

Supplementary table 1 PCR primers for amplification

Patient	Exon	Forward primers	Reverse primers
1	11	GCAGCAGCTGGGTCATCTAT	ACCACACCTGGCCTTCAATA
2	19	CCCAGCTTGCATTAAATAGTCTG	GAAAGTAGAAGAAACATGATTG AACC
3	22	AGGCTATTTTCGTGCC	CACCTGGCCTTCAAT
4	24	TATGGGCAATGGCAGAATATG	TATGCCTGGATGAGGTGTTTG
5	17	AAATTGGAAGGCTATTCCTATGAG	AGGTAGATGTTAAGAAACACCTC TCAC
6	18	TGCCACTGTCAATTGTGCC	GCAAATCCTAGGTGATTCAGTAGC
7	2	TGTTATGTGCAAACCTATTGAAACAAG	AGCTCAGGCAATCCACCTG
8	11	GCAGCAGCTGGGTCATCTAT	ACCACACCTGGCCTTCAATA
9	6	CAGCTTCTCATGGTCAAGAATG	AAGCAGAGAATGAGGGAGGAG

Supplementary table 2 Coverage level through the target region for patients

Patient	Reads (M)	Read length (bp)	Reads aligned to the human genome (Mb)	Reads	Mean coverage	SNPs	Indels,		
				mapped to the target region (Mb)		Non-synonymous	Splice sites	Coding sequence	Splice sites
1	45.32	149	13535.97	13531.65	99.9%	9662	835	751	255
2	39.43	149	11726.17	11715.66	99.8%	12900	1084	972	313
3	56.12	148	16653.27	16631.34	99.8%	9549	753	728	244
4	51.21	149	15252.03	15243.88	99.9%	12766	1006	992	320
5	53.46	148	15863.68	15852.41	99.9%	12681	983	970	314
6	36.7	148	10895.29	10888.65	99.8%	12889	981	976	312
7	43.34	149	12876.37	12866.38	99.8%	12736	1047	945	322
8	49.3	149	14654.79	14648.19	99.9%	12870	971	939	287
9	32.8	149	9720.58	9708.46	99.8%	12708	1024	935	308