

Gen	Family code	ID Proband	Gender (Male, Female)	Cancer site and age at diagnosis	Mutation designation	Exon/ Intron	Mutation type	Mutation status	Loss of protein expression	MSI status	Number of carrier	Number of Noncarrier
MSH2	POINT MUTATIONS											
	VA117	194	F	Uterine-Colon 47-53	c.229_230delAG; p.Ser77CysfsX4	2	Frameshift	MSH2_00100			5	3
	VA22	438	M	Colon-Rectum 43-63	c.229_230delAG; p.Ser77CysfsX4	2	Frameshift	MSH2_00100	MSH2/MSH6-	MSI-H	7	6
	VA188	326	F	Colon carcinoma , 68	c.1226_1227delAG p.Gln409ArgfsX7	7		MSH2_00323	MSH2/MSH6-	MSI-H	4	2
	VA251	432	F	Colon /Endometrium 23-35	c.1216C>T; p.Arg406X	7	Nonsense	MSH2_00312			1	
	VA6	75	M	Colon-Rectum Rectum-Sigma 47	C.1661G>A; p.Ser554Thr	10	Splicing	NEW MSH2_00999	MSH2/MSH6-	MSI-H	18	26
	VA174	305	M	Colon carcinoma , 41	c.2240_2241delTA p.Ile747ArgfsX2	14	Frameshift	MSH2_01137	MSH2/MSH6-	MSI-H	2	
	VA199	368	M	Urinary tract/ Colon carcinoma , 50-52	c.2470C>T; p.Gln824X	15	Nonsense	MSH2_01163	MSH2/MSH6-	MSI-H	3	2
	VA191	430	F	Endometrium, 40	c.2634G>A skipping exon15	15	Splicing	MSH2_00823			2	
	LARGE REARRANGEMENTS											
	VA17	86	F	Endometrium 60	c.646-1019_1386+2420del p.Ile216_Gln462del	4-8	Exons 4-8 deletion	NEW	MSH2-	MSI-H	6	6
	VA20	29	M	Colon carcinoma, 31	c.646-1019_1386+2420del p.Ile216_Gln462del	4-8	Exons 4-8 deletion	NEW			8	2
	VA32	52	F	Endometrium 51	c.646-1019_1386+2420del p.Ile216_Gln462del	4-8	Exons 4-8 deletion	NEW	MSH2/MSH6-	MSI-H	5	3
	VA134	246	F	Endometrium	c.646-1019_	4-8	Exons 4-8	NEW			3	

				42	1386+2420del p.Ile216_Gln462del		deletion						
	VA4	4	F	Colon carcinoma, 39	c.1077-3513_1276+5655 p.R359RfsX16	7	Exon 7 deletion	NEW			8	6	
	VA169	299	M	Colon carcinoma 29-37	c.1077-3513_1276+5655 p.R359RfsX16	7	Exon 7 deletion	NEW	MSH2/MSH6-	MSI-H	1	2	
	VA247	413	M	Colon/ Prostate/Urinary 47-57-60	c.1077-3513_1276+5655 p.R359RfsX16	7	Exon 7 deletion	NEW			4	4	
EPCAM-MSH2	LARGE REARRANGEMENTS IN EPCAM/MSH2												
	VA25	43	M	Colon/ Bladder 33-34	c. 859-1860_MSH2:646-254del	EPCAM exon 7-8 and MSH2 exon 1-3	EPCAM exon 7-8 and MSH2 exon 1-3 deletion	NEW	MSH2/MSH6-	MSI-H	2	0	