

Primers sequences for TP53 SNPs

From Garritano et al., *Human mutation*, 2009

Detailed haplotype analysis at the TP53 locus in p.R337H mutation carriers in the population of Southern Brazil: evidence for a founder effect.

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Due to patterns of migration, selection, and population expansion, founder effects are common among humans. In Southern Brazil, a recurrent TP53 mutation, p.R337H is detected in families with cancer predisposition. We have used whole locus resequencing and high density SNP genotyping to refine TP53 locus haplotype definitions. Haplotyping of 12 unrelated p.R337H carriers using a set of 29 tag SNPs, revealed that all subjects carried the same haplotype and presence of the mutation on this haplotype was confirmed by allele-specific PCR. The probability that this haplotype occurs independently in all index cases was of 3.1×10^{-9} , demonstrating a founder effect. Analysis of the patterns of 103 tumors diagnosed in 12 families showed that the presence of p.R337H is associated with multiple cancers of the Li-Fraumeni Syndrome (LFS) spectrum, with relatively low penetrance before the age of 30 but a lifetime risk comparable to classical LFS. The p.R337H families are mostly distributed along a road axis historically known as the main route used by merchants of Portuguese origin in the XVIII and XIX century. This historical circumstance and the relatively low penetrance before the age of 30 may have contributed to the maintenance of this pathogenic mutation in a large, open population. (c) 2009 Wiley-Liss, Inc.

SNPs	Forward sequence 5' > 3'	Reverse sequence 5' > 3'	Amplicon size (bp)
rs2287499	ACTCTGTTTCCAGGGGAGTG	GGTTGTCCCCAGATCCTGT	93
rs17551157	CTACGCTCCCCCTACCGAGT	AAGAGGTGCAAGACCTGCTG	94
rs17883353	GGGCGACAAGAACGAAAC	ACAACATGAACGAATGTCAGA	113
rs17882227	TGGCCTTTTGAGTTGTTTCC	CGCAATTCCCCTCCTAAGTA	125
rs17885845	GGAATTGCGAGTTTGGAAAGT	CTAATGTCCGGAAGGCTGTG	146
rs9903378	TTCAGTATTTGCCTTGTCTGTT	CCATAAAGGTCCTAAAAGAAAATG	100
rs17881035	TTAACCCCAGGGTCATGAAG	TGACTCCTACACCTCACACCA	135
rs11656607	GCAGGGGATCATTGAGG	TGGCTTCGAAAACAAGTTGG	290
rs5819163	ACATTACCCCATACAATGA	GTGGCTGCTGGTATCAGTCTT	120
rs2078486	AGTGGGGGTGGGAGCAGTA	TGCAATTGTTCTATTTCACTTGTTCC	94
rs1642782	AAGATGCTGGTCCACACAGG	ACCTGCTTGGGCTCAATAAA	104
rs12944939	GTTAACAGGAGGTGGGAGCA	AGAGACAGGGCTTTGCATGT	132
rs8079544	CAGCCATTCTTTTCTGCTC	CTTCCAACCCTGGGTAC	108
rs1642785	CCCCTCTGAGTCAGGAAACA	TCCCACAGGTCTCTGCTAGG	114

rs17878362, rs17883323	TTCCTGAAAACAACGTTCTGG	GGGACTGTAGATGGGTGAA	134
rs1042522	GAAGACCCAGGTCCAGATGA	TGGTAGGTTTTCTGGGAAGG	146
rs1794287	TCCCAGCACTCTCAAAGAGG	CTCCATCTCCTGGCCTCA	194
rs1625895	GGGTTAAGGGTGGTTGTCAG	TTGCACATCTCATGGGGTTA	97
rs12947788	CTGGAAGACTCCAGGTCAGG	TGATGAGAGGTGGATGGGTAG	135
rs12949655	CCAGGCTGGTCTCTAGCCTAC	TAGCAGGCGCTTGTAGTCCT	357
rs858528, rs1641548, rs1641549	CAGTCGAGCCAGGCACAG	TGTGCCTCGTTTCTTTTCTTT	359
rs6503048	TAATCCCAGCTACCTACTCG	CAATGCTTTGAAGGGCCTAA	282
rs17880560, rs1614984	GCTCCATTCTAACTCAGGA	CCGTAATCCTTGGTGAGAGG	130
rs9894946, rs17883532	TCAAAGGGCTTCTTGGGATA	AAGAGAGCAGTGGGTGATGG	138
rs17886760	ATCACCCACTGCTCTCTTGG	CACCCACTGGACCCTAACAC	104
