176	NA17156		3*1	0	0	1	2	0	0	0
		NA17460		3*3	0	0	3	5	1	0	0

		NA17275		3*2	0	0	1	5	0	0	0
		NA17773		3*2	0	0	1	5	0	0	0
LPCR_54278_3423	chr7:150298170-150301593	NA17156		3*2	0	0	1	5	0	0	0
		NA17460		3*2	0	0	4	1	1	0	0
		NA17275		3*2	1	0	2	3	0	0	0
		NA17773		3*4	3	0	3	6	0	0	0
LPCR_240580_9001	chr11:73836950-73845951	NA17156	< 10 ⁻¹⁶	3*13	0	0	1	2	22	10	4
		NA17460		3*9	2	1	5	10	9	0	0
		NA17275		3*9	1	0	2	11	13	0	0
		NA17773	2*10 ⁻⁹	3*10	0	0	1	4	21	4	0
LPCR_240581_8458	chr11:73845929-73854387	NA17156		3*2	0	0	0	6	0	0	0
		NA17460		3*2	0	0	1	5	0	0	0
		NA17275		3*3	0	0	2	5	2	0	0
		NA17773		3*3	0	0	4	4	1	0	0
LPCR_1977_14227	chr21:34657259-34671486	NA17460	0.0008	3*9	18	0	0	0	0	0	9
		NA17275	3*10 ⁻⁷	3*8	6	0	0	0	1	6	11
		NA17773	0.002	3*9	16	1	1	0	0	0	9
LPCR_1983_10796	chr21:34735911-34746707	NA17156		3*7	0	0	4	15	2	0	0
		NA17460		3*2	0	0	2	3	1	0	0
		NA17773		3*4	1	1	3	7	0	0	0
LPCR_778623_3341	chr21:34746238-34749579	NA17460		3*1	0	0	1	1	1	0	0
		NA17773		3*2	0	0	1	5	0	0	0
LPCR_778624_11646	chr21:34749486-34761132	NA17156		3*4	0	0	2	8	2	0	0
		NA17460		3*1	0	0	1	2	0	0	0
		NA17773		3*2	0	0	1	5	0	0	0
LPCR_1988_4624	chr21:34795398-34800022	NA17156		3*1	0	0	1	1	1	0	0
		NA17460		3*1	0	0	1	2	0	0	0
LPCR_1989_6410	chr21:34799969-34806379	NA17156		3*2	0	0	2	3	1	0	0
		NA17460		3*1	0	0	1	2	0	0	0
LPCR_1990_12103	chr21:34806347-34818450	NA17156		3*5	0	0	0	14	1	0	0
		NA17460		3*2	1	0	1	4	0	0	0

Supplemental Table 8. Accuracy of NGS and Sanger technologies assessed by genotyping SNPs in the Illumina Human Hap550 BeadChip.

	NA17156	NA17275	NA17460	NA17773	All Individuals Combined
ABI Sanger	100% (24/24)	96% (24/25)	96% (23/24)	100% (22/22)	98% (93/95)
Roche 454	99% (76/77)	97% (75/77)	98% (78/80)	96% (76/79)	97.4% (305/313)
Illumina GA	100% (77/77)	100% (77/77)	100% (80/80)	100% (79/79)	100% (313/313)
ABI SOLiD	100% (77/77)	100% (77/77)	100% (80/80)	99% (78/79)	99.7% (312/313)

Supplemental Table 9. Discordant genotypes (in bold) between SNPs from the Illumina HumanHap550 array and the sequencing technologies.

Individual	Location	dbSNP rs id	HumanHap550 Genotype	ABI Sanger Genotype	Roche 454 Genotype	Illumina GA Genotype	ABI SOLiD Genotype
NA17275	chr11:73843968	rs7789	GT	TT	GT	GT	GT
NA17460	chr11:73843968	rs7789	GT	TT	GT	GT	GT
NA17773	chr11:73861481	rs633335	CC	-	CT	CC	CC
NA17275	chr21:34743289	rs2070357	CC	CC	CT	CC	CC
NA17773	chr21:34743289	rs2070357	CT	CT	TT	CT	CT
NA17773	chr21:34761833	rs11701051	CT	-	CT	CT	TT
NA17460	chr21:34806395	rs11702354	AG	AG	GG	AG	AG
NA17773	chr21:34806395	rs11702354	AA	AA	GG	AA	AA
NA17275	chr3:38606427	rs7373157	GT	-	GG	GT	GT
NA17156	chr3:38630647	rs9856587	CT	CT	CC	CT	CT
NA17460	chr3:38635940	rs12498069	AC	-	CC	AC	AC

Supplemental Table 10. Comparison of ABI Sanger platform base calls at 258,879 bp (4 samples) sequenced by 3 NGS Technologies.

		Sanger			
		Ref	Het	Alt	N/N
Roche 454	Ref	258,290	12	1	98
	Het	16	203	6	14
	Alt	1	1	104	2
	N/N	131	0	0	0
Illumina GA	Ref	258,388	3	0	96
	Het	28	217	0	15
	Alt	3	0	111	3
	N/N	15	0	0	0
ABI SOLiD	Ref	257,919	6	0	92
	Het	28	200	3	13
	Alt	8	6	108	1
	N/N	480	8	0	7

Supplemental Table 11. Quality metrics for the four DNA Samples in the three NGS technologies when compared to ABI Sanger data (Figure 4).

		Sequencing Accuracy	Variant Accuracy	Coverage Rate	False Positive Rate	False Negative Rate	Variant Discrepancy Rate
Roche 454	ALL	0.99990	0.948	0.9996	0.025	0.031	0.022
	NA17156	0.99990	0.956	0.9994	0.022	0.011	0.034
	NA17275	0.99988	0.919	0.9999	0.033	0.048	0.034
	NA17460	0.99990	0.946	1.0000	0.022	0.032	0.022
	NA17773	0.99993	0.962	0.9990	0.026	0.038	0.000
Illumina GA	ALL	0.99991	1.000	1.0000	0.063	0.000	0.000
	NA17156	0.99992	1.000	1.0000	0.053	0.000	0.000
	NA17275	0.99990	1.000	1.0000	0.086	0.000	0.000
	NA17460	0.99994	1.000	1.0000	0.041	0.000	0.000
	NA17773	0.99990	1.000	0.9999	0.080	0.000	0.000
ABI SOLiD	ALL	0.99985	0.963	0.9984	0.078	0.009	0.028
	NA17156	0.99983	0.967	0.9979	0.072	0.000	0.033
	NA17275	0.99990	0.984	0.9987	0.076	0.000	0.016
	NA17460	0.99982	0.921	0.9984	0.065	0.034	0.047
	NA17773	0.99985	0.988	0.9985	0.101	0.000	0.013

Supplemental Table 12. Discordant genotypes between NGS platforms and ABI Sanger

Found by	NCBI36_sample	Upstream Sequence	Downstream Sequence	Repeats	ABI Sanger genotype	NGS genotypes			Category	
						Roche 454	Illumina GA	ABI SOLiD		
False Positive	Roche 454 only	chr21_34774913_NA17275	GTTTTGCCTTGTTAGCCAAGCTGGTCTC	AACTCCTGACCTCAGGTGATCCACCTGCTT	AluSx	GG	AG	GG	GG	repetitive element & in/del
		chr21_34774913_NA17773	GTTTTGCCTTGTTAGCCAAGCTGGTCTC	AACTCCTGACCTCAGGTGATCCACCTGCTT	AluSx	GG	AG	GG	GG	repetitive element & in/del
		chr2_223627087_NA17275	GGTGGTTCAGGAGACTCTTCTGATCTT	CTAGAAGGGGTAAAGTGGGGTGAACAAGG		TT	CT	TT	TT	
		chr3_38602150_NA17156	ATTGGATGGGTGGGTAGCTGGGTAGATG	GTGGATGGTGTGTGTGTGCCCTTGCCAA	(TGGA) _n	AA	AT	AA	AA	simple repeat & in/del
		chr3_38602150_NA17460	ATTGGATGGGTGGGTAGCTGGGTAGATG	GTGGATGGTGTGTGTGTGCCCTTGCCAA	(TGGA) _n	AA	AT	AA	AA	simple repeat & in/del
		chr3_38602150_NA17773	ATTGGATGGGTGGGTAGCTGGGTAGATG	GTGGATGGTGTGTGTGTGCCCTTGCCAA	(TGGA) _n	AA	AT	AA	AA	simple repeat & in/del
	Illumina GA only	chr11_73842453_NA17275	GATACAGAGATACAGAGATACAGAGATA	AGATATATAGATATATAGATATATAGATAT	(CAGAG A) _n	CC	CC	CT	CC	simple repeat
		chr11_73845036_NA17275	TAGGGAGACTGTCTCAAAAAAAAAAAAA	AAAAGAAAAGAAAAGTTTCTCACATCA	AluSx	GG	GG	AG	GG	homopolymer
		chr21_34749366_NA17156	TTCCTTGGAACTTAAAAAAAAAAAAAAAA	AAACAAAAACAACCAACCAAAAATATATCT	L1PA10	GG	GG	AG	GG	homopolymer & in/del
		chr21_34749366_NA17275	TTCCTTGGAACTTAAAAAAAAAAAAAAAA	AAACAAAAACAACCAACCAAAAATATATCT	L1PA10	GG	GG	AG	GG	homopolymer & in/del
		chr21_34749366_NA17460	TTCCTTGGAACTTAAAAAAAAAAAAAAAA	AAACAAAAACAACCAACCAAAAATATATCT	L1PA10	GG	GG	AG	GG	homopolymer & in/del
		chr21_34749366_NA17773	TTCCTTGGAACTTAAAAAAAAAAAAAAAA	AAACAAAAACAACCAACCAAAAATATATCT	L1PA10	GG	GG	AG	GG	homopolymer & in/del
		chr3_38563723_NA17275	TGTGCTGTGGGAGACAGTATGCGAGTTC	TAAAAAATTAATAAATAAAATTGCCATATG	L1MB3	CC	CC	AC	CC	repetitive element
		chr3_38564670_NA17156	ACAACATCTGGCACTTGATGTCTGTCTCCT	CCCTCTGCTGGTCATTGTGCAGTTCTGGGA		TT	TT	CT	NN	in/del
		chr7_150279372_NA17773	ATGGTGGCCCTGGAGTCTCTAAGTTCC	GGGCTCACTCTGGCCCGCTAGCAGCCTC		AA	AA	AG	AA	in/del
		chr7_150286149_NA17773	CGCATGGCCTCGATGTCGTCGGCCGACG	GGCGCGGCGCAGCTGGCGCAGCTTTCTCG		AA	AA	AG	AA	low coverage
		chr7_150298286_NA17460	GGGGAGAAAAGAGGCCCACTCTCCCCC	CACACACCCCAAGCTGGCCCTCTGCTGC		TT	TT	CT	TT	homopolymer
	ABI SOLiD only	chr11_73842038_NA17460	GAATACCTACTTACTACATAGGGTTGTT	TGAGGGTTTTAAAGCTAAATGAATCAAG	MIR	AA	AA	AA	CC	repetitive element
		chr11_73843792_NA17460	AGAACCCAAGGCTTTTATTTTTATTTT	TCAAAATAGAAGTACCTTTTTCTTTTAT		TT	TT	TT	CT	
		chr21_34741405_NA17275	CTAAGAAGGGTCTGGCTTACACAGGGGC	ATGAGACGTGGCAGGCATAGCTGGGCTGCT		CC	CC	CC	TT	
chr21_34741865_NA17773		ACGGAGGCCCTCTCTATCTGCTGGGGCA	CCCTCCGGTGCCCGCTGGAAGGCAGAGC		GG	GG	GG	CG	in/del	
chr21_34749352_NA17275		ACATCCTGCACATGTTCCCTGGAACTTA	AAAAAAAAAAAAAGAAACAAAAACAACCAA	L1PA10	AA	AA	AA	AT	homopolymer & in/del	
chr21_34774705_NA17156		AGAAAATGAAATTGAGGAGGGGACTCA	TTTTTTTTTTTTTTTGGAGACAGAGTCTCA	AluSx	CC	CC	CC	CT	homopolymer & in/del	
chr21_34807310_NA17460		CCAAGATCAATGTTTTTCTTTATCC	GATTTTAAATCAAGTACACTTTATTGAGG		TT	TT	TT	CT		
chr21_34807310_NA17773		CCAAGATCAATGTTTTTCTTTATCC	GATTTTAAATCAAGTACACTTTATTGAGG		TT	TT	TT	CT		
chr3_38589927_NA17773		TTGCTGTGAGATCTCTGTTTTTCTTTT	TTAAAAAATTTTCCCTCAGCAATTTGGA	L1MEe	TT	TT	TT	CT	homopolymer	
chr3_38596373_NA17156		GCACTCCCGGCTTGCCTGCGCGCACGCG	GCGCACACACACACACACCCCCACACAC	(CA) _n	CC	CC	CC	TT	simple repeat & in/del	
chr3_38602169_NA17773		GGGTAGATGAGTGGATGGTGTGTGTGTG	CCCTTGCCAACTTACCACAAGTTGCCAA		GG	GG	GG	GT	simple repeat & in/del	
chr3_38643193_NA17156		TCAGATCCCAGGGAGTTAAAAAAAAAAA	CCACCCAAAGAAAAGGTAATTAACCTGGA		AA	AA	AA	AC	homopolymer	
chr3_38645101_NA17156		TAGAGAGCCACACCCCGGAGTGAGGC	AGAGACGGGCTCAGAGCAAGAAAAGCTGGC		CC	CC	CC	TT		
chr7_150276467_NA17460		CCCCTTCTCCCTCCCCGCTCACCC	TGTCCTGCGCCTGCGGAAGGACAACCTGC		TT	GT	GT	GG		

		chr7_150279376_NA17773	TGCCCCCTGGAGTCTCTAAGTTCAGGG	CTCACTCTGGCCCGCTAGCAGCCTCAGTT		CC	CC	CC	CG	in/del	
		chr7_150290648_NA17156	ACTCACACTTACACACACACACGAGAGA	AGAGAGAGAGAGAGAGAGAGAGAGAGGAGT	(GA)n	CC	CC	CC	CG	simple repeat & in/del	
		chr7_150290648_NA17275	ACTCACACTTACACACACACACGAGAGA	AGAGAGAGAGAGAGAGAGAGAGAGAGGAGT	(GA)n	CC	CC	CC	CG	simple repeat & in/del	
		chr7_150290648_NA17460	ACTCACACTTACACACACACACGAGAGA	AGAGAGAGAGAGAGAGAGAGAGAGAGGAGT	(GA)n	CC	CC	CC	CG	simple repeat & in/del	
	Roche 454 and Illumina GA	chr21_34741860_NA17156	GCAGAACGGAGGCCCTCTATCTGCTG	GGCAGCCCTCCGGTGCCCCGCTGGAAGGC		GG	CG	CG	NN	in/del	
		chr7_150276467_NA17460	CCCCTTCTCCCTCCCCGCTCACCC	TGTCCGTGCGCTGCGGAAGGACAACTTGC		TT	GT	GT	GG		
	Illumina GA and ABI SOLiD	chr2_223624619_NA17773	ATAATCCTACCTGCAGAAAGACCCCCC	CCGCCAGGCACAACGATTTTACAGACGAGG		CC	CC	CG	CG	homopolymer & in/del	
		chr3_38564670_NA17275	ACAACATCTGGCACTTGATGTCTGTCT	CCCTCTGTGGTTCATTGTGCAGTTCTGGA		TT	TT	CT	CT	simple repeat & in/del	
		chr3_38564670_NA17460	ACAACATCTGGCACTTGATGTCTGTCT	CCCTCTGTGGTTCATTGTGCAGTTCTGGA		TT	TT	CT	CT	simple repeat & in/del	
		chr3_38564670_NA17773	ACAACATCTGGCACTTGATGTCTGTCT	CCCTCTGTGGTTCATTGTGCAGTTCTGGA		TT	TT	CT	CT	simple repeat & in/del	
		chr3_38596386_NA17773	GCGTGCGCGCACGCGCGCACACACAC	CACACACCCCCACACACACCACACACACC	(CA)n	AA	AA	AC	AC	simple repeat & in/del	
		chr3_38596396_NA17156	ACGCGCGCGCACACACACACACACACC	CACACACACCACACACACCAGGAGCCAGA	(CA)n	CC	CC	AA	AA	simple repeat & in/del	
		chr3_38596400_NA17156	GCGCGCACACACACACACACACCCAC	CACACCACACACACCAGGAGCCAGAGTCA	(CA)n	AA	AA	CC	CC	simple repeat & in/del	
		chr3_38638394_NA17275	TTACTGAAGTGGGATAGCCCTGGGGTA	CTCCTAGTGCCTACCTCTAGCTGCCTAC	MIR	CC	CC	CG	CG	repetitive element & in/del	
	chr3_38638394_NA17773	TTACTGAAGTGGGATAGCCCTGGGGTA	CTCCTAGTGCCTACCTCTAGCTGCCTAC	MIR	CC	CC	CG	CG	repetitive element & in/del		
	All 3 NGS	chr11_73843968_NA17275	TCAGAGTTCTGCTATACACAAATACGCA	GTTTCAATGGTACTTCCAGGGACCGCATG		TT	GT	GT	GT		
		chr11_73843968_NA17460	TCAGAGTTCTGCTATACACAAATACGCA	GTTTCAATGGTACTTCCAGGGACCGCATG		TT	GT	GT	GT		
		chr21_34759676_NA17460	ATGGGTAGGAAAGAACAATAAATCTACT	GCACAATGTTGATATTTAAAGATAAACAT		CC	CT	CT	CT		
		chr21_34807876_NA17156	ATCTATCCAATCCAATTGCGTGTGAAC	TCTACTATGAGCCATTGGTACCAGGAATC		GG	AG	AG	AG		
		chr3_38596474_NA17156	GGAACAGAGAAGGAAATGAGAGACAGC	GGAATCCCCCTCTGTGCTGGAGATCATTG		CC	TT	TT	TT		
		chr3_38639168_NA17156	CAATCCCCCTCCACCCAGGCTGGTCTCT	TCCCTCCCCGGCACTTGACGCTGGCCCC		TT	GT	GT	GT		
		chr7_150284380_NA17773	CAGGCGGTGGGAGCAGGGCCAGGAGCCC	CCGAGGTAGCAGGGCAGCTCCCTTACATGA		GG	AG	AG	AG		
		chr7_150293045_NA17773	CAAGAGGCCAACAGTGAATGAGAGAGA	GGAGGGAGAGACAGAGACAGGGAGAGAG	GA-rich	AA	AG	AG	AG		
	chr7_150293190_NA17156	CAGGTTGAGGGGCCCAAGCTCAGACCC	GACCCACAGAAGCAGCATCTCCAGACAGA		TT	GT	GT	GT			
	False Negative	Roche 454 only	chr11_73843069_NA17275	TAACTCACAACAAAATAATTTGTATTA	TTTATGATTATGCCATGGAGTGTGAGGAC		AT	TT	AT	AT	in/del
			chr11_73845036_NA17460	TAGGGAGACTGTCTCAAAAAAAAAAAAA	AAAAGAAAAGAAAAGAAGTTTCTCACATCA	AluSx	AG	GG	AG	NN	homopolymer
			chr21_34743289_NA17773	CAGTGAAACAGTGTCAATTAATCCACCCC	CACCCCTTACAACAGCCAAAAAGAAATCCA		CT	TT	CT	CT	
			chr21_34806395_NA17460	AGTGTCAAATGCGTTGTTCCCTGGAGAG	AAGGCTTATTTCCAGAAAAGTACTCTGACGT		AG	GG	AG	AG	
chr21_34806395_NA17773			AGTGTCAAATGCGTTGTTCCCTGGAGAG	AAGGCTTATTTCCAGAAAAGTACTCTGACGT		AA	GG	AA	AA	In/del	
chr3_38622646_NA17773			TAGATCTTCCCTGCGGACCTGAGGGTCT	GGGGAGCAAGGGGGCAGAGGTCAACCCTCAC		GT	GG	GT	GT		
chr3_38630647_NA17156			CCCACCAGCCCTCTCCCTTGCCCCCT	CCACGGGGTGGGGGAGTGTGTGGATAG		CT	CC	CT	CT	in/del	
chr3_38633238_NA17275			CAGGGGCAGAAAGAGCAAGGGTGGTGG	TGAGAATCCTATCCCATCACCAGCCCCACT		GT	GG	GT	GT	in/del	
chr7_150275327_NA17460			CTGGACCAGACTCCAGGGCGTCCCCC	ACCCACCTGCACCTCCCTCACCTGTTGAGC		CT	CC	CT	CT	homopolymer	
chr7_150293045_NA17275		CAAGAGGCCAACAGTGAATGAGAGAGA	GGAGGGAGAGACAGAGACAGGGAGAGAG	GA-rich	AG	AA	AG	AG	simple repeat		
ABI SOLiD only	chr11_73844514_NA17460	TCTTGGCTTACTGCAACCTCCACCTCCC	GGTTCAAACAATTTCTCTGCTCAGCCTCC	AluSx	AG	AG	AG	GG	repetitive element		
	chr7_150278902_NA17460	AGAGGTTCCCTCTGCCACCCACTCTT	CCAGCCTGCCACCCACTGGCCACGCTCTGG		CT	CC	CT	CC	adjacent variants		

		chr7_150278903_NA17460	GAGGTTCCCCTGTCACCCCACTCTTC	CAGCCTGCCACCCACTGGCCACGCTCTGGT		CG	CC	CG	CC	adjacent variants
All 3 NGS		chr7_150299914_NA17773	CTGTGAGGACTGGCAGAGGAGGAGCCAC	GCCCCATGGCGATGTGGGAGCACAGGTGT		AC	AA	AA	AA	
		chr7_150275444_NA17773	TGGAGAGGGGATGTTGAGGAGGCTGGG	GTGGGGCGGGCATCGAGGAGCTCCTGG		AG	GG	GG	GG	
		chr2_223624557_NA17275	CAGGCAGCCTCAATCTTAGAAGGGTTA	GCTGAAAGGGTCTCAAAGGTCACGTGGTT		AG	AA	AA	AA	
Variant Discrepant Calls	Roche 454 only	chr21_34743289_NA17275	CAGTGAACAGTGTCTTAATCCACCCC	CACCCCTTACAACAGCCAAAAAGAAATCCA		CC	CT	CC	CC	
		chr3_38643196_NA17156	GATCCCAGGGAGTTAAAAAACC	CCCAAAGAAAGGTAATTAAGCTGGAGGG		GG	AG	GG	GG	
		chr7_150279919_NA17460	GGGCAAGGGGGCAAGGGAGGAGGGGAG	TGCTGCGCCCTCAGAGCGAGCATCAGAGG		CC	CG	CC	CC	
		chr7_150286470_NA17275	GCGCCCGCCGACCGCACCGACTCCC	GGCCGTCAGCGCCAGCAGCGGGCAGCTT		AG	AA	AG	NN	
		chr7_150293045_NA17156	CAAGAGGCCAACCCAGTGAATGAGAGAGA	GGAGGGAGAGAGACAGAGACAGGGAGAGAG	GA-rich	GG	AG	GG	GG	simple repeat
		chr7_150293045_NA17460	CAAGAGGCCAACCCAGTGAATGAGAGAGA	GGAGGGAGAGAGACAGAGACAGGGAGAGAG	GA-rich	GG	AG	GG	GG	simple repeat
	ABI SOLiD only	chr11_73842457_NA17460	CAGAGATACAGATACAGATACAGA	ATATAGATATATAGATATATAGATATAGAT	(TA)n	GG	GG	GG	GT	simple repeat
		chr11_73843069_NA17460	TAAGTACAACAAAATAATTTGTATTA	TTTATGATTATGCCCATGGAGTGTGAGGAC		AT	AT	AT	AA	
		chr11_73845278_NA17275	CAACATATTAATGTGGCAGTATATTTTC	TAATGTTTGCAAAGTAAGATATTTTAGAA	Arthur1	AG	AG	AG	GG	repetitive element
		chr11_73845278_NA17460	CAACATATTAATGTGGCAGTATATTTTC	TAATGTTTGCAAAGTAAGATATTTTAGAA	Arthur1	AG	AG	AG	GG	repetitive element
		chr21_34746100_NA17460	GTCCTGTCAGAGACTGGCCCCTGCCTCA	TGAGCTGAGTGGCAACCCACTCACTCAA		CT	CT	CT	CC	
		chr3_38592130_NA17156	AGGGCCGTGGGACCACCCGTGGGGGCC	CACGCTCTGCGGGCACTTTTGAGGAGGAT		GG	GG	GG	CG	
		chr7_150279131_NA17773	TGGCCGACAGTTGCCGAAGATGCTAGC	TACATGAGGGCTGGGGCGTGGGCACGTGG		AG	AG	AG	GG	
		chr7_150283286_NA17156	AGGAGCCAGGCGCGGGTGAAGAGGGCCA	GCCCCACGGTCCCAAAGCTTCTACTTCC		AG	AG	AG	AA	
Roche 454 and ABI SOLiD	chr7_150298282_NA17156	CTCTGGGGAGAAAAGAGGCCAGTCTCC	CCCTCACACCCCCAAGCTGGCCCTTCTG		TT	CT	TT	CT	in/del	

Supplemental Table 13. Analysis of eleven heterozygous indels identified by ABI Sanger. Allele1 and Allele2 refer to the base calls detected at the locus. Homopolymer Length is the length of the homopolymeric region flanking the reported indel.

ABI SANGER						Roche 454		
NCBI36	Sample	Allele1	Allele2	Homopolymer Length	resolved by 454	NCBI36	Allele1	Allele2
chr21_34795794	NA17773	-	T	5	no			
chr21_34806868	NA17156	-	T	6	no			
chr2_223624624	NA17773	-	GCC		yes	chr2_223624624	-	GCC
chr3_38633475	NA17156	-	GAGGGCTGCAAGCTG		yes	chr3_38633476	-	GAGGGCTGCAAGCTG
chr3_38633475	NA17275	-	GAGGGCTGCAAGCTG		partial	chr3_38633465	A	-
						chr3_38633469	CTG	GCAA
chr3_38633475	NA17460	-	GAGGGCTGCAAGCTG		yes	chr3_38633476	-	GAGGGCTGCAAGCTG
chr3_38638411	NA17275	CCTCCTAGCTGCCTAC	G		yes	chr3_38638394	CCTCCTAGCTGCCTAC	G
chr3_38638411	NA17773	CCTCCTAGCTGCCTAC	G		yes	chr3_38638394	CCTCCTAGCTGCCTAC	G
chr7_150282302	NA17460	-	A	11	no			
chr7_150282302	NA17773	-	A	11	no			
chr7_150299420	NA17156	-	G	6	no			

Supplemental Table 14. Forty three additional indels not found by ABI Sanger and identified by Roche 454 in the 88-kb overlapping ABI Sanger sequences

Coord NCBI36	Sample	alt/ref	Total Reads	Alt reads	upstream sequence	downstream sequence	repeat
chr11_73841786	NA17460	-/A	23	8	GATTAGGCCACTGCATCCAGCCTGAGCCACAGAGCGAGACCCCTGTCT	AAAAAAAAAAAAATCTGCCTCAAGGTCTGTTTCTAGGAAACCAACTTCA	AluJo
chr21_34742194	NA17773	T/-	24	15	GGTTTGTGGCTGTGCTACTGCACCCTCTGCCTCTGTACGGCATGGTG	CCTCCCTATGCATCTGTGTCTGAATTTCCCTCTCCGATAAGGACTCCAG	MLT1C
chr21_34749371	NA17275	A/-	11	5	CACATCCTGCATGTTCCTTGGAACTTAAAAAAAAAAAAAAAAAGAAAC	AAAACAACCAACCAAAAAATATCTAAAAATGCATCTGTTAGCAATTGAC	
chr21_34774705	NA17156	C/-	25	14	TATCTCTGCAAAGAGAGGAAAGAAAATGAAATTGAGGAGGGGGACTCA	TTTTTTTTTTTTTTTTGAGACAGAGTCTACTCTATTGCCAGGCTGGAG	AluSx
chr21_34774890	NA17275	GCCT/CACCA	17	4	TACAGGCGCACACCACCACACCTGTATTTTTGTAGAGATGGAGTTTT	CCTTGTTAGCCAAGCTGGTCTCGAACTCTGACCTCAGGTGATCCACCTG	AluSx
chr21_34774898	NA17275	AGCCAA/GGTCAG	17	4	CACACCACCACACCTGTATTTTTGTAGAGATGGAGTTTTGCCTTGT	GCCAAAGCTGGTCTCGAACTCTGACCTCAGGTGATCCACCTGCTTTGGTC	AluSx
chr21_34774884	NA17773	A/-	12	3	TGAGACTACAGGCGCACACCACCACACCTGTATTTTTGTAGAGATGG	GTTTTGCCTTGTAGCCAAGCTGGTCTCGAACTCTGACCTCAGGTGATC	AluSx
chr21_34774890	NA17773	GCCT/CACCA	13	4	TACAGGCGCACACCACCACACCTGTATTTTTGTAGAGATGGAGTTTT	CCTTGTTAGCCAAGCTGGTCTCGAACTCTGACCTCAGGTGATCCACCTG	AluSx
chr21_34774898	NA17773	AGCCAA/GGTCAG	15	5	CACACCACCACACCTGTATTTTTGTAGAGATGGAGTTTTGCCTTGT	GCCAAAGCTGGTCTCGAACTCTGACCTCAGGTGATCCACCTGCTTTGGTC	AluSx
chr21_34774940	NA17773	GCTTTGGT/CGGC	13	3	CTTGTTAGCCAAGCTGGTCTCGAACTCTGACCTCAGGTGATCCACCT	CCTTGCTCCCAAAGTCTGGGATTATAGGCATGAGCCACCATGCCTGA	AluSx
chr21_34807620	NA17156	CT/TC	9	6	GATGACCATAACCTACCACAGACAGGGTCTCTCTCTCTCTCTCTCT	TTTTCCCTGTTCTTGCGCCAGCTGCCTGACAATTTGTGGGATTAAGCT	(TC)n
chr21_34807620	NA17460	TC/TC	15	13	GATGACCATAACCTACCACAGACAGGGTCTCTCTCTCTCTCTCTCT	TTTTCCCTGTTCTTGCGCCAGCTGCCTGACAATTTGTGGGATTAAGCT	(TC)n
chr21_34807620	NA17773	CTTTTC/TCTTTT	12	8	GATGACCATAACCTACCACAGACAGGGTCTCTCTCTCTCTCTCTCT	TTTTCCCTGTTCTTGCGCCAGCTGCCTGACAATTTGTGGGATTAAGCT	(TC)n
chr21_34807776	NA17156	-/ATCC	21	6	TATTTCTTTTCAGTGACTGCTCACTCTCTCTCTCTCTCTCTCTCTCT	TCCATCCATCCATCCATCCATCCATCCATCCATCCATCCATCCAACTTTC	(TCCA)n
chr3_38564678	NA17156	- /TTTTCTACTCTCTTCT CCCCTCCT	42	31	CGATTAGTTCAGACAACATCTGGCACTTGATGTCTGTCTTCCCTCCT	CTGGTCATTGTGCAGTTCTGGAAATTA AAAAGGTGACAGCCAGGCTAAAA	
chr3_38564674	NA17275	- /TCCTTTTTCTACTCTC TTCTCCCC	52	38	TGCTCGATTAGTTCAGACAACATCTGGCACTTGATGTCTGTCTTCCC	CCTGCTGGTCATTGTGCAGTTCTGGAAATTA AAAAGGTGACAGCCAGGCT	
chr3_38564678	NA17460	- /TTTTCTACTCTCTTCT CCCCTCCT	60	42	CGATTAGTTCAGACAACATCTGGCACTTGATGTCTGTCTTCCCTCCT	CTGGTCATTGTGCAGTTCTGGAAATTA AAAAGGTGACAGCCAGGCTAAAA	
chr3_38564678	NA17773	- /TTTTCTACTCTCTTCT CCCCTCCT	52	39	CGATTAGTTCAGACAACATCTGGCACTTGATGTCTGTCTTCCCTCCT	CTGGTCATTGTGCAGTTCTGGAAATTA AAAAGGTGACAGCCAGGCTAAAA	
chr3_38593427	NA17773	-/T	33	7	GAAGGGAGGCAGCATCTTCATGACGCACCCTGGGACCTGCACAGAGCC	CCTTTCCTGTGTCAGGGCTGGGCCAAGGCTGGACCTCGGGGTTCCCGGGTC	
chr3_38596394	NA17156	CCCCACA/ACACACC	24	12	CACCTCCCGCTTGCCTGCGCGCACGCGCGCACACACACACACACAC	CCCACACACACACACACACCCAGGAGCCAGAGTCAATGGGCTCAAGAGA	(CA)n
chr3_38596370	NA17773	GCGCGCGC/-	22	14	GACTTCCCTCATGAATCCCTGTGGCACTCCCGCTTGCCTGCGCGCAC	CGCGCGCACACACACACACACACCCACACACACACACACCCAGGA	(CA)n
chr3_38596394	NA17773	C/ACA	26	13	CACCTCCCGCTTGCCTGCGCGCACGCGCGCGCACACACACACACAC	CCCACACACACACACACACCCAGGAGCCAGAGTCAATGGGCTCAAGAGA	(CA)n
chr3_38599800	NA17156	T/-	15	3	GGGAGGGGGCTGTGTGGGCTGGCATTAGCTCCCTCTCCAGCAGCCTC	AATTTTCTCATCCCCAGGCAGATCTGCTGGGAAATCGCAGGGCTCCGTGT	
chr3_38602134	NA17156	GGTAGCTGGGTAGA/T GTGTGTGTGTGTG	13	5	GGTAGATGGATTGATAGAAGGGTAAGTAAATGGATAAATGGATGGGTG	GTAGCTGGGTAGATGAGTGGATGGTGTGTGTGTGGCCCTTGCCAACTTA	(TGGA)n
chr3_38602154	NA17156	GATG/TGTGT	17	10	GGTAAGTAAATGGATAAATGGATGGGTGGGTAGCTGGGTAGATGAGTG	ATGGTGTGTGTGTGGCCCTTGCCAACTTACCACAAGGTTGCCAATGACC	(TGGA)n
chr3_38602154	NA17460	GATG/TGTGT	22	9	GGTAAGTAAATGGATAAATGGATGGGTGGGTAGCTGGGTAGATGAGTG	ATGGTGTGTGTGTGGCCCTTGCCAACTTACCACAAGGTTGCCAATGACC	(TGGA)n
chr3_38602134	NA17773	GGTAGCTGGGTAGA/T GTGTGTGTGTGTG	16	4	GGTAGATGGATTGATAGAAGGGTAAGTAAATGGATAAATGGATGGGTG	GTAGCTGGGTAGATGAGTGGATGGTGTGTGTGTGGCCCTTGCCAACTTA	(TGGA)n

chr3_38602154	NA17773	GATG/TGTGT	18	11	GGTAAGTAAATGGATAATTGGATGGGTGGGTAGCTGGGTAGATGAGTG	ATGGTGTGTGTGTGGCCCTTGGCCAACCTACCACAAGGTTGCCAATGACC	(TGGA)n
chr3_38602661	NA17460	-/C	25	12	AGCTCCTGCCTGAAAGCCCAAGGAGCCCTTGGAGGGCACTGACACCTA	TCCCCCCCGCCATTTCTCCAGAGCACCCACTACTGTTCTCTCCACAG	
chr3_38624928	NA17275	T/-	28	18	TGCCTCTGGCCTTCTTCTGTTCTCTGAAGACATCCTGCTGCCAGTG	CCCCATGCTCTCAGAAGACACGGGAAGCCCTGGGCAGGGCCTCAGCTCTC	
chr3_38624928	NA17773	T/-	33	16	TGCCTCTGGCCTTCTTCTGTTCTCTGAAGACATCCTGCTGCCAGTG	CCCCATGCTCTCAGAAGACACGGGAAGCCCTGGGCAGGGCCTCAGCTCTC	
chr3_38638341	NA17460	-/A	60	16	CAGAAGCAGTTTCTGGAACCTGTGCAATGGTTATAGGAGGCAGGACTT	GGAAAAAAGGAGCAGCAGAGTGTGTTACTGAAGTGGGGATAGCCCTGGGG	
chr3_38643195	NA17275	CACCC/GCCG	27	13	CTTGGGCTGCCTAAGTCTTCAGATCCCAGGGAGTTTAAAAAAAAC	ACCCAAAGAAAAGGTAATTAAGTCTGGAGGGTATTTTCATGTGTTGTG	
chr3_38643195	NA17773	CACCC/GCCG	31	13	CTTGGGCTGCCTAAGTCTTCAGATCCCAGGGAGTTTAAAAAAAAC	ACCCAAAGAAAAGGTAATTAAGTCTGGAGGGTATTTTCATGTGTTGTG	
chr7_150277455	NA17773	ACT/-	20	7	ACAGTAGTGAATCAAACCAGCATTTTTTTTTTTTTTTTTTTTTTTTTT	CTGAAAGAACATACAGTAGTATAGCTTAGCACAGCACAGAACAGAAATGC	
chr7_150280333	NA17773	-/CTCTCTTT	30	10	TGCAGCTGCCTTGCCACCATGTCTCTCTCCACTGTCTTCTCTCTTT	TCTCTCTCTCTTTTTCTGTCTCTCTCCACCCCTCCACCCACT	(TC)n
chr7_150286415	NA17275	A/GAC	6	2	AGCGACTCGCTGCTGGGTGCCGCGGGCGTCAGGTCCACGTCCACCACC	CGGCCCCCGGGCGCCCGCGCCCGCCGCGCCCGCCGACCGCACCAGCAGAC	GC_rich
chr7_150290675	NA17156	-/A	12	5	CTTACACACACACAGAGAGACAGAGAGAGAGAGAGAGAGAGAGAGAG	AGTGACTGCTGGCCCTTCTCTTTGTGCGAGATTAAGAAATTTTCTTTT	
chr7_150290653	NA17460	-/AG	13	3	CAGCACAAGCCTGTAACCTCACACTTACACACACACACAGAGAGACAGAG	GAGAGAGAGAGAGAGAGAGAGAGAGAGGAGTACTGCTGGCCCTTCTCTTTGTGCG	(GA)n
chr7_150298286	NA17156	TC/-	5	2	CCACCAGGAAGCCACTCTGGGGAGAAAAGAGGCCAGTCTCCCCC	CACACACCCCAAGCTGGCCCTTGTGTCGCCCATGGCCCCCTCCCCAG	
chr7_150298336	NA17275	-/C	5	2	ACACACCCCAAGCTGGCCCTTCTGCTGCCCATGGCCCCCTCCCCA	AGCCCAAGGCCACCATCAAAGTCTGCTCCCATCTCAGACAGCTCTCGCA	
chr7_150298336	NA17460	-/C	3	2	ACACACCCCAAGCTGGCCCTTCTGCTGCCCATGGCCCCCTCCCCA	AGCCCAAGGCCACCATCAAAGTCTGCTCCCATCTCAGACAGCTCTCGCA	
chr7_150299654	NA17275	-/A	26	14	GGGTGGGGGACAATGGGAGAGAGGGAGAGAGGCAAGGAGCAAGTTCC	GGGTGCTGAGGCGAGGGAAGAAGACAGGAGGAGGACCAGAGAGGAGATA	

Supplemental Table 15. Primer pairs used for long-range PCR amplification of the targeted interval (Perlegen Sciences mapped to NCBI build 36) and PCR amplification around discordant calls for validation by independent sequencing.

Application		Sequence	NCBI36 coordinates	Amplicon size (bp)
LR-PCR	LR-1527027_1	GAGGGGACAGAAGACCAGGCTGGCTAATG	chr7:150284619-150284648	3087
	LR-1527027_2	GGGCTGTCATCATGTTTCATCCTCAATTTGAG	chr7:150287673-150287705	
	LR-1977_1	GTAGTACCTTAGTACCCTTGCAGCCCGTTTC	chr21:34657259-34657290	14227
	LR-1977_2	CAAGATAGCAGGGCAGGGAATAAAGATCAGTA	chr21:34671453-34671485	
	LR-1983_1	TGTGTCTGAGGATTGTCTGGCGTGAGG	chr21:34735911-34735938	10796
	LR-1983_2	GCTCATTGGCATCTGTTTCATTGGAGTAAGCAG	chr21:34746674-34746706	
	LR-1985_1	GGGACCCACATTGCTCTTAGAACTATTC	chr21:34760612-34760640	14476
	LR-1985_2	CTTGTAGCTGAACCCAACGTATCACAACTTA	chr21:34775055-34775087	
	LR-1986_1	TGATACAGTTGGGTTGAGCTACAAGGGTAAC	chr21:34775063-34775094	10469
	LR-1986_2	CTTCTCTTTGCTAGTTGAACCCGCTACTACAT	chr21:34785499-34785531	
	LR-1987_1	CAACTAGCAAAGAGAAGACAGCGTCCTT	chr21:34785515-34785543	9922
	LR-1987_2	TATAGTGGATAACCTTGACTTACCCGGTCCTA	chr21:34795404-34795436	
	LR-1988_1	CAGAGAGTAGGACCGGGTAAGTCAAGGTTATC	chr21:34795398-34795430	4624
	LR-1988_2	CCACGCCAGGTCTTCTTTAGGATCTAGTCT	chr21:34799991-34800021	
	LR-1989_1	ATTACTGTAGCTCAAACGAGGCCAGACTAGAT	chr21:34799969-34800001	6410
	LR-1989_2	CGCATTTGACACTTGACTGGGATACACTA	chr21:34806349-34806378	
	LR-1990_1	CGGTAGTGTATCCAGTCAAGTGTCAAA	chr21:34806347-34806375	12103
	LR-1990_2	CCGTAAGTAGTGTCTATGTATTGGCAACTGTT	chr21:34818417-34818449	
	LR-240580_1	GGGCCAGGCTAGGTTTGTATTGCGATGTCAG	chr11:73836950-73836982	9001
	LR-240580_2	GATCTAACACGAGAGGGCTGGGACGGTGGA	chr11:73845920-73845950	
	LR-240581_1	CCCAGCCCTCTCGTGTAGATCATAGACA	chr11:73845929-73845958	8458
	LR-240581_2	CACAGAGCCAAGCATAACAGCCAAGATTAGAAT	chr11:73854354-73854386	
	LR-240582_1	CTTTCATGATTCTAATCTTGGCTGTATGCTTG	chr11:73854347-73854379	8219
	LR-240582_2	CATCCTCCACCCACATTCCTTAGTTCT	chr11:73862538-73862565	
	LR-382423_1	ATAGAGATTCTCATAGTCTTGGGCTACACTG	chr2:223616214-223616246	11004
	LR-382423_2	CCTTCTAGGTCTCAGCTACGTGACGAAAT	chr2:223627188-223627217	
	LR-54275_1	CCAAGGATGAGGGAGTCTGCGTCACAAG	chr7:150269610-150269638	11143
	LR-54275_2	TGGTCATCTACACGGCTGTCTTACACCCTAC	chr7:150280720-150280752	
	LR-54276_1	GCAGCCGAGTAGGGTGTGAAGACAGCCGTGTA	chr7:150280713-150280745	4905
	LR-54276_2	CTGTGCAGGGCCTGGTCTCCACTCTCGATCTA	chr7:150285585-150285617	
	LR-54277_1	TCTAGAATCCAGTCATCATAAACCGCCATA	chr7:150286689-150286719	11487
	LR-54277_2	AAACTCCTCCCTTAGAGAACTTCTGCGTTTAG	chr7:150298143-150298175	
	LR-54278_1	GAGTTTCAGGAACGCTTCAGGGAAATACCAAC	chr7:150298170-150298202	3423
	LR-54278_2	GTTTCGCTCACAGCAAATCAGGGTTCTAACTG	chr7:150301561-150301592	
	LR-721496_1	CTGGACGTGCATGTAAATCTTGATTGGTTA	chr3:38553978-38554010	11101
	LR-721496_2	ACCCATGCCCTGACAGATCTAGTTCT	chr3:38565051-38565078	
	LR-721498_1	TGACATTCTGAGGGACAATTGCAGGAATAGAC	chr3:38575485-38575517	8720
	LR-721498_2	GAGCCCTAGTGACGTGTTTCAGTTGCCTAGA	chr3:38584174-38584204	
	LR-721499_1	CACCTCCTCATTTCTGCCTCAACTCAA	chr3:38584039-38584066	11568
	LR-721499_2	CAAACCTGATGCAGGACTAACCCACTACCTC	chr3:38595574-38595606	
	LR-721500_1	CTCAGGGGTTGCCAACTCCAATGCTTAC	chr3:38595323-38595351	11083
	LR-721500_2	TTCCTGAGTCTAGCTCCACCTGATAACTGCC	chr3:38606373-38606405	
LR-721501_1	CAGCTGTTTTGGTGGTGAGATGGCAGTTATC	chr3:38606353-38606384	11969	
LR-721501_2	CCAGATTGGGAGGTAGGGGCTGAAGACTTACT	chr3:38618289-38618321		
LR-721502_1	CCCAACCCTGTGCTTTGTTTTAGACTAGAAC	chr3:38618395-38618427	11967	
LR-721502_2	CATTGTCTCGTGTGTGAATTCCTTGTTACA	chr3:38630329-38630361		

	LR-721503_1	TGTAACAAGGGAAATTCACACACGAGACA	chr3:38630330-38630359	11821
	LR-721503_2	TAACAAATAATTCCCTCCCTCCTGTACGGTAG	chr3:38642118-38642150	
	LR-721504_1	ACCGTCGAATGTGGGGAGACTACCGTA	chr3:38642100-38642127	11891
	LR-721504_2	TCAGGGCACACAGAATTTTGGAGAATGTCTAA	chr3:38653958-38653990	
	LR-721505_1	ATTGTAAGACCACAGTCGCTCATCCCCTAGAT	chr3:38653478-38653510	11811
	LR-721505_2	GGCTCTCGACCTCTGTGTTATTGATCGTC	chr3:38665258-38665288	
	LR-778623_1	ATGGATACAGGCCCTAAATTTGAGAGTCTAT	chr21:34746238-34746269	3341
	LR-778623_2	AGGATGATTTCTGATGGCATATTTAGGTAAGA	chr21:34749546-34749578	
	LR-778624_1	CAGCTGAGGAATAGGCCACTTTTATCAGATAG	chr21:34749486-34749518	11646
	LR-778624_2	ACTTATTCTCCTGCTTACCTTCCTAGTCCAA	chr21:34761099-34761131	
False positive Validation	FP1-F	CAGTGTTCTACCCCCCTCTG	chr21:34759544+34759840	297
	FP1-R	CTGGTTCTCTGGCTGGGTAA		
	FP2-F	GCTCACTCCTCTTCTCCAA	chr21:34807745+34808040	296
	FP2-R	GAATCGTTGCCAGCTACTCC		
	FP3-F	TCAATGGGCTCAAGAGAAGG	chr3:38596428+38596733	306
	FP3-R	GTTGAGTCAGGCCATCTTG		
	FP4-R	GAGGATGGTGCACATGATGA	chr3:38638951+38639257	307
	FP4-F	CCCTACCAGCAGTGTGGTTT		
	FP5-F	GGCTGTGCAGTGTCAAGTTA	chr7:150292849+150293143	287
	FP5-R	GGAGGCAGGAGCTTAGGACT		
FP67-F	GAGTTAGGGAGCCCAAGAGG	chr7:150293004+150293290	271	
FP67-R	CCCCAAGACCCTATAAATG			
False Negative Validation	FN1-F	AGGCAGAAGACAAAGGCAAG	chr7:150299734+150300033	300
	FN1-R	GCAGAGTCCAGTGGTGTGAG		
	FN2-F	GGGCTAGGAATGGAAGAAGG	chr7:150275218+150275518	301
	FN2-R	TCCAACATTTTCAGCTTCTGG		
	FN3-F	GGAACAAAACCTGCCATA	chr2:223624363+223624672	310
	FN3-R	TTAACCTCTCCGCACCTCAC		