

MUTATIONAL ANALYSIS_TH GENE

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tccaggagcc tccgtggcct gtgccccac cccagcacag cctcctgacc 2189186
cgtgcatccc ctctgcctc agGGCTTCTC GGACCAGGTG TACCGCCAGC 2189136 EX6

GCAGGAAGCT GATTGCTGAG ATCGCCTTCC AGTACAGGCA gtgagggggc 2189086
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gggtgggagq gaaggacaaa ggggaggacc catcttgtca ccagcatcag 2188986
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c.698G>A (p.R233H)
 (p.Arg233His)

ggggccgggg atggtcagcc aagcgcceca cccagcgcg gctccagccc 2188036
gtcccggctc ggcagtgacc cgcgtggccc cttgcaagAGC GCACGGGCTT 2187986 EX9

CCAGCTGCGG CCTGTGGCCG GCCTGCTGTC CGCC**C**GGGAC TTCCTGGCCA 2187936
GCCTGGCCTT CCGCGTGTTT CAGTGCACCC AGTATATCCG CCACGCGTCC 2187886
TCGCCCATGC ACTCCCCTGA GCCgtgagtq cgcgccttgq cegccagccc 2187836
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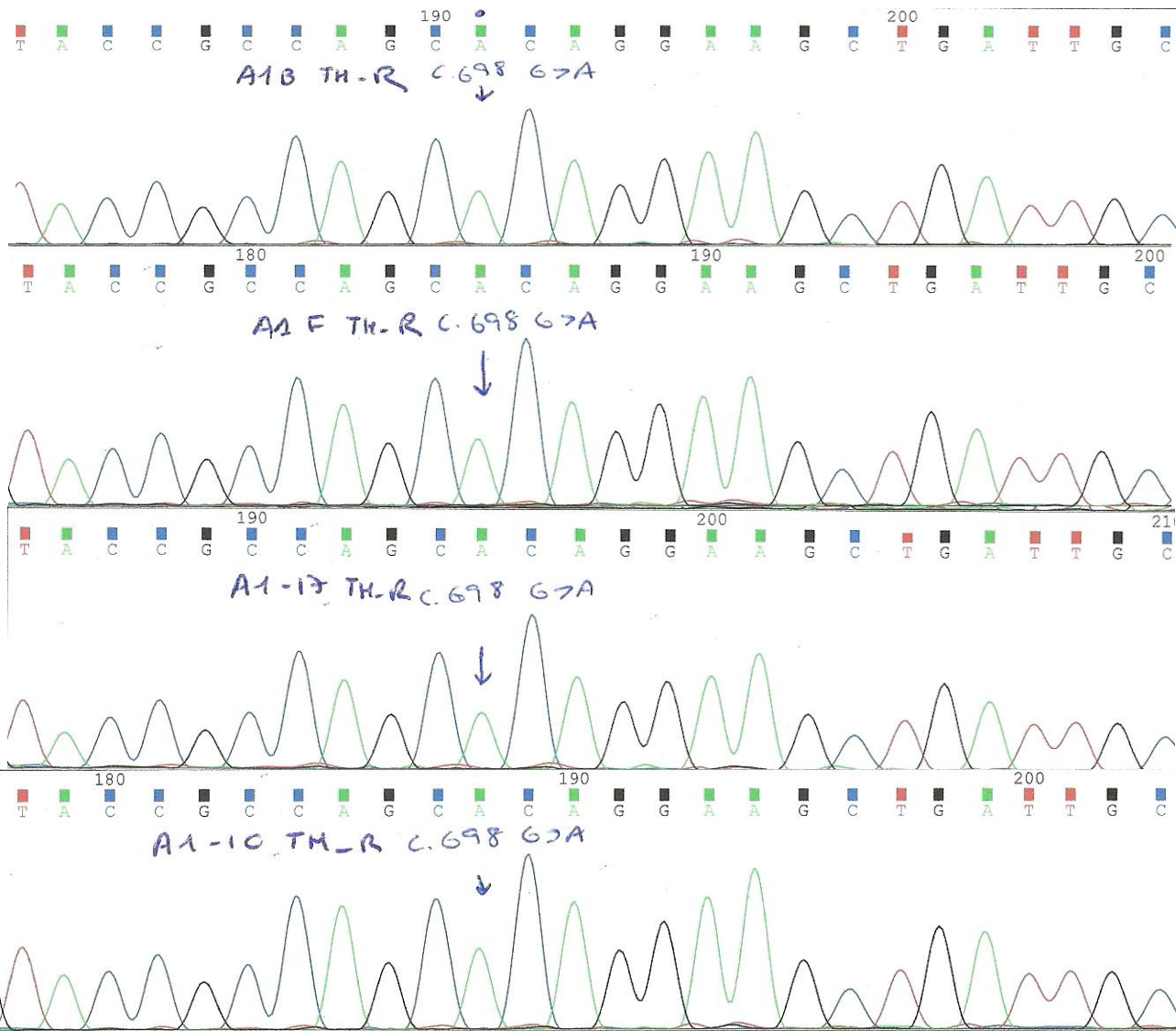
c.982C>T (p.R328w)
 (p.Arg328Trp)

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CA**C**Ggtgggt tgaccctcc ctgcagggcc tgggggtg gtttgggggt 2187186
ctgaatccag gcctcacct cttgccgtcc aggctgaggc ctctcctcc 2187136

c.1196C>T (p.T399M)
 (p.Thr399Met)

c.698G>A



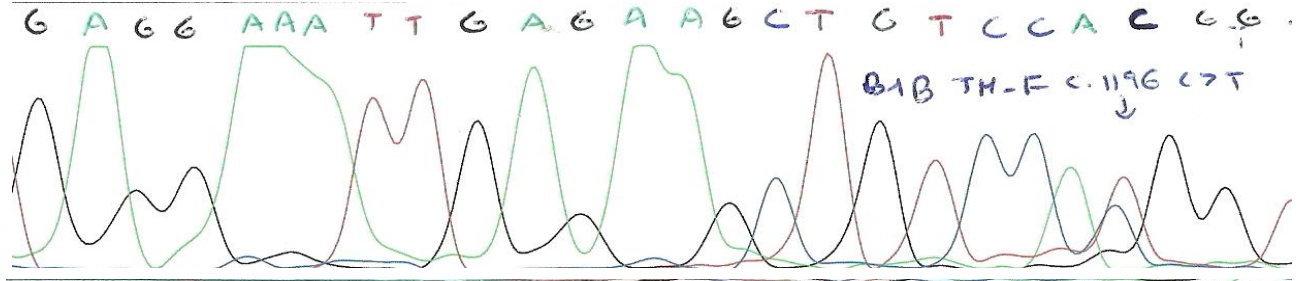
alignment position

SeqReFc.698G_A	145	185	179	182	189						
A5 Blood	GCCAGCGCAG	GAAGCTGATT	GCTGAGATCG	CCTTCCAGTA	CAGGCAGTGA	GGGGCCCCCTG	CGCTCGGGAC	CCAGACTCCG	TCCTGCAGGC	TGACGCTGGA	CCTGGGGGGT
A6 Fibroblasts	GCCAGCACAG	GAAGCTGATT	GCTGAGATCG	CCTTCCAGTA	CAGGCAGTGA	GGGGCCCCCTG	CGCTCGGGAC	CCAGACTCCG	TCCTGCAGGC	TGACGCTGGA	CCTGGGGGGT
A7 Clon 10	GCCAGCACAG	GAAGCTGATT	GCTGAGATCG	CCTTCCAGTA	CAGGCAGTGA	GGGGCCCCCTG	CGCTCGGGAC	CCAGACTCCG	TCCTGCAGGC	TGACGCTGGA	CCTGGGGGGT
A8 Clon 17	GCCAGCACAG	GAAGCTGATT	GCTGAGATCG	CCTTCCAGTA	CAGGCAGTGA	GGGGCCCCCTG	CGCTCGGGAC	CCAGACTCCG	TCCTGCAGGC	TGACGCTGGA	CCTGGGGGGT

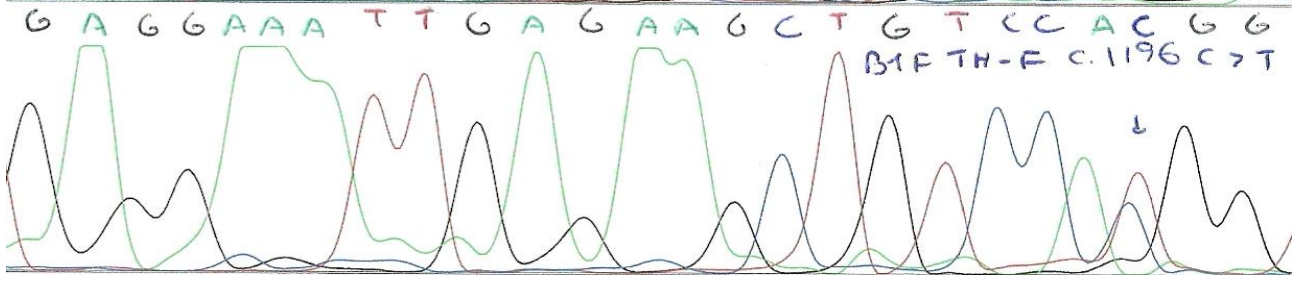


c.1196C>T

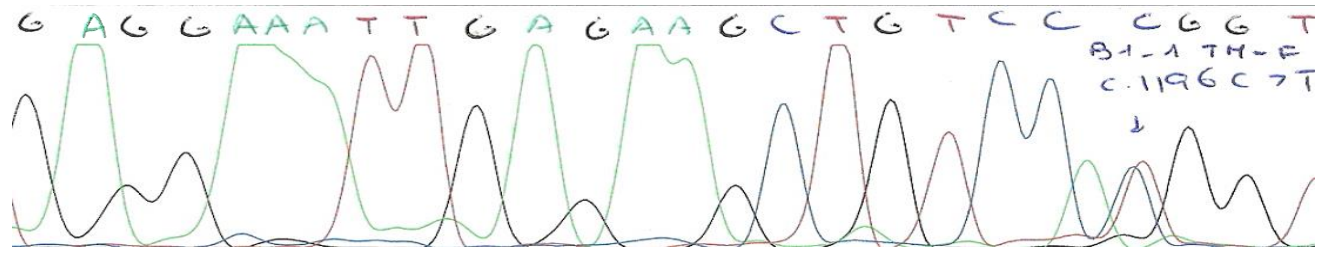
Blood



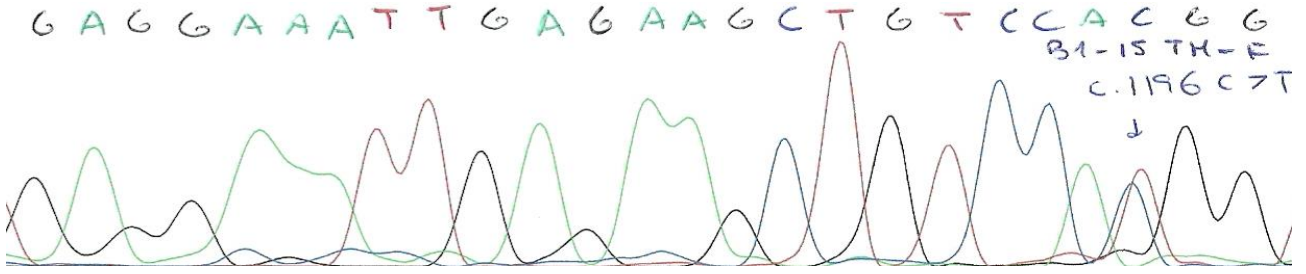
Fibroblasts



Clon 1



Clon 15



alignment position	1.....	11.....	21.....	31.....	41.....	51.....	61.....	71.....	81.....	91.....	10.....		
SeqReFc.1196C_T	1	tgacc	ccaggg	ctgcctctcc	ccatccttca	tcctcctctgc	tccacaggac	attggcCTGG	CGTCCCTGGG	GGCCTCGGAT	GAGGAAATTG	AGAAGCTGTC	CACGGTGGGT
A17 Blood	1	-----	-----	-----	-----	-----	-----	-----CTGG	CGTCCCTGGG	GGCCTCGGAT	GAGGAAATTG	AGAAGCTGTC	CATGGTGGGT
A18 Fibroblasts	1	-----	-----	-----	-----	-----	-----	-----	CGTCCCTGGG	GGCCTCGGAT	GAGGAAATTG	AGAAGCTGTC	CATGGTGGGT
A19 Clon 1	1	-----	-----	-----	-----	-----	-----	-----	-GTCCTGGG	GGCCTCGGAT	GAGGAAATTG	AGAAGCTGTC	CATGGTGGGT
A20 Clon 15	1	-----	-----	-----	-----	-----	-----	-----	-----CCTCGGAT	GAGGAAATTG	AGAAGCTGTC	CATGGTGGGT	
									****	*****	*****	*****	*****

