Supplementary Table 1 **hMLH1/hMSH2 Gene Variants**

NB: All the gene variants listed below have been published in the ICG-HNPCC database, or in other relevant papers. It should be noted, however, that the various sources differ considerably in the way alterations are presented, and the amount of detail provided. The authors of this review have interpreted the available information for presentation in the format shown, and in some cases have inferred details not specifically given in the original publication. In several instances, variants have been published which are inconsistent with the consensus sequence of hMLH1/hMSH2, and could not easily be re-interpreted. In such cases, variants have been entered as given in the publication, with possible alternative interpretations given afterwards in Italics.

Spaces within the table indicate that relevant information was not available.

The 'cancer' column lists the types of cancer found in the affected kindred(s). The extent to which such information is provided varies considerably, and in many cases very little clinical information is provided. Information in this column should thus be interpreted with particular care.

In the ID column, identification codes separated by commas refer to different families with identical alterations, whereas identification codes separated by a forward slash refer to the same kindred, which has been included in more than one publication under different IDs.

Classification of mutations, as given in the mutation/polmorphism (M/P) column, has been performed by the authors of the review for the purpose of summarising the information contained in this table, and is based on the predicted consequences of each variant, incorporating the results of functional assays where available. These classifications do not necessarily reflect the interpretation given in the original publications.

Although the functional domains of the bacterial homologues of hMLH1 and hMSH2 have been studied, information specific to the human genes is scarce. This issue has been addressed in several recent papers [Schmutte et al., 2001;Tomer et al., 2002;Kondo et al., 2001], but details remain to be be confirmed and discussion of the current evidence is outwith the scope of this review.

(a) Germline hMLH1 Alterations

ID	EXON	Nucleotide Change	cDNA change	Cancer	Origin	Frequency	Reference	ICG	M/P
Healthy controls	5'UTR	g-a nt 1-93				0.5	[Ito et al., 1999]	Y	4
13	5'UTR	c-t nt 1-42			Germany		[MullerKoch et al., 2001]	N	4
14	5'UTR	a-g nt 1-28			Germany		[MullerKoch et al., 2001]	N	4
Case 8	5'UTR	c-t, nt 1 -21			USA		[Samowitz et al., 2001]	N	4

1	5'UTR	t-c, nt 1-21			Germany		[HolinskiFeder et al., 2001]	N	4
2	5'UTR	a-g, nt 1 –21 (17?)			Germany		[HolinskiFeder et al., 2001]	N	4
	5'UTR	g-a nt 1-17			Switzerland		[Heinimann et al., 1999]	N	4
Family 2	Ex 1	G-A nt 1	loss of translation start site	co	Australia		[Scott et al., 2001]	N	2
72	Ex 1	T-A nt 2	Met1Lys	co	Germany		[Wehner et al., 1997]	Y	2
3	Ex 1	T-A nt 2	Met1Lys	co	Germany		[HolinskiFeder et al., 2001]	N	2
FF10, F34	Ex 1	T-G nt 2	Met1Arg	co/ut	French		[Wang et al., 1997]	N	2
F2	Ex 1	C-A nt 5	Ser2STOP		Italy		Piepoli, unpub.	Y	1
NL-205, NLB-1069	Ex 1	17bp del nt 17	frameshift from codon 5	co/HNPCC	Netherlands		[Wijnen et al., 1997]	Y	1
	Ex 1	G-A nt 39	Glu13Glu		Scotland	0.014	[Farrington et al., 1998]	N	4
2841/ 4F	Ex 1	del G nt 63	frameshift from codon 20	co	USA		[Moslein et al., 1996]	Y	1
39603	Ex 1	G-T nt 69 (*67*)	STOP		UK		Norbury, unpub.	Y	1
110	Ex 1	del A nt 73	frameshift from codon 24	co	Germany		[Wehner et al., 1997]	Y	1
4	Ex 1	del A nt 73	frameshift from codon 24	co	Germany		[HolinskiFeder et al., 2001]	N	1
Patient 377	Ex 1	del A nt 73	frameshift from codon 24		Czech Republic		Krepelova, unpub.	Y	1
166	Ex 1	A-T nt 73	Ile25Phe	co/re	USA African		[Weber et al., 1997]	Y	2/3
40547	Ex 1	T-C nt 74	Ile25Thr		UK		Norbury, unpub.	Y	2/3
	Ex 1	G-T codon 26 (*C-T nt 76*)	Glu26STOP (*Gln26STOP*)	co	Portugal		[Cravo et al., 1999]	N	1
	Ex 1	del A nt 84	frameshift – STOP codon 35		Asia		Deffenbaugh, unpub	Y	1
52	Ex 1	C-T nt 83	Pro28Leu		Germany		[Wehner et al., 1997]	Y	2/3

5	Ex 1	C-T nt 83	Pro28Leu		Germany	[HolinskiFeder et al., 2001]	N	2/3
NLB-100	Ex 1	del AG nt 101	frameshift from codon 33	co/HNPCC	Netherlands	[Wijnen et al., 1997]	Y	1
3	Ex 1	T-G nt 104	Met35Arg	co/en/br	Sweden	[Tannergard et al., 1995]	Y	2
h9	Ex 1	ins AA nt 105	frameshift from codon 34		England	Bunyan, unpub.	Y	1
30	Ex 1	ins AA nt 105	frameshift from codon 34		England	[Coleman et al., 2001]	N	1
477	Ex 1	T-G nt 107	Ile36Ser	co/ut	Finland	[Aaltonen et al., 1998]	N	2
Family 3	Ex 1	A-G nt 112	Asn38Asp	co	Australia	[Scott et al., 2001]	N	2
MD815	IVS 1	g-a nt 116+1	splice defect	co	Scotland	[Farrington et al., 1998]	N	1
Family 1	IVS 1	g-a nt 116+1	splice defect	co	Australia	[Scott et al., 2001]	N	1
MD985	IVS 1	g-t nt 116+1	splice defect	co	Scotland	Farrington/Dunlop, unpub.	Y	1
2	Ex 2	C-T nt 131	Ser44Phe	co/en/br/ga	Sweden	[Bronner et al., 1994]	Y	2
15	Ex 2	C-T nt 131	Ser44Phe	co	Sweden	[Tannergard et al., 1995]	Y	2
6	Ex 2	ins G nt 151	frameshift from codon 51		Germany	[HolinskiFeder et al., 2001]	N	1
Case 1 – BR	Ex 2	C-T nt184	Gln62STOP	co/en	USA	[Liu et al., 1995a]	Y	1
91	Ex 2	C-T nt184	Gln62STOP		Germany	[Wehner et al., 1997]	Y	1
127	Ex 2	C-T nt184	Gln62STOP	co	Germany	[Lamberti et al., 1999]	Y	1
234	Ex 2	C-T nt184	Gln62STOP	co	Germany	[Lamberti et al., 1999]	N	1
7	Ex 2	C-T nt184	Gln62STOP	co	Germany	[HolinskiFeder et al., 2001]	N	1

N498	Ex 2	C-A nt184	Gln62Lys	co/HNPCC	Norway	0/100	[Wijnen et al., 1997]	Y	2
8	Ex 2	del AA nt 190	frameshift from codon 64		Germany		[HolinskiFeder et al., 2001]	N	1
N2104	Ex 2	A-G nt 191	Asn64Ser	co/HNPCC	Norway	0/100	[Wijnen et al., 1997]	Y	2/3
MD1446	Ex 2	A-G nt 191	Asn64Ser	co	Scotland		Farrington, unpub.	N	2
	Ex 2	ins C nt 198	frameshift – STOP codon 78		Asia		Deffenbaugh, unpub.	Y	1
7	Ex 2	C-T nt 198	Thr66Thr		Germany	0.01	[Wehner et al., 1997]	Y	4
Case 9	Ex 2	C-T nt 198	Thr66Thr		USA		[Samowitz et al., 2001]	N	4
MFI	Ex 2	G-A nt 199	Gly67Arg		USA		[Herfarth et al., 1997]	Y	2
JPN-28	Ex 2	G-A nt 199	Gly67Arg	co/HNPCC	Japan		[Sasaki et al., 1997]	Y	2
VS015	Ex 2	G-A nt 199	Gly67Arg		Switzerland		[Hutter et al., 1998]	Y	2
1652	Ex 2	G-A nt 199	Gly67Arg		Switzerland		[Heinimann et al., 1999]	Y	2
OG/98-2989	Ex 2	G-A nt 199	Gly67Arg	co	Slovenia		[RavnikGlavac et al., 2000]	N	2
69	Ex 2	G-C nt 199	Gly67Arg	co/en	Sweden		[Tannergard et al., 1995]	Y	2
Patient 304	Ex 2	G-C nt 199	Gly67Arg		Czech Republic		Krepelova, unpub.	Y	2
MD1759	Ex 2	G-C nt 199	Gly67Arg	co	Scotland		Farrington/Dunlop, unpub.	N	2
FF52	Ex 2	GGG-TGG	Gly67Trp	co/b/ly/leu/Rh	France		[Wang et al., 1999]	N	2
18	Ex 2	T-A nt 203	Ile68Asn	co/en/pa/ga	Sweden		[Tannergard et al., 1995]	Y	2
10	IVS 2	A-G nt 208-2	in frame del Ex 3 codon 70102	co/ga	Sweden		[Tannergard et al., 1995]	Y	1
Family 4	IVS 2	del 4bp, +1,+2,208,209	loss of intron/exon junction	co	Australia		[Scott et al., 2001]	N	1
	Ex 3	T-C or A nt 228	Cys77Arg	co/HNPCC	Italy		[Panariello et al., 1998]	N	2

YKY	Ex 3	G-A nt 229	Cys77Tyr	co	England		[Tomlinson et al., 1997]	N	2
IMS2	Ex 3	G-A nt 229	Cys77Tyr		Australia		Bennet, unpub.	Y	2
9	Ex 3	T-G nt 238	Phe80Val		Germany		[HolinskiFeder et al., 2001]	N	3/4
1	Ex 3	T-G nt 238	Phe80Val		Germany		[MullerKoch et al., 2001]	N	3/4
HK	Ex 3	1 bp del	frameshift codon 82	co/HNPCC	England		[Beck et al., 1997a]	N	1
DF5	Ex 3	C-T nt 245	Thr82Ile	co/HNPCC	USA		[Syngal et al., 1999]	N	2/3
10	Ex 3	A-G nt 250	Lys84Glu	co	Germany		[HolinskiFeder et al., 2001]	N	2
171	Ex 3	A-G nt 250	Lys84Glu	co	Germany		[Lamberti et al., 1999]	Y	2
	Ex 3	A-G nt 277	Ser93Gly	co			[Quaresima et al., 1998]	Y	2/3
FF44, FF75	Ex 3	C-T nt 298	Arg100STOP	co/b/ut/urt	France		[Wang et al., 1999]	N	1
Case 1	Ex 3	C-T nt 298	Arg100STOP	co/sk	USA		[Samowitz et al., 2001]	N	1
61	Ex 3	G-A nt 304	Glu102Lys		USA		de la Chapelle, unpub.	Y	2
R-RM2	Ex 3	Ex 3 deleted	deletion Ex 3 codon 70102	co/ut/k	Italy		[Viel et al., 1997]	N	1
115	Ex 35	Ex 3-5 deleted	deletion Ex 35 codon 70151	co/en	Finland		[Aaltonen et al., 1998]	N	1
9	IVS 3	g-a nt 306+1	inframe deletion exon 3	co/ut/lv	Germany/Czech Republic		[Pistorius et al., 2000]	N	1
11	IVS 3	g-a, 306+1	inframe deletion exon 3	co/ut/lv	Germany		[HolinskiFeder et al., 2001]	N	1
H16	IVS 3	g-a nt 306+1	inframe deletion exon 3	co/ut	Iran		[Planck et al., 1999]	N	1
Case 10	IVS 3	c-g nt 306+15		co	USA		[Samowitz et al., 2001]	N	4
	IVS 3	c-a nt 307-29			Sweden	0.0038	[Liu et al., 1998a]	Y	3/4

12	IVS 3	c-a nt 307-29			Germany	[HolinskiFeder et al., 2001]	N	3/4
Case 11	IVS 3	c-a nt 307-29		co	USA	[Samowitz et al., 2001]	N	3/4
28,51,67,477	Ex 4	T-G nt 320	Ile107Arg	co/en/HNPCC	Finland	[NystromLahti et al., 1996]	Y	2
614,661,811	Ex 4	T-G nt 320	Ile107Arg	co	Finland	[Salovaara et al., 2000]	N	2
355	Ex 4	C-T nt 332	Arg111Val	co	Japan	[Nomura et al., 2000]	N	2
	Ex 4	C-G nt 341 (*350*)	Thr117Arg	co/HNPCC	Switzerland	[Heinimann et al., 1999]	N	2
0434*	Ex 4	C-G nt 350	Thr117Arg	co/b/br/ov	Switzerland	[Buerstedde et al., 1995]	Y	2
LG	Ex 4	C-T nt 350	Thr117Met	co	USA	[Liu et al., 1996]	Y	2
5	Ex 4	C-T nt 350	Thr117Met	co/en/sa	Moldavia	[Maliaka et al., 1996]	Y	2
SB-2575	Ex 4	C-T nt 350	Thr117Met	co	Poland	StachowKurzawski, unpub.	Y	2
h83	Ex 4	C-T nt 350	Thr117Met		England	Bunyan, unpub.	Y	2
BES-SG624	Ex 4	C-T nt 350	Thr117Met		France	[Dieumegard et al., 2000]	Y	2
84	Ex 4	C-T nt 350	Thr117Met		USA	de la Chapelle, unpub.	Y	2
26	Ex 4	C-T nt 350	Thr117Met		Germany	[Raedle et al., 1999]	Y	2
Patient 431	Ex 4	C-T nt 350	Thr117Met		Czech Republic	Krepelova, unpub.	Y	2
Family 5	Ex 4	C-T nt 350	Thr117Met	co	Australia	[Scott et al., 2001]	N	2
FF39	Ex 4	C-T nt 350	Thr117Met	co/urt/ga/se	France	[Wang et al., 1999]	N	2
	Ex 4	C-T nt 350	Thr117Met			Deffenbaugh, unpub.	Y	2
Case2	Ex 4	C-T	Missense	co	Poland	[Debniak et al., 2000]	N	2

Case 1	Ex 4	ins AA nt 352	frameshift from codon 118	co	Poland		[Debniak et al., 2000]	N	1
1 kindred	Ex 4	A-G nt 375	Ala125Ala	co	Scotland		[Farrington et al., 1998]	N	4
870	Ex 4	C-G nt 378	Tyr126STOP	co	Finland		[Salovaara et al., 2000]	N	1
FF53	Ex 4	del C	frameshift from codon 126	co/leu	France		[Wang et al., 1999]	N	1
Family 6	Ex 4	G-A nt 381 (38*0 or A-G nt 381, exon 5*)	loss of intron/exon junction	co	Australia		[Scott et al., 2001]	N	1
34	Ex 411	-	in frame del Ex 411	co	Sweden		[Wahlberg et al., 1999]	Y	1
SK1	IVS 4	t-a nt 380+2	splice defect		Slovakia		Bartosova & Fridrichova, unpub.	Y	1
1044	IVS 4	a-g nt 381-2	splice defect	co/en/ov/re	France		[Mauillon et al., 1996]	Y	1
1500/5	IVS 4	a-g nt 381-2	splice defect		Belgium/ Switzerland		[Heinimann et al., 1999]	Y	1
338	Ex 5	G-C nt 382	Ala128Pro		Italy		[Pensotti et al., 1997]	Y	2/3
12*,11*,10*,17*	Ex 5	G-T nt 397	Gly133STOP	co/ut/ov	Japan		[Nakahara et al., 1997]	N	1
MD1092	Ex 5	ins 1 bp	frameshift from codon 143	co	Scotland		Farrington/Dunlop, unpub.	Y	1
02	IVS 5	g-t nt 453+1	splice defect	co	Lithuania		Avizienyte, unpub.	Y	1
	IVS 5	g-t nt 453+1	splice defect		Latin American / Caribbean		Deffenbaugh, unpub.	Y	1
CC8	IVS 5	t-c nt 453+2	splice defect		Spain		Caldes, unpub.	Y	1
Finnish Kindreds	IVS 5	t-c nt 454-51			Finland	0.04	[NystromLahti et al., 1996]	Y	3/4
Patient 478	IVS 5	t-c nt 454-51			Czech Republic		Krepelova , unpub.	Y	3/4
	IVS 5	a-g nt 454-16			Sweden		[Wahlberg et al., 1999]	N	3/4
	IVS 5	a-g nt 454-13					Lindblom, unpub.	Y	3/4

102	IVS 5	g-a nt 454-1	out of frame del Ex 6 codon 152182		Sweden	[Liu et al., 1998a]	Y	1
10 Kindreds	IVS 5	g-a nt 454-1	out of frame del Ex 6 codon 152182	co/HNPCC	Finland	[NystromLahti et al., 1995]	Y	1
4 kindreds	IVS 5	g-a nt 454-1	out of frame del Ex 6 codon 152182	co	Finland	[Salovaara et al., 2000]	N	1
13	IVS 5	g-a nt 454-1	out of frame del Ex 6 codon 152182		Germany	[HolinskiFeder et al., 2001]	N	1
274	IVS 5	g-a nt 454-1	out of frame del Ex 6 codon 152182	co	USA	[Cunningham et al., 2001]	N	1
62	IVS 5	g-t nt 454-1	out of frame del Ex 6 codon 152182, or splice defect	co/HNPCC	Finland	[Aaltonen et al., 1998]	N	1
	Ex 6	C-T nt 479	Ala160Val			Deffenbaugh, unpub.	Y	3
33	Ex 6	ins A nt 504	frameshift from codon 168		USA	de la Chapelle, unpub.	Y	1
	Ex 6	ins AG nt 523	frameshift from codon 174	co		[Quaresima et al., 1998]	N	1
404	Ex 6	GG-AT	Glu177STOP	co/re/ut	USA African	[Weber et al., 1999]	N	1
FF7	Ex 6	A-G nt 544	Arg182Gly	co/b/liver	French	[Wang et al., 1997]	N	2
668	IVS 6	a-g nt 545+3	in frame del Ex 6 codon 152182		Italy	[Pensotti et al., 1997]	Y	1
16	IVS 6	a-g nt 546-2	out of frame del Ex 7 codon 183196	co/ga/k/sa/rp	Sweden	[Tannergard et al., 1995]	Y	1
163	IVS 6	a-g nt 546-2	out of frame del Ex 7 codon 183196	co/re/ut//ly	USA England	[Weber et al., 1997]	Y	1
DUV-SG135	IVS 6	a-g nt 546-2	out of frame del Ex 7 codon 183196	co/ov	France	[Dieumegard et al., 2000]	Y	1
H17	IVS 6	a-g nt 546-2	out of frame del Ex 7 codon 183196	co/ov/br/ut/ga	Sweden	[Planck et al., 1999]	Y	1
OZ-1	Ex 7	T-G nt 554	Val185Gly	co/gb/bd/si/ov	Australia	[KohonenCorish et al., 1996]	Y	2
Family 7	Ex 7	T-G nt 554	Val185Gly	co	Australia	[Scott et al., 2001]	N	2

	Ex 7	T-G nt 554	Val185Gly		Europe		Deffenbaugh, unpub.	Y	2
	Ex 7	T-C nt 577	Ser193Pro	co	Italy		[Panariello et al., 1998]	N	2
SMAL001	Ex 7	del A nt 583	frameshift from codon 195		Malta		Hutter, unpub.	Y	1
FF72	Ex 7	A-T nt 586	Lys196STOP	Co/ut/si/ga	France		[Wang et al., 1999]	N	1
Patient 494	IVS 7	g-a nt 588+5	splice defect?		Czech Republic		Krepelova, unpub.	Y	3
1 kindred	IVS 7	g-c nt 588+11			Scotland		[Farrington et al., 1998]	N	4
207	IVS 7	a-g nt 589-2	out of frame del Ex 8 codon 197226	co/HNPCC	USA		[Luce et al., 1995]	Y	1
A-AV24	IVS 7	a-g nt 589-2	out of frame del Ex 8 codon 197226		Italy		[Viel et al., 1997]	Y	1
DF1751	IVS 7	a-g nt 589-2	out of frame del Ex 8 codon 197226	co/HNPCC	USA	1/180	[Syngal et al., 1999]	Y	1
3003	Ex 8	G-C nt 595	Glu199Gln	co	Sweden		[Wahlberg et al., 1999]	Y	2/3
24	Ex 8	G-A nt 637	Val213Met	co/HNPCC	Portugal		[Fidalgo et al., 2000]	N	2/3
39356	Ex 8	G-A nt 637	Val213Met		Asia		Norbury, unpub.	Y	2/3
9	Ex 8	C-T nt 649	Arg217Cys	co	Japan		[Miyaki et al., 1995]	Y	2/3
SNU-H1006	Ex 8	C-T nt 649	Arg217Cys	co/HNPCC	Korea		[Han et al., 1996]	Y	2/3
15*	Ex 8	C-T nt 649	Arg217Cys	co	Japan		[Nakahara et al., 1997]	N	2/3
GDLV-11#II-9	Ex 8	G-A nt 655	Val219Ile	co	Italy	0.06	[Curia et al., 1999]	N	3/4
78	Ex 8	A-G nt 655	Ile219Val	re	England		[Tomlinson et al., 1997]	N	3/4
Patient 3	Ex 8	A-G nt 655	Ile219Val	co/es	England		[Beck et al., 1997b]	N	3/4
12*	Ex 8	A-G nt 655	Ile219Val	co/ut	Japan		[Nakahara et al., 1997]	N	3/4
	Ex 8	A-G nt 655	Ile219Val		USA	0.13	[Liu et al., 1995a]	Y	3/4

	Ex 8	A-G nt 655	Ile219Val		Italy	0.33	Piepoli, unpub.	Y	3/4
	Ex 8	A-G nt 655	Ile219Val		Sweden	0.34	[Tannergard et al., 1995]	Y	3/4
HNL-1,HNL-2	Ex 8	A-G nt 655	Ile219Val		Japan		[Sasaki et al., 1997]	N	3/4
	Ex 8	A-G nt 655	Ile219Val		Scotland	0.327	[Farrington et al., 1998]	N	3/4
4 kindreds	Ex 8	A-G nt 655	Ile219Val		Switzerland		[Heinimann et al., 1999]	N	3/4
Family 9	Ex 8	A-G nt 655	Ile219Val		Australia		[Scott et al., 2001]	N	3/4
16	Ex 8	A-G nt 655	Ile219Val		Germany		[HolinskiFeder et al., 2001]	N	3/4
	Ex 8	A-G nt 655	Ile219Val		USA	0.396	[Cunningham et al., 2001]	N	3/4
92	Ex 8	A-G nt 655	Ile219Val	co	Japan	0.038	[Nomura et al., 2000]	N	3/4
0434*, 1033*, 1515*	Ex 8	A-G nt 655	Ile219Val	co/HNPCC	Switzerland		[Buerstedde et al., 1995]	N	3/4
F90	Ex 8	A-C nt 655	Ile219Val		Italy		[Montera et al., 2000]	N	3/4
	Ex 8	A-C nt 655	Ile219Leu		USA/Ger	0.31	[Moslein et al., 1996]	Y	3/4
	Ex 8	A-C nt 655	Ile219Leu		Finland		[Liu et al., 1995a]	N	3/4
4082	Ex 8	del A nt 663	frameshift from codon 222		Uruguay		[Sarroca et al., 2000]	Y	1
FF37	Ex 8	del AGTC	frameshift from codon 225226	co/ut/ov	France		[Wang et al., 1999]	N	1
Family 8	Ex 8	del G nt 673	fameshift, STOP	co	Australia		[Scott et al., 2001]	N	1
88 or 12A	Ex 8	C-T nt 676	Arg226STOP	co	Germany		[Moslein et al., 1996]	Y	1
12	Ex 8	C-T nt 676	Arg226STOP	co	Germany		[Wehner et al., 1997]	Y	1
14,15	Ex 8	C-T nt 676	Arg226STOP	co	Germany		[HolinskiFeder et al., 2001]	N	1
331	Ex 8	C-T nt 676	Arg226STOP	co	Japan		[Nomura et al., 2000]	N	1

DF2842	Ex 8	C-T nt 676	Arg226STOP	co/HNPCC	USA		[Syngal et al., 1999]	N	1
6	Ex 8	G-T nt 677	splice defect	co/st	Moldavia		[Maliaka et al., 1996]	Y	1
NL-20	Ex 8	G-A nt 677	Arg226Gln – splice defect	co/re/en/urt/ov /b	Netherlands		[Wijnen et al., 1996]	Y	1
DF168	Ex 8	G-A nt 677	Arg226Gln – splice defect	co/HNPCC	USA		[Syngal et al., 1999]	N	1
	Ex 8	G-A nt 677	Arg226Gln – splice defect		Europe		Deffenbaugh, unpub.	Y	1
	IVS 8	g-a nt 677+1	splice defect		Latin American / Caribbean		Deffenbaugh, unpub.	Y	1
NL-27	IVS 8	del g nt 677+1	splice defect	co/en/k	Netherlands		[Wijnen et al., 1996]	Y	1
NLB-296	IVS 8	ins t nt 677+5	splice defect	co/HNPCC	Netherlands		[Wijnen et al., 1997]	Y	1/2
Cases & Controls	IVS 8	a-g nt 677+39			USA		[Samowitz et al., 2001]	N	4
MD533	IVS 8	del ta nt 678-3	splice defect	co	Scotland		[Farrington et al., 1998]	Y	1
77	IVS 8	del ta nt 678-3	in frame del Ex 910 codon 227295	co	USA		[Liu et al., 1996]	Y	1
F102	IVS 8	g-a	out of frame deletion exon 8	co	Italy		[Montera et al., 2000]	N	1
H38	IVS 8	g-t, splice acceptor site	out of frame deletion exon 9	co	Sweden		[Planck et al., 1999]	Y	1
SNU-H1010	Ex 9	del T nt 692	frameshift from codon 230	co/HNPCC	Korea		[Han et al., 1996]	Y	1
GDLG-20#II1*	Ex 9	G-A nt 702	Glu234Glu	co	Italy	0/100	[Curia et al., 1999]	N	4
Control	Ex 9	G-A nt 702	Glu234Glu		USA		[Samowitz et al., 2001]	N	4
311	Ex 9	G-A nt 731	Gly244Asp	co	Italy		[Pensotti et al., 1997]	Y	2
H6	Ex 9	C-A nt 755	Ser252STOP	co	USA		[Papadopoulos et al., 1994]	Y	1
S1532, GE602	Ex 9	T-G nt 779	Leu260Arg	co	Italy		[Montera et al., 2000]	N	2

VS006	Ex 9	del ATC nt 784	in frame del Ile codon262	co	Portugal		[Hutter et al., 1998]	Y	2
24*	Ex 9	C-T nt 790	splice defect	co	Hong Kong		[Chan et al., 1999]	N	1
GE042	IVS 9	g-c nt 790+1	in frame del Ex 910 codon 227295	co	Spain		[Hutter et al., 1998]	Y	1
Patient 424	IVS 9	g-c nt 790+1	in frame del Ex 910 codon 227295		Czech Republic		Krepelova, unpub.	Y	1
21	IVS 9	g-a nt 790+1	splice defect	co	USA		[Cunningham et al., 2001]	N	1
19	IVS 9	g-a nt 790+1	splice defect		Germany		[HolinskiFeder et al., 2001]	N	1
18	IVS 9	del g nt 790+1	probable splice defect		Germany		[HolinskiFeder et al., 2001]	N	1
SNUH-H7	IVS 9	t-a nt 790+2	splice defect	co/pa	Korea		[Han et al., 1995]	Y	1
JD	IVS 9	ins t nt 790+3	in frame del Ex 910 codon 227295	co	USA		[Liu et al., 1996]	Y	1
OZ-2	IVS 9	ins t nt 790+3	in frame del Ex 910 codon 227295	co/b/k	Australia		[KohonenCorish et al., 1996]	Y	1
67	IVS 9	a-g nt 790+4	splice defect	co	Germany		[Wehner et al., 1997]	Y	2
17	IVS 9	a-g nt 790+4	splice defect	co	Germany		[HolinskiFeder et al., 2001]	N	2
	IVS 9	a-g nt 790+10				0.01	[Wahlberg et al., 1999]	Y	4
Patient 534	IVS 9	a-g nt 790+10			Czech Republic		Krepelova, unpub.	Y	4
1834	IVS 9	del ttagATCGT nt 791-4	frameshift from codon 264		Switzerland		Heinimann, unpub.	Y	1
Case 2	IVS 9	a-g nt 791-2	splice defect	co/ov/es/sp	USA		[Samowitz et al., 2001]	N	1/2
NL-6	IVS 9	g-c nt 791-1	splice defect	co/re	Netherlands		[Wijnen et al., 1996]	Y	1
DF357	IVS 9	g-t nt791-1	splice defect	co	USA		[Syngal et al., 1999]	N	1
Family10	Ex 10	A-G nt 790 (*791*)	splice defect	co	Australia		[Scott et al., 2001]	N	1

33633	Ex 10	A-G nt 791	splice site mutation – unclear pathogenicity		UK		Norbury, unpub.	Y	1
	Ex 10	del ATCG nt 791	frameshift from codon 264	co			[Peltomaki et al., 1997]	N	1
143	Ex 10	del ATCG nt 791	frameshift from codon 264	co	Germany		[Lamberti et al., 1999]		1
20	Ex 10	del ATCG nt 791	frameshift from codon 264	co	Germany		[HolinskiFeder et al., 2001]	N	1
198	Ex 10	C-T nt 793	Arg265Cys	co	Sweden		[Wahlberg et al., 1999]	Y	2/3
198	Ex 10	C-T nt 793	Arg265Cys		Portugal		Hutter, unpub.	Y	2/3
MD1535, MD1654	Ex 10	C-T nt 793	Arg265Cys	co	Scotland		Farrington/Dunlop, unpub.	N	2/3
A-PD1*	Ex 10	G-A nt 794	Arg265His	co/bl	Italy	0/196	[Viel et al., 1997]	Y	2/3
80	Ex 10	A-G nt 803	Glu268Gly	co	Sweden		[Liu et al., 1998a]	Y	2
NL-30	Ex 10	C-G nt 806	Ser269STOP	co/HNPCC	Netherlands		[Wijnen et al., 1997]	Y	1
1805	Ex 10	del TCCTT nt 811-815	frameshift from codon 271		Switzerland		Heinimann, unpub.	Y	1
	Ex 10	C-T nt 842	Ala281Val				Deffenbaugh, unpub.	Y	3
Patient 519	Ex 10	del A nt 856	frameshift from codon 286		Czech Republic		Krepelova, unpub.	Y	1
4	Ex 10	A-C	Lys286Gln	co/HNPCC	England		[Beck et al., 1997c]	Y	2
MD1435	Ex 10	del A nt 861	frameshift from codon 287	co	Scotland		Farrington/Dunlop, unpub.	N	1
NL-37	Ex 10	A-G nt 883	Ser295Thr splice defect?	co/HNPCC	Netherlands		[Wijnen et al., 1997]	Y	1
TF2	Ex 10	A-G nt 883	Ser295Thr splice defect?	co	France		[Wang et al., 1997]	N	1
Control	IVS 10	t-a nt 885-24			USA		[Samowitz et al., 2001]	N	3/4
SNU-H19	Ex 11	ins T nt 888	frameshift from codon 296		Korea		Park, unpub.	Y	1
FF2	Ex 11	G-T nt 889	Glu297STOP	co	France		[Wang et al., 1997]	N	1

A-AV2	Ex 11	C-T nt 901	Gln301STOP	Co/k/br	Italy	[Viel et al., 1997]	Y	1
SIF1	Ex 11	A-T nt 912	Asp304Val	co	Italy	[Montera et al., 2000]	N	2
NL-24	Ex 11	ins GC nt 923	frameshift from codon 307	co/en	Netherlands	[Wijnen et al., 1996]	Y	1
Case 3	Ex 11	ins A nt 938	frameshift from codon 313	co	USA	[Samowitz et al., 2001]	N	1
	Ex 11	del C nt 954	frameshift from codon 318	co	Italy	[Panariello et al., 1998]	N	1
GDLG-31#III-11	Ex 11	del C nt 954	frameshift from codon 318	co	Italy	[Curia et al., 1999]	N	1
1515*	Ex 11	T-C nt 977	Val326Ala	co	Switzerland	[Buerstedde et al., 1995]	Y	3/4
3273	Ex 11	T-C nt 977	Val326Ala	co/HNPCC	USA	[Liu et al., 1996]	Y	3/4
DAC	Ex 11	T-C nt 977	Val326Ala		France	[Dieumegard et al., 2000]	N	3/4
CFS70,CFS136	Ex 11	T-C nt 977	Val326Ala	co	Italy	[Fornasarig et al., 2000]	N	3/4
A-AV18	Ex 11	T-C nt 977	Val326Ala	co/br/ov/ut/re/ ga	Italy	[Genuardi et al., 1998]	N	3/4
GDLG-26#II-4	Ex 11	T-C nt 977	Val326Ala	co	Italy	[Curia et al., 1999]	N	3/4
1515*	Ex 11	T-C nt 977	Val326Ala	co/HNPCC	Switzerland	[Buerstedde et al., 1995]	N	3/4
	Ex 11	A-C nt 986	His329Pro	co	France	[Wang et al., 1997]	N	2/3
96	Ex 11	A-C nt 986	His329Pro	co	Germany	[Lamberti et al., 1999]	Y	2/3
2	Ex 11	A-C nt 986	His329Pro		Germany	[MullerKoch et al., 2001]	N	2/3
21	Ex 11	A-C nt 986	His329Pro	co	Germany	[HolinskiFeder et al., 2001]	N	2/3
RIE-SG125	Ex 11	del A nt 994	frameshift from codon 331		France	[Dieumegard et al., 2000]	Y	1
SNUH-H4	Ex 11	del G nt 1005	frameshift from codon 335	co/en	Korea	[Han et al., 1995]	Y	1
A-PD1*	Ex 11	del C nt 1010	frameshift from codon 337	co/bl	Italy	[Viel et al., 1997]	Y	1

VS003	Ex 11	ins C nt 1012	frameshift from codon 338	co	Switzerland		[Hutter et al., 1998]	Y	1
Patient 339	Ex 11	ins G nt 1027	frameshift from codon 343		Czech Republic		Krepelova, unpub.	Y	1
22	IVS 11	c-t nt 1038+51			Germany		[HolinskiFeder et al., 2001]	N	4
15	IVS 11	c-t nt 1038+51			Germany		[MullerKoch et al., 2001]	N	4
	IVS 11	(TA)n-(T)n length polymorphism				0.73	[Kowalski et al., 1997]	Y	4
	IVS 11	ins a nt1039-7	None apparent		Scotland/USA	0.007	[Farrington et al., 1998]	Y	3/4
39	IVS 11	g-a nt 1039-1	out of frame del Ex 12 codon 347470		Western Europe		[Peltomaki et al., 1997]	Y	1
F16259	Ex 12	8bp del nt 1068	frameshift from codon 356	co	Germany		[Lamberti et al., 1999]	Y	1
23	Ex 12	8bp del nt 1068	frameshift from codon 356	co	Germany		[HolinskiFeder et al., 2001]	N	1
GDLV-52#II-2	Ex 12	A-G nt 1090	Thr364Ala		Italy		[Curia et al., 1999]	N	2/3
F33,F4	Ex 12	G-A nt 1132/del TC nt 1133	frameshift from codon 378		Italy		Piepoli, unpub.	Y	1
319	Ex 12	T-A nt 1151	Val384Asp	co	Japan	0.038	[Nomura et al., 2000]	N	2/3
	Ex 12	T-A nt 1151	Val384Asp		Japan	0.03	[Konishi et al., 1996]	N	2/3
Family 11	Ex 12	ins 4bp nt 1164	stop codon	co	Australia		[Scott et al., 2001]	N	1
Case 12	Ex 12	C-T nt 1165	Arg389Trp	co	USA		[Samowitz et al., 2001]	N	2
MD1529	Ex 12	G-A nt 1166	Arg389Gln	co	Scotland		Farrington/Dunlop, unpub.	N	2
	Ex 12	G-A nt 1166	Arg389Gln		Latin American / Caribbean		Deffenbaugh, unpub.	Y	2
DP6	Ex 12	del T nt 1190	frameshift	co/ut	Canada		[Millar et al., 1999]	N	1
MD442	Ex 12	del T nt 1211	frameshift from codon 404	co	Scotland		Farrington/Dunlop, unpub.	N	1
CFS111	Ex 12	G-A nt 1217	Ser406Asn	co	Italy		[Fornasarig et al., 2000]	N	2/3

	Ex 12	G-A nt 1217	Ser406Asn		Finland	0.015	[Wu et al., 1997]	Y	2/3
209	Ex 12	G-A nt 1217	Ser406Asn	co	USA		[Cunningham et al., 2001]	N	2/3
Control	Ex 12	G-A nt 1217	Ser406Asn		USA		[Samowitz et al., 2001]	N	2/3
HNPC-036	Ex 12	ins GTCAGCC nt 1217	frameshift from codon 406	co	England		[Bapat et al., 1999]	Y	1
2	Ex 12	G-A nt 1321	Ala441Thr	co/br	Sweden		[Tannergard et al., 1995]	Y	2/3
Patient 466	Ex 12	G-A nt 1321	Ala441Thr	co	Czech Republic		Krepelova, unpub.	Y	2/3
83	Ex 12	G-A nt 1321	Ala441Thr	co	USA		[Cunningham et al., 2001]	N	2/3
NET	Ex 12	-	frameshift codon 450	co/HNPCC	England		[Beck et al., 1997a]	N	1
DF1846	Ex 12	A-T nt 1381	Lys461STOP	co/HNPCC	USA		[Syngal et al., 1999]	N	1
PB	Ex 12	2 bp del	frameshift codon 461	co/HNPCC	England		[Beck et al., 1997a]	N	1
RA	Ex 12	Ex 12 deleted	deletion Ex 12 codon 347470	co/HNPCC	USA		[Papadopoulos et al., 1994]	N	1
Q1	Ex 12	Ex 12 deleted	deletion Ex 12 codon 347470	co/sk/HNPCC	French Canada		[Bapat et al., 1999]	N	1
NL67	Ex 12	genomic del of ~25kb	unknown		Netherlands		Wijnen, unpub.	Y	1
8	IVS 12	g-c nt 1409+1	out of frame del Ex 12 codon 347470		Finland		Holmberg, unpub.	Y	1
MD949	IVS 12	g-t nt 1409+1	out of frame del Ex 12 codon 347470	co/ut	Scotland		Farrington/Dunlop, unpub.	N	1
25	IVS 12	g-a nt 1406*(1410)*-14			Germany		[HolinskiFeder et al., 2001]	N	3/4
MD817	IVS 12	1.2kb intronic del	in frame del Ex 13 codon 470520	co	Scotland		[Farrington et al., 1998]	Y	1
MS	Ex 13	del AAAG nt 1410	frameshift from codon 470	co/HNPCC	USA		[Liu et al., 1996]	Y	1
24	Ex 13 *(Ex12)*	del AGAA(G) nt 1411 (*or AGAA nt 1408)*	frameshift from codon 471		Germany		[HolinskiFeder et al., 2001]	N	1
CC22	Ex 13	del C nt 1420	frameshift from codon 474		Spain		Caldes Trnidad, unpub.	Y	1

DES-SG407	Ex 13	G-A nt 1421	Arg474Gln		Japan	[Lu et al., 1996]	Y	2
C009	Ex 13	del A nt 1448	frameshift from codon 483	co	England	[Froggatt et al., 1996]	Y	1
	Ex 13	T-A nt 1456	Asp485Glu	co/HNPCC	Switzerland	[Heinimann et al., 1999]	N	2/3
h5	Ex 13	C-T nt 1459	Arg487STOP		England	Bunyan, unpub.	Y	1
MAR-SG363	Ex 13	C-T nt 1459	Arg487STOP		France	[Dieumegard et al., 2000]	Y	1
CC29	Ex 13	C-T nt 1459	Arg487STOP		Spain	Caldes Trinidad, unpub.	Y	1
Patient 470	Ex 13	C-T nt 1459	Arg487STOP		Czech Republic	Krepelova, unpub.	Y	1
2,8	Ex 13	C-T nt 1459	Arg487STOP	co/HNPCC	Portugal	[Fidalgo et al., 2000]	N	1
MD1269	Ex 13	C-T nt 1459	Arg487STOP	co	Scotland	Farrington/Dunlop et al., unpub.	N	1
Family 15	Ex 13	del TGAT nt 1460	stop codon	co	Australia	[Scott et al., 2001]	N	1
1S or A4T	Ex 13	G-A nt 1474	Ala492Thr	co	USA	[Moslein et al., 1996]	Y	2
5A or 17383	Ex 13	ins C nt 1490	frameshift from codon 497	co/re	USA	[Moslein et al., 1996]	Y	1
CRC11	Ex 13	ins C nt 1490	frameshift from codon 497	co	Japan	[Sasaki et al., 1997]	Y	1
10,11,12,14	Ex 13	ins C nt 1490	frameshift from codon 497	co/ut/ov/ga/re	Germany/Czech republic	[Pistorius et al., 2000]	N	1
144	Ex 13	ins C nt 1490	frameshift from codon 497	co	Germany	[Lamberti et al., 1999]	Y	1
CH1	Ex 13	ins C nt 1490	frameshift from codon 497	co/HNPCC	Czech	[Wijnen et al., 1997]	Y	1
Patient 469	Ex 13	ins C nt 1490	frameshift from codon 497		Czech Republic	Krepelova, unpub.	Y	1
26	Ex 13	ins C nt 1490	frameshift from codon 497		Germany	[HolinskiFeder et al., 2001]	N	1
	Ex 13	del G nt 1490	frameshift from codon 497	co	Finland	[Peltomaki et al., 1997]	N	1
241-1	Ex 13	del G nt 1490	frameshift from codon 497	co/re/ga	USA England	[Weber et al., 1997]	Y	1

3118	Ex 13	T-C nt 1517	Val506Ala	co/HNPCC	USA		[Liu et al., 1996]	Y	2/3
DF260*	Ex 13	T-C nt 1517	Val506Ala	co/HNPCC	USA		[Syngal et al., 1999]	N	2/3
	Ex 13	T-C nt 1517	Val506Ala		Native American		Deffenbaugh, unpub.	Y	2/3
Family 14	Ex 13	G-T nt 1535	stop codon	co	Australia		[Scott et al., 2001]	N	1
GC7237	Ex 13	ins T nt 1553	Glu519STOP	co/cx/b/br/ov/ bc/rp/l	England		[Kolodner et al., 1995]	Y	1
Family 13	Ex 13	ins T nt 1554	stop codon	co	Australia		[Scott et al., 2001]	N	1
645	Ex 13-16	22.4kb deletion (Alu mediated)	deletion Ex 13-16 codon 470632	co/en	France		[Mauillon et al., 1996]	Y	1
A-MD10	IVS 13	g-t nt 1558+1	splice defect		Italy		[Viel et al., 1997]	Y	1
	IVS 13	g-a nt 1558+10			Spain		Trinidad, unpub.	Y	4
	IVS 13	g-a nt 1558+11		co/sk/br/d/pa	Switzerland		[Hutter et al., 1996]	Y	4
Family 12	IVS13	t-a nt 1558+13	cryptic splice site	co	Australia		[Scott et al., 2001]	N	2/3
	IVS 13	g-a nt 1558+14			Sweden	0.06	[Tannergard et al., 1995]	Y	4
	IVS 13	g-a nt 1558+14			Germany	0.08	[Wehner et al., 1997]	N	4
	IVS 13	g-a nt 1558+14			USA	0.042	[Cunningham et al., 2001]	N	4
	IVS 13	g-a nt 1558+14			Scotland/USA	0.014	[Farrington et al., 1998]	N	4
cases & controls	IVS 13	g-a nt 1558+14			USA		[Samowitz et al., 2001]	N	4
	IVS 13	g-a nt 1558+14			Italy		[Percesepe et al., 2001]	N	4
27	IVS 13	g-a nt 1558+14	NA		Germany		[HolinskiFeder et al., 2001]	N	4
85	IVS 13	a-c nt 1559-2	out of frame del Ex 14 codon 520556		Finland		Holmberg, unpub.	Y	1
40	IVS 13	g-t nt 1559-1	out of frame del Ex 14 codon	co/HNPCC	Finland		[NystromLahti et al., 1996]	Y	1

			520556						
39992	IVS 13	a-g nt 1580-2			UK		Norbury, unpub.	Y	1
	Ex 14	del GT nt 1572	frameshift – STOP codon 526				Deffenbaugh, unpub.	Y	1
Case 13	Ex 14	C-T nt 1590	Phe530Phe	co	USA		[Samowitz et al., 2001]	N	4
113	Ex 14	del C nt 1622	frameshift from codon 540	co	Germany		[Lamberti et al., 1999]	Y	1
28	Ex 14	del C nt 1622	frameshift from codon 540	co	Germany		[HolinskiFeder et al., 2001]	N	1
SNUH-H2	Ex 14	A-T nt 1625	Gln542Leu	co/HNPCC	Korea		[Han et al., 1995]	Y	2
5	Ex 14	T-A nt 1640	Leu547STOP	co	Germany		[Lamberti et al., 1999]	Y	1
29	Ex 14	T-A nt 1640	Leu547STOP	co	Germany		[HolinskiFeder et al., 2001]	N	1
I-771	Ex 14	ins TTATA nt 1644	frameshift from codon 548		Italy		Radice and Bertario, unpub.	Y	1
SNU-H14	Ex 14	T-C nt 1646	Leu549Pro	co/HNPCC	Korea		[Han et al., 1996]	Y	2/3
TF1	Ex 14	A-C nt 1652	Asp551Thr	co	French		[Wang et al., 1997]	N	2
VS012	Ex 14	A-C nt 1652	Asp551Thr		Switzerland		[Hutter et al., 1998]	Y	2
30	Ex 14	C-T nt 1652*(3)*	Asp551AAT?		Germany		[HolinskiFeder et al., 2001]	N	?
88	Ex 14 – 15	Ex 14-15	out of frame del Ex 14-15	co	Sweden		[Wahlberg et al., 1999]	Y	1
12,18,21,31,41	IVS 14	4bp ins/ 3bp del nt 1667+2	silencing of allele	co/ut/am	Denmark		[Jager et al., 1997]	Y	1
5	IVS 14	3’splice site+7	aberrant transcript	co	England		[Beck et al., 1997c]	N	2/3
	IVS 14	a-g nt 1668-19				0.38	Miller, unpub.	Y	4
		a-g nt 1668-19			Germany	0.5	[Wehner et al., 1997]	Y	4

0434*,1033*, 1515*,1553*	IVS 14	a-g nt 1668-19			Switzerland		[Buerstedde et al., 1995]	Y	4
		a-g nt 1668-19			Sweden	0.4	[Tannergard et al., 1995]	Y	4
	IVS 14	a-g nt 1668-19			Sweden	0.4	[Liu et al., 1995a]	Y	4
CC-7	IVS 14	a-g nt 1668-19			Spain		Caldes Trinidad, unpub.	Y	4
	IVS 14	a-g nt 1668-19			Scotland/USA	0.063	[Farrington et al., 1998]	N	4
	IVS 14	a-g nt 1668-19			USA	0.56	[Cunningham et al., 2001]	N	4
cases & controls	IVS 14	a-g nt 1668-19			USA		[Samowitz et al., 2001]	N	4
MAL-SG164	IVS 14	c-a nt 1668-3	deletion Ex 15 codon 556577		France		[Dieumegard et al., 2000]	Y	1
FF48, FF70	Ex 15	G-T nt 1672	Glu558STOP	co	France		[Wang et al., 1999]	N	1
OG/97-4266	Ex 15	C-T nt 1684	Gln562STOP	co	Slovenia		[RavnikGlavac et al., 2000]	N	1
1808	Ex 15	del CTCA nt 1690-1693	frameshift from codon 564		Switzerland		Heinimann, unpub.	Y	1
VD001	Ex 15	A-T nt 1693	Ile565Phe	co	Switzerland		[Hutter et al., 1998]	Y	3
SNUH-H1	Ex 15	T-C nt 1721	Leu574Pro	co/ut/sk/re	Korea		[Han et al., 1995]	Y	2/3
OZ-4	Ex 15	G-A nt 1731	out of frame del Ex 15 codon 556577	co/en/ov	Australia		[KohonenCorish et al., 1996]	Y	1
1801	Ex 15	G-A nt 1731	out of frame del Ex 15 codon 556577		Switzerland		Heinimann, unpub.	Y	1
13	Ex 15	G-A nt 1731	out of frame del Ex 15 codon 556577	co	Germany/Czech Republic		[Pistorius et al., 2000]	N	1
Family 16	Ex 15	G-A nt 1731	out of frame del Ex 15 codon 556577	co	Australia		[Scott et al., 2001]	N	1
EB	Ex 15	Ex 15 deleted	out of frame del Ex 15 codon 556577	co/HNPCC	USA		[Liu et al., 1996]	N	1
NL-25	IVS 15	g-a nt 1731+1	splice defect	co/en	Netherlands		[Wijnen et al., 1996]	Y	1

F197	IVS 15	g-a	out of frame del exon 15	co	Italy	[Montera et al., 2000]	N	1
D-105	IVS 15	a-t nt 1732-2	in frame del Ex 16 codon 578632	co/HNPCC	Denmark	[Wijnen et al., 1996]	Y	1
3, 5	IVS 15	a-t nt 1732-2	in frame del Ex 16 codon 578632	co/ut/bl/si	Denmark	[Jager et al., 1997]	Y	1
21	Ex 16	A-G nt 1733	Glu578Gly	co/pa	Sweden	[Tannergard et al., 1995]	Y	2
ENDO003	Ex 16	A-G nt 1733	Glu578Gly		USA	de la Chapelle, unpub.	Y	2
JPN-1	Ex 16	C-G nt 1744	Leu582Val	co	Japan	[Han et al., 1995]	Y	2/3
31	Ex 16	T-C nt 1745	Leu582Pro		Germany	[HolinskiFeder et al., 2001]	N	2/3
01	Ex 16	del TT nt 1747	frameshift from codon 583		Lithuania	Avizienyte – unpub	Y	1
4 kindreds	Ex 16	ins C nt 1756	frameshift from codon 586	co/pa/en/ov	Korea	[Han et al., 1996;Han et al., 1995;Yuan et al., 1998]	Y	1
	Ex 16	ins C nt 1758	frameshift from codon 586 – STOP codon at 592		Asia	Deffenbaugh, unpub.	Y	1
	Ex 16	del T nt 1764	frameshift from codon 588	co	Korea	[Yuan et al., 1998]	N	1
63	Ex 16	del T nt 1769	frameshift from codon 590	co/br	Sweden	[Liu et al., 1998a]	Y	1
316 or 3F	Ex 16	del TAGA nt 1769	frameshift from codon 590	co	USA	[Moslein et al., 1996]	Y	1
15980 or 13A	Ex 16	del TAGA nt 1769	frameshift from codon 590	co/en	Germany	[Moslein et al., 1996]	Y	1
NL-28	Ex 16	del AG nt 1779	frameshift from codon 593	co/en	Netherlands	[Wijnen et al., 1996]	Y	1
Patient 531	Ex 16	del AG nt 1783-84	frameshift from codon 595		Czech Republic	Krepelova, unpub.	Y	1
CFS25	Ex 16	del AG	frameshift from codon 595	co	Italy	[Fornasarig et al., 2000]	N	1
A-AV14	Ex 16	del AG	frameshift from codon 595	co/sk/ga/l	Italy	[Genuardi et al., 1998]	N	1
32	Ex 16	del TAGA nt 1790*(1769?)*	frameshift?		Germany	[HolinskiFeder et al., 2001]	N	1

33	Ex 16	C-G nt 1802*(1807)?*	*Pro603Arg?*		Germany	[HolinskiFeder et al., 2001]	N	?
3	Ex 16	C-G nt 1807?	*Pro603Arg?*		Germany	[MullerKoch et al., 2001]	N	?
DF2722	Ex 16	A-T nt 1810	Lys604STOP	co	USA	[Syngal et al., 1999]	N	1
MD770	Ex 16	G-C nt 1818	Gly606Ala		Scotland	Farrington/Dunlop, unpub.	N	3/4
14	Ex 16	T-A nt 1820	Leu607His	co/HNPCC	Portugal	[Fidalgo et al., 2000]	N	2
h78	Ex 16	ins T nt 1822	frameshift from codon 607		England	Bunyan, unpub.	Y	1
MD329	Ex 16	del AAG nt 1846	in frame deletion Lys codon 616	co	Scotland	[Farrington et al., 1998]	Y	2
HNP-8	Ex 16	del AAG nt 1846	in frame deletion Lys codon 616	co	Japan	[Miyaki et al., 1995]	Y	2
NL-29,NL-40,NL-59	Ex 16	del AAG nt 1846	in frame deletion Lys codon 616	co/en/pa/st/re/ HNPCC	Netherlands	[Wijnen et al., 1996]	Y	2
I-202	Ex 16	del AAG nt 1846	in frame deletion Lys codon 616	co/en/urt	Italy	[Wijnen et al., 1996]	Y	2
42/43	Ex 16	del AAG nt 1846	in frame deletion Lys codon 616	co	USA Germany	[Weber et al., 1997]	Y	2
R-RM3	Ex 16	del AAG nt 1846	in frame deletion Lys codon 616	co/st/si	Italy	[Viel et al., 1997]	Y	2
GUE-SG618	Ex 16	del AAG nt 1846	in frame deletion Lys codon 616		France	[Dieumegard et al., 2000]	Y	2
1760	Ex 16	del AAG nt 1846	in frame deletion Lys codon 616		Switzerland	Heinimann, unpub.	Y	2
JR or P4	Ex 16	del AAG nt 1846	in frame deletion Lys codon 616	co/HNPCC	USA	[Liu et al., 1996]	Y	2
14	Ex 16	del AAG nt 1846	in frame deletion Lys codon 616	co/TS	USA	[Hamilton et al., 1995]	Y	2
15980	Ex 16	del AAG nt 1846	in frame deletion Lys codon 616		USA	[Moslein et al., 1996]	Y	2
5 Kindreds	Ex 16	del AAG nt 1846	in frame deletion Lys codon 616	co/bl/b/br/du/ ut/si/sk/ga/m/ urt/l/ov	France	[Wang et al., 1999]	N	2
DF336	Ex 16	del AAG nt 1846	in frame deletion Lys codon 616	co	USA	[Syngal et al., 1999]	N	2

GDLG-29#III-8	Ex 16	del AAG nt 1846	in frame deletion Lys codon 616	co	Italy		[Curia et al., 1999]	N	2
32	Ex 16	del AAG nt 1846	in frame deletion Lys codon 616	co	England		[Coleman et al., 2001]	N	2
34	Ex 16	del AG nt 1848*(7)*	frameshift from codon 616		Germany		[HolinskiFeder et al., 2001]	N	1/2
Case 4	Ex 16	del AAG nt 1852-1854	in frame deletion lys codon 618	co	USA		[Samowitz et al., 2001]	N	2
GE111	Ex 16	A-T nt 1852	Lys618STOP	co	Switzerland		[Hutter et al., 1998]	Y	1
MD696	Ex 16	AA-GC nt 1852	Lys618Ala	co	Scotland		[Farrington et al., 1998]	Y	2/3
55-1	Ex 16	AA-GC nt 1852	Lys618Ala	co/re/rp/ly	USA/England		[Weber et al., 1999]	Y	2/3
756	Ex 16	AA-GC nt 1852	Lys618Ala	co/re	France		[Mauillon et al., 1996]	Y	2/3
16	Ex 16	AA-GC nt 1852	Lys618Ala		Netherlands		[Tannergard et al., 1995]	Y	2/3
CC12	Ex 16	AA-GC nt 1852	Lys618Ala		Spain		Caldes Trinidad, unpub.	Y	2/3
VD007	Ex 16	AA-GC nt 1852	Lys618Ala		Switzerland		Hutter, unpub.	Y	2/3
51**	Ex 16	AA-GC nt 1852	Lys618Ala		USA		de la Chapelle, unpub.	Y	2/3
Family 17	Ex 16	AA-GC nt 1852	Lys618Ala	co	Australia		[Scott et al., 2001]	N	2/3
28	Ex 16	AA-GC nt 1852	Lys618Ala	co/HNPCC	Portugal		[Fidalgo et al., 2000]	N	2/3
DF260*	Ex 16	AA-GC nt 1852	Lys618Ala	co/HNPCC	USA	1/186	[Syngal et al., 1999]	N	2/3
NLB-526	Ex 16	AA-GC nt 1852	Lys618Ala	co/HNPCC	Netherlands		[Wijnen et al., 1997]	Y	2/3
Case 14, control	Ex 16	AA-GC nt 1852	Lys618Ala		USA		[Samowitz et al., 2001]	N	2/3
	Ex 16	AA-GC nt 1852	Lys618Ala			0.99%	Deffenbaugh, unpub.	Y	2/3
4	Ex 16	AA-GC nt 1852	Lys618Ala		Germany		[MullerKoch et al., 2001]	N	2/3
6	Ex 16	A-G nt 1852	Lys618Gln	co/HNPCC	England		[Beck et al., 1997c]	N	2/3
18	Ex 16	A-G nt 1853	Lys618Arg	co	Germany/Czech Republic		[Pistorius et al., 2000]	N	2/3

	Ex 16	A-C nt 1853	Lys618Thr	co	Portugal	[Cravo et al., 1999]	N	2/3
	Ex 16	A-C codon 618	Lys618Tyr (*Thr*)	co	Italy	[de Leon et al., 1999]	N	2/3
F103	Ex 16	A-C nt 1853	Lys618Thr		Italy	[Montera et al., 2000]	N	2/3
US-6/4A	Ex 16	A-C nt 1853	Lys618Thr	co	USA	[Han et al., 1995]	Y	2/3
35	Ex 16	AA-GC nt 1855/1856 *(1852?)*	*Lys618Ala?*		Germany	[HolinskiFeder et al., 2001]	N	2/3
Case 5*	Ex 16	del T nt 1877	frameshift from codon 626	co/ut	USA	[Samowitz et al., 2001]	N	1
73	Ex 16	del TCTCTTT nt 1877	frameshift from codon 626	co/en/br	France	[Mauillon et al., 1996]	Y	1
EC	Ex 16	TTC.TCT-TCC.ACT	Phe.SerSer.Thr(626/627)	co/HNPCC	England	[Beck et al., 1997a]	N	2
MTS-K14	Ex 16	del GGAAA nt 1884	frameshift from codon 628	MT/sa	Germany	[Kruse et al., 1998]	Y	1
NL-3,NL-4,NL-204	Ex 16	G-A nt 1896	in frame del Ex 16 codon 578632	co/HNPCC	Netherlands	[Wijnen et al., 1996]	Y	1
	Ex 16	del G nt 1896	frameshift /splicing defect	co/HNPCC	Switzerland	[Heinimann et al., 1999]	N	1
NL-14	Ex 16	genomic del of 5.5kb			Netherlands	Wijnen, unpub.	Y	1
14 kindreds	Ex 16	3.5kb deletion	in frame del Ex 16 codon 578632	co/HNPCC	Finland	[NystromLahti et al., 1995]	Y	1
20 kindreds	Ex 16	3.5kb deletion	in frame del Ex 16 codon 578632	co/HNPCC	Finland	[NystromLahti et al., 1996]	N	1
9 Kindreds	Ex 16	3.5kb deletion	in frame del Ex 16 codon 578632	co/HNPCC	Finland	[Aaltonen et al., 1998]	N	1
5 Kindreds	Ex 16	3.5kb deletion	in frame del Ex 16 codon 578632	co	Finland	[Salovaara et al., 2000]	N	1
7 Kindreds	Ex 16	3.5kb deletion	in frame del Ex 16 codon 578632	co/HNPCC	Finland	[Papadopoulos et al., 1994]	N	1
7,14,27,58,98, 131,142	Ex 16	3.5kb deletion	in frame del Ex 16 codon 578632		Sweden/Finland	[Tannergard et al., 1995]	Y	1
VS009	Ex 16	3.5kb deletion	in frame del Ex 16 codon 578632		Switzerland	[Hutter et al., 1998]	Y	1
K-25/HNP85	Ex 16	Ex 16 deleted	deletion Ex 16 codon 578632	co/en	Japan	[Akiyama et al., 1996]	N	1

Case 2	IVS16	g-t in splice donor site	splice defect	co	Sweden		[Nilbert et al., 1999]	N	1
DF 397	IVS 16	g-a nt 1896+1		co/HNPCC	USA		[Syngal et al., 1999]	N	1
1033*	IVS 16	del G nt 1896+1	frameshift /splicing defect	co	Switzerland		[Buerstedde et al., 1995]	Y	1
1 kindred	Ex 17	G-A nt 1908	Leu636Leu		Scotland/USA		[Farrington et al., 1998]	N	4
36	Ex 17	C-T nt 1943	Pro648Leu		Germany		[HolinskiFeder et al., 2001]	N	2/3
5	Ex 17	C-T nt 1943	Pro648Leu		Germany		[MullerKoch et al., 2001]	N	2/3
	Ex 17	T-G nt 1958	Leu653Arg		Europe		Deffenbaugh, unpub.	Y	2/3
37	Ex 17	G-T nt 1958 *(1959)*	Leu653Leu?		Germany		[HolinskiFeder et al., 2001]	N	4
Family 18	Ex 17	G-T nt 1959	Leu653Leu	co	Australia		[Scott et al., 2001]	N	4
	Ex 17	G-T nt 1959	Leu653Leu		Sweden	0.0038	[Liu et al., 1995a]	Y	4
1533*	Ex 17	G-T nt 1959	Leu653Leu	co/HNPCC	Switzerland		[Buerstedde et al., 1995]		4
SKI	Ex 17	G-T nt 1959	Leu653Leu		Slovakia		Bartosova & Fridrichova, unpub.	Y	4
1 kindred	Ex 17	G-T nt 1959	Leu653Leu		Scotland/USA		[Farrington et al., 1998]	N	4
CC-32	Ex 17	G-T nt 1959	Leu653Leu		Spain		Caldes Trinidad, unpub.	Y	4
36003	Ex 17	G-T nt 1959	Leu653Leu		UK		Norbury, unpub.	Y	4
	Ex 17	G-T nt 1959	Leu653Leu	co	USA	0.083	[Cunningham et al., 2001]	N	4
cases & controls	Ex 17	G-T nt 1959	Leu653Leu		USA		[Samowitz et al., 2001]	N	4
	Ex 17	G-T nt 1959	Leu653Leu		Italy		[Percesepe et al., 2001]	N	4
38469	Ex 17	C-T nt 1961	Pro654Leu		UK		Norbury, unpub.	Y	2/3

OG/97-3941	Ex 17	T-C nt 1964	Ile655Thr	co	Slovenia	[RavnikGlavac et al., 2000]	N	2/3
38	Ex 17	T-C nt 1964	Ile655Thr		Germany	[HolinskiFeder et al., 2001]	N	2/3
MD804	Ex 17	C-T nt 1975	Arg659STOP	co/ut	Scotland	[Farrington et al., 1998]	Y	1
72,83	Ex 17	C-T nt 1975	Arg659STOP	co/en/HNPCC	Finland	[NystromLahti et al., 1996] & unpub.	Y	1
179	Ex 17	C-T nt 1975	Arg659STOP	co/ut	Finland	[Aaltonen et al., 1998]	N	1/2
A262	Ex 17	C-T nt 1975	Arg659STOP	co	Finland	[Loukola et al., 1999]		1
C219	Ex 17	C-T nt 1975	Arg659STOP	co	Finland	[Wu et al., 1997]	N	1
20198 or 5F	Ex 17	C-T nt 1975	Arg659STOP	co	USA	[Moslein et al., 1996]	Y	1
OZ-3	Ex 17	del CG nt 1975	in frame del Ex 17 codon 633663	co/b/en/re	Australia	[KohonenCorish et al., 1996]	Y	1
Family 20	Ex 17	del GA nt 1975 (*1976*)	stop codon	co	Australia	[Scott et al., 2001]	N	1
7	Ex 17	G-C nt 1976	Arg659Pro	co/HNPCC	Finland	[NystromLahti et al., 1996]	Y	2
NL-56	Ex 17	G-C nt 1976	Arg659Pro	co/HNPCC	Netherlands	Wijnen, unpub.	Y	2
51*	Ex 17	G-A nt 1976	Arg659Glu		USA	de la Chapelle, unpub.	Y	2
1	Ex 17	G-T nt 1976	Arg659Leu	co/HNPCC	Portugal	[Fidalgo et al., 2000]	N	2
39	Ex 17	A-C nt 1985 *(1984)*	*Thr662Pro*		Germany	[HolinskiFeder et al., 2001]	N	2/3
6	Ex 17	A-C nt 1984 (*?*)	*Thr662Pro*		Germany	[MullerKoch et al., 2001]	N	2/3
CHI-SG277	Ex 17	A-G nt 1988	deletion Ex 17 codon 633663		France	[Dieumegard et al., 2000]	Y	1
Family 19	Ex 17	G-A nt 1989	stop codon	co	Australia	[Scott et al., 2001]	N	1

I-337,I670	Ex 17	G-T nt 1989	splice defect		Italy	Radice and Bertario, unpub.	Y	1
FF66	Ex 17	G-T nt 1989	in frame del exon 17	co/br/bl/ga/ov/du	France	[Wang et al., 1999]	N	1
SCA	Ex 17	-	del Ex 17		France	[Dieumegard et al., 2000]	N	
156	IVS 17	g-t nt 1989+1	in frame del Ex 17 codon 633663	co	Germany	[Lamberti et al., 1999]	Y	1
40	IVS 17	g-t nt 1989+1	in frame del Ex 17 codon 633663	co	Germany	[HolinskiFeder et al., 2001]	N	1
R-MD6	IVS 17	g-t nt 1989+5	in frame del Ex 17 codon 633663	co/ga/ut/br	Italy	[Viel et al., 1997]	Y	1
16	IVS 17	t-g nt1990-22			Germany	[MullerKoch et al., 2001]	N	3/4
Patient 478	IVS 17	c-g nt 1990-3	splice defect?		Czech Republic	Krepelova, unpub.	Y	2
FF5	IVS 17	del nt 1990-16 --> -2	in frame del exon 18	co/si	France	[Wang et al., 1999]	N	1
SNU-YC32	Ex 18	C-T	Asp667Asp		Korea	[Yuan et al., 1998]	N	4
Patient 386	Ex 18	G-T nt 2028	Leu676Leu		Czech Republic	Krepelova , unpub.	Y	4
C010	Ex 18	G-A nt 2041	Ala681Thr	co	England	[Froggatt et al., 1996]	Y	2/3
BT-1881/WH-821	Ex 18	G-A nt 2041	Ala681Thr		Poland	Kurzawski, unpub.	Y	2/3
Case 6	Ex 18	G-A nt 2041	Ala681Thr		Poland	[Debniak et al., 2000]	Y	2/3
7	Ex 18	A-G nt 2066	Gln689Arg		Germany	[MullerKoch et al., 2001]	N	2/3
SGE109	Ex 18	C-G nt 2093	Ser698STOP		Switzerland	Hutter, unpub.	Y	1
NL-9,NLB-35	IVS 18	g-a nt 2103+1	Splice defect	co/st/en/b/k/sk/HNPCC	Netherlands	[Wijnen et al., 1996], [Wijnen et al., 1997]	Y	1
1813	IVS 18	g-a nt 2103+1	splice defect		Switzerland	Heinimann, unpub.	Y	1
IRI-SG119	IVS 18	g-a nt 2103+3	splice defect		France	[Dieumegard et al., 2000]	Y	2

1121*	IVS 18	a-t nt 2104-2	splice defect	co/b/ov/ur/re	Switzerland		[Buerstedde et al., 1995]	Y	1
	Ex 19	A-T nt 2104	splice defect	co/HNPCC	Switzerland		[Heinimann et al., 1999]	N	1
DF1448	Ex 19	del AG nt 2104-2105	splice site loss	co/HNPCC	USA		[Syngal et al., 1999]	N	1
	Ex 19	C-T nt 2118	Gly706Gly	co	Scotland	0.035	[Farrington et al., 1998]	N	4
AM	Ex 19	G-A nt 2135	Trp712STOP	co/HNPCC	USA		[Liu et al., 1996]	Y	1
h44,h87	Ex 19	G-A nt 2135	Trp712STOP		England		Bunyan, unpub.	Y	1
29	Ex 19	G-A nt 2135	Trp712STOP	co	England		[Coleman et al., 2001]	N	1
VS004,008,010	Ex 19	G-A nt 2141	Trp714STOP	co/ut/st/gl/br/sk	Switzerland		[Hutter et al., 1998]	Y	1
	Ex 19	G-A nt 2141	Trp714STOP	co/HNPCC	Switzerland		[Heinimann et al., 1999]	N	1
C008	Ex 19	G-A nt 2141	Trp714STOP	co	England		[Froggatt et al., 1996]	Y	1
H50	Ex 19	G-A nt 2141	Trp714STOP	co	Sweden		[Planck et al., 1999]	Y	1
GE111*	Ex 19	G-A nt 2146	Val716Met	co	Switzerland		[Hutter et al., 1998]	Y	2/3
CFS56,CFS144	Ex 19	G-A nt 2146	Val716Met	co	Italy		[Fornasarig et al., 2000]	N	2/3
A-AV23	Ex 19	G-A nt 2146	Val716Met	co/ut	Italy		[Genuardi et al., 1998]		2/3
AUB	Ex 19	G-A nt 2146	Val716Met	co/l	France		[Dieumegard et al., 2000]	N	2/3
DF1751	Ex 19	G-A nt 2146	Val716Met	co/HNPCC	USA		[Syngal et al., 1999]	N	2/3
Family 22	Ex 19	G-A nt 2146	Val716Met	co	Australia		[Scott et al., 2001]	N	2/3
	Ex 19	G-A nt 2146	Val716Met		Netherlands	0/100	[Wijnen et al., 1998a]	N	2/3
41	Ex 19	G-A nt 2146	Val716Met		Germany		[HolinskiFeder et al., 2001]	N	2/3
8,9	Ex 19	G-A nt 2146	Val716Met		Germany		[MullerKoch et al., 2001]	N	2/3

Case 15	Ex 19	G-A nt 2146	Val716Met	co	USA		[Samowitz et al., 2001]	N	2/3
CHO	Ex 19	C-T nt 2152	His718Tyr		France		[Dieumegard et al., 2000]	N	2/3
Case 16	Ex 19	C-T nt 2152	His718Tyr	co	USA		[Samowitz et al., 2001]	N	2/3
	Ex 19	C-T nt 2152	His718Tyr		Scotland	0.021	[Farrington et al., 1998]	N	2/3
3	Ex 19	T-A nt 2163	Tyr721STOP	co/HNPCC	Portugal		[Fidalgo et al., 2000]	N	1
154	Ex 19	ins AAATAT nt 2167	in frame insertion codon 723	co	Germany		[Lamberti et al., 1999]	Y	1
42	Ex 19	ins AAATAT nt 2167	in frame insertion codon 723	co	Germany		[HolinskiFeder et al., 2001]	N	1
24	Ex 19	del CA nt 2177	frameshift from codon 726	co/ph	France		[Mauillon et al., 1996]	Y	1
L7	Ex 19	del CACA nt 2177	frameshift from codon 726	co/HNPCC	USA		[Papadopoulos et al., 1994]	Y	1
	Ex 19	C-G nt 2185	Leu729Val		Scotland	0.021	[Farrington et al., 1998]	N	2/3
HNP-004	Ex 19	ins AACA nt 2190	frameshift from codon 730	co	French Canadian		[Bapat et al., 1999]	Y	1
1797	Ex 19	ins AAAC nt 2198	frameshift, codon 730 > truncation codon 733		USA		[Risinger et al., 1996]	Y	1
DF2913	Ex 19	ins AAAC nt 2198	frameshift, codon 730 > truncation codon 733	co/HNPCC	USA		[Syngal et al., 1999]	N	1
IMS1	Ex 19	del C nt 2224	frameshift from codon 742		Australia		[KohonenCorish et al., 1996]	Y	1
h112	Ex 19	C-A nt 2250	Tyr750STOP		England		Bunyan, unpub.	Y	1
DF171	Ex 19	C-G nt 2250	Tyr750STOP	co/HNPCC	USA		[Syngal et al., 1999]	N	1
SNU-H1009	Ex 19	del AA nt 2251	frameshift from codon 751	co/HNPCC	Korea		[Han et al., 1996]	Y	1
h18	Ex 19	del AA nt 2251	frameshift from codon 751		England		Bunyan, unpub.	Y	1
	Ex 19	A-G nt 2253	Lys751Arg		Scotland		Farrington/Dunlop, unpub.	N	3/4

Family 21	Ex 19	A-G nt 2253	Lys751Arg	co	Australia		[Scott et al., 2001]	N	3/4
17	Ex 19	ins AA nt 1853	frameshift	co	Germany/Czech Republic		[Pistorius et al., 2000]	N	1
FF74	Ex 19	A-T nt 2263	Arg755Trp	co/br	France		[Wang et al., 1999]	N	2
DF232	Ex 19	G-T nt 2265	Arg755TSer	co/HNPCC	USA		[Syngal et al., 1999]	N	2
DG	Ex 19	ins TGTT nt 2268	frameshift from codon 756	co/HNPCC	USA		[Liu et al., 1996]	Y	1
L2516	Ex 19	ins TGTT nt 2270	frameshift from codon 756	co/HNPCC	USA		[Papadopoulos et al., 1994]	Y	1
A-AMD7	Ex 19	ins T nt 2270	STOP757Leu		Italy		[Viel et al., 1997]	Y	1
A-MD1	3' UTR	del ttc nt 2268+33			Italy	0.04	[Viel et al., 1997]	Y	4
166	3' UTR	del ttc nt 2268+33			USA		Weber & RodriguezBigas, unpub.	Y	4
GDLG-21#III2, GDLG-20#II1*	3' UTR	del ttc nt 2268+33		co	Italy	0.02	[Curia et al., 1999]	N	4
VS0005	3' UTR	del ttc nt 2268+33			Switzerland		Hutter, unpub.	Y	4

(b) Germline hMSH2 Alterations

ID	EXON	Nucleotide Change	cDNA change	Cancer	Origin	Frequency	Reference	ICG	M/P
MD1157	Ex 1	A-T nt 1	Met1Leu	co	Scotland		[Farrington et al., 1998]	Y	2/3
344, 350	Ex 1	C-T nt 23	Thr8Met	co	Japan	0.01	[Nomura et al., 2000]	N	2/3
Case 17, control	Ex 1	C-T nt 126	Phe42Phe		USA		[Samowitz et al., 2001]	N	4
DP5	Ex 1	del 29bp nt 134	frameshift codon 45	co/ut	Canada		[Millar et al., 1999]	N	1

33	Ex 1	C-G nt 138	His46Gln	co	Scotland		[Bubb et al., 1996]	N	2
	Ex 1	del C nt 161	frameshift from codon 54 – STOP at codon 63				Deffenbaugh, unpub.	Y	1
8001	Ex 1	C-T nt 181	Gln61STOP		Uruguay		Sarroca et al., unpub.	Y	1
5	Ex 1	del G nt 201	frameshift from codon 67	co/en	Sweden		[Wahlberg et al., 1997]	Y	1
38439	Ex 1	ins gg track ggggg nt 205	frameshift		UK		Norbury, unpub.	Y	1
NLB-50162	Ex 1	2.1kb genomic deletion	deletion Ex 1 codon 1-70	co/HNPCC	Netherlands		[Wijnen et al., 1998b]	Y	1
621	Ex 1		deletion Ex 1 codon 1-70				[Papadopoulos et al., 1994]	N	1
NL-48, NL-53	Ex 1-6	2.5kb genomic deletion	deletion exon 1-6		Netherlands		[Wijnen et al., 1998b]	Y	1
NLB-376	IVS 1	a-g nt 212-2	splicing defect	co/HNPCC	Netherlands		[Wijnen et al., 1997]	N	1
1	IVS 1	g-c nt 211+8			Germany		[Holinski-Feder et al., 2001]	N	4
	IVS 1	c-g nt 211+9			Scotland	0.38	[Bubb et al., 1996]	Y	4
	IVS 1	c-g nt 211+9			Scotland/USA	0.148	[Farrington et al., 1998]	N	4
3 Kindreds	IVS 1	c-g nt 211+9			Switzerland		[Heinimann et al., 1999]	N	4
2	IVS 1	c-g nt 211+9			Germany		[Holinski-Feder et al., 2001]	N	4
cases & controls	IVS 1	c-g nt 211+9			USA		[Samowitz et al., 2001]	N	4
	IVS 1	9th base pair			USA	0.12	[Weber et al., 1997]	N	4
	Ex 2	G-A nt 219	Lys73Lys		Scotland/USA	0.007	[Farrington et al., 1998]	Y	4

352	Ex 2	del CT nt 223-224	frameshift	co	Japan		[Nomura et al., 2000]	N	1
NLB-600	Ex 2	del AG nt 227	frameshift from codon 75	co/HNPCC	Netherlands		[Wijnen et al., 1997].	Y	1
1820	Ex 2	del TT nt 261-262	frameshift		Macedonia		Heinimann, unpub.	Y	1
I-334	Ex 2	del TT nt 278	frameshift from codon 93	co/HNPCC	Italy		[Calistri et al., 2000]	Y	1
82	Ex 2	del TCT nt 279-281	frameshift from codon 93	co	Sweden		[Wahlberg et al., 1999]	Y	1
	Ex 2	G-A nt 287	Arg96His		Netherlands	0.01	[Wijnen et al., 1995]	Y	3
130	Ex 2	ins 22bp nt 289	frameshift from codon 97	MT	Germany		[Kruse et al., 1998]	Y	1
3	Ex 2	ins 22bp nt 289	frameshift from codon 97		Germany		[Holinski-Feder et al., 2001]	N	1
4	Ex 2	del AGTTGA nt297?	codon 99/100 deleted?		Germany		[Holinski-Feder et al., 2001]	N	2
523	Ex 2	G-C nt 319	Ala107Pro		Czech Republic		Krepelova, unpub.	Y	2/3
	Ex 2	A-G nt 329	Lys110Arg		USA	0.02	[Weber et al., 1997]	Y	3
	Ex 2	G-A nt 339	Lys113Lys		Czech Republic	0.0076	Krepelova, unpub.	Y	4
5	Ex 2	G-A nt 339	Lys113Lys		Germany		[Holinski-Feder et al., 2001]	N	4
6	Ex 2	del A nt 344	frameshift from codon 115		Germany		[Holinski-Feder et al., 2001]	N	1
NA-17/NA-86	Ex 2	5.4kb genomic deletion	deletion Ex 2 codon 71-122	co/HNPCC	Netherlands		[Wijnen et al., 1998b]	Y	1
HNP-O-3	Ex 2-8	genomic deletion	frameshift del exons 2-8 codons 71-462	co/ut/ga/urt	UK		[Bapat et al., 1999]	N	1
	IVS 2	del T from polyT tract			USA	0.05	[Weber et al., 1997]	Y	4
114-1-OL	Ex3	A-G nt 380	Asn127Ser		Nigeria		de la Chapelle, unpub.	Y	2/3

Case 5*,18,19	Ex3	A-G nt 380	Asn127Ser	co/ut	USA		[Samowitz et al., 2001]	N	2/3
MTS-K10(n)	Ex 3	del A nt 380	frameshift from codon 127	MT/pr	Germany		[Kruse et al., 1998]	Y	1
1097He	Ex 3	del TC nt 387	frameshift from codon 129	co/TS	Switzerland		[Buerstedde et al., 1995]	Y	1
	Ex 3	del nt 388-389	frameshift ->STOP codon 131	co/HNPCC	Switzerland		[Heinimann et al., 1999]	N	1
	Ex 3	C-T nt 399	Asp133Asp		African Am		Johnson, unpub.	Y	4
control	Ex 3	C-T nt 399	Asp133Asp		USA		[Samowitz et al., 2001]	N	4
HNP-1/ K-8/HNP10	Ex 3	del T nt 406	frameshift from codon 136	co/ut/urt/ov	Japan		[Miyaki et al., 1995]	Y	1
69	Ex 3	A-G nt 416	Asn139Ser	co	Scotland		[Bubb et al., 1996]	N	2/3
338	Ex 3	T-G nt 435	Ile145Met		Czech Republic		Krepelova, unpub.	Y	3/4
	Ex 3	T-G nt 435	Ile145Met				Deffenbaugh, unpub.	Y	3/4
	Ex 3	G-A nt 446	Gly149Asp				Deffenbaugh, unpub.	Y	2/3
	Ex 3	C-T nt 459	Ser153Ser		USA		[Moslein et al., 1996]	Y	4
175	Ex 3	C-A nt 471	Gly157Gly	co	Japan	0.028	[Nomura et al., 2000]	N	4
	Ex 3	C-A nt 471	Gly157Gly	co	Japan	0.02	[Konishi et al., 1996]	N	4
10	Ex 3	T-A nt 482	Val161Asp		Germany		[Muller-Koch et al., 2001]	N	1/2
4F	Ex 3	G-C nt 499	Asp167His	co	USA		[Moslein et al., 1996]	N	2
control	Ex 3	G-C nt 499	Asp167His		USA		[Samowitz et al., 2001]	N	2
	Ex 3	A-G nt 505	Ile168Val		Asia		Deffenbaugh, unpub.	Y	3/4
K-79/HNP185	Ex 3	del ATAC codon 169-170	frameshift from codon 169		Japan		[Bai et al., 1999]	N	1
5	Ex 3	del G nt 513	frameshift from codon 171	co	Hong Kong		[Chan et al., 1999]	N	1

2	Ex 3	del T nt 518	frameshift from codon 173	co	Germany/Czech Republic		[Pistorius et al., 2000]	N	1
24/223	Ex 3	del T nt 518	frameshift from codon 173	co	Germany		[Wehner et al., 1997]	Y	1
7	Ex 3	del T nt 518	frameshift from codon 173	co	Germany		[Holinski-Feder et al., 2001]	N	1
8064	Ex 3	del AA nt 530	frameshift		Uruguay		Sarroca, unpub.	Y	1
4	Ex 3	C-T nt 547	Gln183STOP	co	Portugal		[Fidalgo et al., 2000]	N	1
control	Ex 3	A-G nt 557	Asn186Ser		USA		[Samowitz et al., 2001]	N	3/4
HNPCC-8	Ex 3	del TGAGGCTCT nt 561	in frame del codon 187-190	co	Slovenia		[Ravnik-Glavac et al., 1998]	Y	1/2
16	Ex 3	TC-CT nt 569/570	Leu190Pro	co	Germany/Czech Republic		[Pistorius et al., 2000]	N	2/3
8	Ex 3	CT-TC nt 569/570		co	Germany		[Holinski-Feder et al., 2001]	N	2/3
	Ex 3	C-T nt573	Leu191Leu		Scotland/USA	0.021	[Farrington et al., 1998]	N	4
Case 20	Ex 3	C-T nt 573	Leu191Leu		USA		[Samowitz et al., 2001]	N	4
199	Ex 3	A-G nt 593	Glu198Gly	co	Sweden		[Wahlberg et al., 1999]	Y	2
D/19	Ex 3	T-C nt 595	Cys199Arg	gl/co/re	Hong Kong		[Leung et al., 1998]	Y	2
-	Ex 3	G-T nt 613	Glu205STOP	co	Poland		Kurzawski, unpub.	Y	1
HNP-055	Ex 3	C-T nt 643	Gln215STOP	co	France		[Bapat et al., 1999]	Y	1
11	Ex 3(*4?*)	A-G nt 646	Ile216Val/possible splice defect		Germany		[Muller-Koch et al., 2001]	N	2/3
3 kindreds	Ex 3	2.2kb genomic deletion	Ex 3 deleted	co/HNPCC	Netherlands		[Wijnen et al., 1998b]	Y	1
CO07	Ex 3		in frame del Ex 3 codon 123-215	co	England		[Froggatt et al., 1996]	N	1

HNP-048	Ex 3		in frame del Ex 3 codon 123-215	co/ga/ut/sk	UK	[Bapat et al., 1999]	N	1
9	Ex 4	del GGTCC, ins TAAT nt 675?	frameshift?		Germany	[Holinski-Feder et al., 2001]	N	1/2
HNP4/H114/K-43	Ex 4	ins A nt 688	frameshift from codon 228/230	co/ut/ov	Japan	[Miyaki et al., 1995]	Y	1
FF51	Ex 4		frameshift from codon 228	co/ov/urt	France	[Wang et al., 1999]	N	1
3	Ex 4	del TT nt 696	frameshift	co	Germany/Czech Republic	[Pistorius et al., 2000]	N	1
10	Ex 4	del TT nt 696	frameshift	co	Germany	[Holinski-Feder et al., 2001]	N	1
DF268, DF951	Ex 4	del AA nt 704-705	frameshift --> Truncation, codon 235	co	USA	[Syngal et al., 1999]	N	1
MD818	Ex 4	C-T nt 754	Gln252STOP	co/ut	Scotland	[Liu et al., 1995b]	Y	1
MD968	Ex 4	C-T nt 754	Gln252STOP	co/ut	Scotland	Farrington/Dunlop, unpub.	N	1
3	Ex 4	G-T	STOP	co	Poland	[Debniak et al., 2000]	N	1
FF17	Ex 4		in-frame del exon 4	co/ut	France	[Wang et al., 1999]	N	1
46	IVS 4	g-a nt 792+1	splice defect	co	USA	[Cunningham et al., 2001]	N	1
Case 21	IVS 4	a-g nt792+49			USA	[Samowitz et al., 2001]	N	4
MD1521	IVS 4	g-t nt 793-1	splice defect	co	Scotland	Farrington/Dunlop, unpub.	Y	1/2
	Ex 5	C-T nt 806	Ser 269Leu		Latin American / Caribbean	Deffenbaugh, unpub.	Y	2/3
MD825	Ex 5	del GTCT nt 808	in frame del Exon 5	co/ut/pa/st	Scotland	[Liu et al., 1995b]	Y	1
DF597	Ex 5	C-T nt 815	Ala272Val	co	USA	[Syngal et al., 1999]	N	3

Case 22	Ex 5	C-T nt 815	Ala272Val		USA		[Samowitz et al., 2001]	N	3
GALI	Ex 5	ins T nt 840	frameshift from codon 280	co/ut/bl/b	France		[Lazar et al., 1994]	Y	1
BLA-SG101	Ex 5	ins T nt 840	frameshift from codon 280		France		[Dieumegard et al., 2000]	Y	1
NL-10, NL-39	Ex 5	C-T nt 862	Gln288STOP	co/st/k/urt	Netherlands		[Wijnen et al., 1998b]	Y	1
1	Ex 5	C-T nt 862	Gln288STOP	co/ut/ov/pa	Germany/Czech Republic		[Pistorius et al., 2000]	N	1
278(n)	Ex 5	C-T nt 862	Gln288STOP	MT/Hodgkins	Germany		[Kruse et al., 1998]	Y	1
11	Ex 5	C-T nt 862	Gln288STOP	co	Germany		[Holinski-Feder et al., 2001]	N	1
6	Ex 5	C-T codon 298 (*?*)		co	Sweden		[Wahlberg et al., 1999]	Y	1
NL-38	Ex 5	G-A nt 913	Ala305Thr	co/HNPCC	Netherlands	0/100	[Wijnen et al., 1997]	Y	2/3
354	Ex 5	G-A nt 942	Splice mutation	co	Japan		[Nomura et al., 2000]	N	1
	Ex 5	Ex 5 deleted	deletion Ex 5 cod 265-314	co			[Parsons et al., 1995]	N	1
FF8	Ex 5	Ex 5 deleted	deletion Ex 5 cod 265-314	co/ut/leu	France		[Wang et al., 1997]	N	1
FF27	Ex 5	Ex 5 deleted	deletion Ex 5 cod 265-314	co/si/leu/br/l/chg	France		[Wang et al., 1999]	N	1
11,42	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co/ut/ov/urt/bo	Denmark		[Jager et al., 1997]	Y	1/2
119	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co	Germany		[Lamberti et al., 1999]	Y	1/2
20681/2A	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co/HNPCC	USA		[Moslein et al., 1996]	Y	1/2
3177	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co/HNPCC			[Liu et al., 1996]	Y	1/2
-	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co	UK		[Froggatt et al., 1995]	Y	1/2
HNP7	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co/ut/b/sk	Japan		[Miyaki et al., 1995]	Y	1/2

667,349	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co/ut/pa/b	Italy		[Pensotti et al., 1997]	Y	1/2
C, MF, RB	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co	Canada		[Liu et al., 1994]	Y	1/2
C017,C022	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co	UK		[Froggatt et al., 1996]	Y	1/2
N-HS3, O-4, HNP-020, HNP-038	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co/ut/br/ov/bs/ ga/urt/ sk/th/l/bo	Norway, UK, Newfoundland		[Bapat et al., 1999] & unpub.	Y	1/2
124	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co			unpub.	Y	1/2
TK1882	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co	Poland		Kurzawski. unpub.	Y	1/2
K-39/HNP108	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co	Japan		[Lu et al., 1996]	N	1/2
23	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co	Hong Kong		[Chan et al., 1999]	N	1/2
	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co/HNPCC	Italy		[de Leon et al., 1999]	N	1/2
SI505	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co	Italy		[Montera et al., 2000]	N	1/2
42	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co	Portugal		[Fidalgo et al., 2000]	N	1/2
3 Kindreds	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co	USA		[Syngal et al., 1999]	N	1/2
10 Kindreds	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co	Newfoundland		[Froggatt et al., 1999]	N	1/2
K-57/HNP143	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co	Japan		[Lu et al., 1996]	N	1/2
GDLM-2#III-1	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co	Italy		[Curia et al., 1999]	N	1/2
4	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co/re/lv	Germany/Czech Republic		[Pistorius et al., 2000]	N	1/2
3 kindreds	IVS 5	a-t nt 942+3	in-frame deletion Ex 5				Deffenbaugh, unpub.	Y	1/2
12	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co	Germany		[Holinski-Feder et al., 2001]	N	1/2
Case 6	IVS 5	a-t nt 942+3	in-frame deletion Ex 5	co	USA		[Samowitz et al., 2001]	N	1/2
	IVS 5	del 10bp from poly A			USA	0.032	[Weber et al., 1997]	Y	4

		tract							
6	IVS 5	g-a nt 943-1	in-frame deletion Ex 6	co/ut/ga	Germany/Czech Republic		[Pistorius et al., 2000]	N	1/2
DF3002	Ex 6	G-T nt 945	Gly315Val	co	USA		[Syngal et al., 1999]	N	2/3
SI524	Ex 6		Gly321Asp (*Gly322Asp*)	co	Italy		[Montera et al., 2000]	N	2/3
1	Ex 6	G-A nt 965	Gly322Asp	co/br	Russia		[Maliaka et al., 1996]	Y	2/3
JM	Ex 6	G-A nt 965	Gly322Asp	co	England		[Tomlinson et al., 1997]	Y	2/3
Family 24	Ex 6	G-A nt 965	Gly322Asp	co	Australia		[Scott et al., 2001]	N	2/3
	Ex 6	G-A nt 965	Gly322Asp	co	Finland	.067	[Salovaara et al., 2000]	N	2/3
DF1448, DF1370	Ex 6	G-A nt 965	Gly322Asp	co	USA		[Syngal et al., 1999]	N	2/3
HNPCC-1	Ex 6	G-A nt 965	Gly322Asp	co/extracolon	England		[Beck et al., 1997c]	N	2/3
CI-2556/SM-1820	Ex 6	G-A nt 965	Gly322Asp		Poland		Kurzawski, unpub.	Y	2/3
19	Ex 6	G-A nt 965	Gly322Asp	co	Portugal		[Fidalgo et al., 2000]	N	2/3
9 Kindreds	Ex 6	G-A nt 965	Gly322Asp	co	Sweden		[Liu et al., 1998b]	N	2/3
	Ex 6	G-A nt 965	Gly322Asp		Scotland / USA	0.02	[Liu et al., 1995a]	Y	2/3
case 23,24, control	Ex 6	G-A nt 965	Gly322Asp		USA		[Samowitz et al., 2001]	N	2/3
	Ex 6	G-A nt 965	Gly322Asp		Norway	0.02	[Borresen et al., 1995]	N	2/3
5 Kindreds	Ex 6	G-A nt 965	Gly322Asp		Czech Republic		Krepelova, unpub.	Y	2/3
	Ex 6	G-A nt 965	Gly322Asp		Spain		Caldes, unpub.	Y	2/3
	Ex 6	G-A nt 965	Gly322Asp		Germany	0.01	[Wehner et al., 1997]	N	2/3

GDLM-9#II-2	Ex 6	G-A nt 965	Gly322Asp	co	Italy	0/100	[Curia et al., 1999]	N	2/3
13	Ex 6	G-A nt 965	Gly322Asp	co	Germany	0.01	[Holinski-Feder et al., 2001]	N	2/3
186	Ex 6	CGC-GAC	Missense		Germany		[Brieger et al., 1999]	Y	2/3
K-55/HNP137	Ex 6	C-T nt 968	Ser323Cys	co	Japan		[Akiyama et al., 1996]	N	2/3
331*	Ex 6	G-A nt 972	Gln324Gln	co	Japan	0.00	[Nomura et al., 2000]	N	4
Case 18	Ex 6	G-C nt 982	Ala328Pro	co	USA		[Samowitz et al., 2001]	N	2/3
GDLM-8#II-1, GDLV-35#III-5	Ex 6	C-T nt 984	Ala328Ala	co	Italy	0/100	[Curia et al., 1999]	Y	4
	Ex 6	C-T nt 984	Ala328Ala		Sweden		[Wahlberg et al., 1999]	Y	4
Family 25	Ex 6	C-T nt 984	Ala328Ala	co	Australia		[Scott et al., 2001]	N	4
Case 25, control	Ex 6	C-T nt 984	Ala328Ala		USA		[Samowitz et al., 2001]	N	4
114-1-OL*	Ex 6	T-C nt 997	Cys333Arg		Nigeria		de la Chapelle, unpub.	Y	2/3
IMS5	Ex 6	G-A nt 998	Cys333Tyr		Australia		Bennett, unpub.	Y	2/3
59	Ex 6	A-T nt 1000	Lys334STOP	co/urt/bl	USA - German		[Weber et al., 1997]	Y	1
	Ex 6	C-T nt 1006	Pro336Ser	co	Switzerland		[Heinimann et al., 1999]	N	2
Family 23	Ex 6	C-T nt 1009	Gln337STOP	co	Australia		[Scott et al., 2001]	N	1
	Ex 6	G-A nt 1012	Gly338Arg				Deffenbaugh et al., unpub	Y	2/3
NL-21	Ex 6	del AA nt 1016	frameshift from codon 339	co/pr/en/ov/urt	Netherlands		[Wijnen et al., 1995]	Y	1
GDLG-49#IV-2	Ex 6	G-A nt 1024	Val342Ile	co	Italy	0/100	[Curia et al., 1999]	N	3/4
h86	Ex 6	del G nt 1059	frameshift from codon 353		England		Bunyan, unpub.	Y	1

RH	Ex 6	173bp ins nt 1076-1092	frameshift from codon 359	co/HNPCC	USA		[Liu et al., 1996]	Y	1
	Ex 6	G-A, polyA region	-		USA	0.036	[Weber et al., 1997]	N	4
NL-33/NLB-50490	Ex 6	13kb genomic deletion	deletion Ex 6 codon 315-359	co/HNPCC	Netherlands		[Wijnen et al., 1998b]	Y	
OZ-5	Ex 2-6	Ex 2-6 deleted	deletion Ex 2-6 codon 71-359	co/en/b	Australia		[Kohonen-Corish et al., 1996]	N	1
FF15	IVS 6	g-a nt 1076+1	Splice defect	co/bl/ut/ga	France		[Wang et al., 1999]	N	1
14	IVS 6	g-a nt 1076-35*(+35)*			Germany		[Holinski-Feder et al., 2001]	N	4
	IVS 6	t-c nt 1077-10			Norway	0.05	[Borresen et al., 1995]	Y	3/4
1 kindred	IVS 6	t-c nt 1077-10			Scotland		[Farrington et al., 1998]	N	3/4
15	IVS 6	t-c nt 1076(*1077*)-10			Germany		[Holinski-Feder et al., 2001]	N	3/4
cases & controls	IVS 6	t-c nt 1076–8			USA		[Samowitz et al., 2001]	N	3/4
30	Ex 7	ins A nt 1097	frameshift from codon 366, termination codon 388	co/en	Sweden		[Liu et al., 1998a]	Y	1
H23	Ex 7	ins A nt 1097	frameshift from codon 366, termination codon 388	re/ut/ga	Sweden		[Planck et al., 1999]	N	1
2	Ex 7	ins TA nt 1127	frameshift from codon 376	co/en/br/sk	Russia		[Maliaka et al., 1996]	Y	1
NL-7	Ex 7	del T nt 1137	frameshift from codon 379	co/en/es/st	Netherlands		[Wijnen et al., 1995]	Y	1
He1587	Ex 7	C-T nt 1147	Arg383STOP	co/en	Switzerland		[Buerstedde et al., 1995]	Y	1
6	Ex 7	C-T nt 1147	Arg383STOP	co/re/ut/br	Germany/Czech Republic		[Pistorius et al., 2000]	N	1
177	Ex 7	C-T nt 1147	Arg383STOP	co	Germany		[Lamberti et al., 1999]	N	1

	Ex 7	C-T nt 1147	Arg383STOP	co/HNPCC	Switzerland		[Heinimann et al., 1999]	N	1
	Ex 7	C-T nt 1165	Arg389STOP		French Canada		Foulkes, unpub.	Y	1
HNPCC-2	Ex 7	C-T nt 1165	Arg389STOP	co/extracolon	England		[Beck et al., 1997c]	N	1
DP7	Ex 7	C-T nt 1165	Arg389STOP	co/ ut	Canada		[Millar et al., 1999]	N	1
201	Ex 7	C-T nt 1165	Arg389STOP		Germany		[Brieger et al., 1999]	Y	1
449	Ex 7	C-T nt 1168	Leu390Phe	co	USA - African		[Weber et al., 1999]	N	2/3
	Ex 7	C-T nt 1168	Leu390Phe		Japan	0.02	[Konishi et al., 1996]	N	2/3
MD1466	Ex 7	A-T nt 1177	Lys393STOP	co	Scotland		Farrington/Dunlop, unpub.	N	1
C021	Ex 7	C-T nt 1189	Gln397STOP		UK		[Froggatt et al., 1996]	Y	1
CA135	Ex 7	ins CA	frameshift from codon 399	co	Italy		[Montera et al., 2000]	N	1
MD528	Ex 7	C-T nt 1216	Arg406STOP	co	Scotland		[Liu et al., 1995b]	Y	1
8	Ex 7	C-T nt 1216	Arg406STOP	co/HNPCC	USA		[Leach et al., 1993]	Y	1
7	Ex 7	C-T nt 1216	Arg406STOP	co	Sweden		[Wahlberg et al., 1999]	Y	1
24	Ex 7	C-T nt 1216	Arg406STOP	co/ut/ga/ph	Germany/Czech Republic		[Pistorius et al., 2000]	N	1
240-1	Ex 7	C-T nt 1216	Arg406STOP	co/re/ut	USA (Italian)		[Weber et al., 1997]	Y	1
IG-1296/4	Ex 7	C-T nt 1216	Arg406STOP		Poland		[Debniak et al., 2000]	Y	1
17	Ex 7	ins T nt 1222	frameshift from codon 408		Germany		[Holinski-Feder et al., 2001]	N	1
59	Ex 7	del AG nt 1226	frameshift from codon 409		Sweden		[Liu et al., 1998a]	Y	1
330 or 14A	Ex 7	del AG nt 1226	frameshift from codon 409	co	Germany		[Moslein et al., 1996]	Y	1
225	Ex 7	del AG nt 1226	frameshift from codon 409	co	Germany		[Lamberti et al., 1999]	N	1

HNPCC-MO-2	Ex 7	del CCTA nt 1243	frameshift from codon 414		Italy		[de Leon et al., 1999]	Y	1
A-MD1	Ex 7	del CCTA nt 1243	frameshift from codon 414	co/ga/ut/b	Italy		[Viel et al., 1997]	Y	1
BR120	Ex 7	del G nt 1249	frameshift		Brazil		[Cassandra M.Corvello, 1999]	Y	1
2/H62	Ex 7	C-T nt 1255	Gln419STOP	co/en/ga	Japan		[Miyaki et al., 1995]	Y	1
	Ex 7	C-A	Gln419Lys		Japan	0.01	[Konishi et al., 1996]	N	2/3
RV	Ex 7		out of frame del codon 359-426	co	USA		[Liu et al., 1996]	N	1
NL-5	Ex 7	6 kb genomic deletion	genomic deletion		Netherlands		Wijnen, unpub.	Y	1
167	Ex 7-10		del exon 7-10	co	Sweden		[Wahlberg et al., 1999]	Y	1
3	IVS 7	g-c nt 1276+1	splice defect	co/en/br	Moldavia		[Maliaka et al., 1996]	Y	1
	IVS 7	c-a nt 1276+49			African Am		Johnson, unpub.	Y	4
NL-23/NL-220	Ex 8	C-T nt 1285	Gln429STOP	co/st/pr/cx/sk/b/ HNPCC	Netherlands		[Wijnen et al., 1995]	Y	1
90	Ex 8	del CT nt 1316-1317	frameshift from codon 439, STOP codon 441	co	USA		[Cunningham et al., 2001]	N	1
O3	Ex 8	T-C nt 1319	Leu440Pro		Lithuania		Avizienyte, unpub.	Y	2/3
MD1699	Ex 8	ins A nt 1320	frameshift from codon 440	co	Scotland		Farrington/Dunlop, unpub.	N	1
	Ex 8	T-G nt 1339	Phe447Val		Netherlands	0/100	[Wijnen et al., 1998a]	N	3/4
DF1754	Ex 8	del AG nt 1352-1353	frameshift - truncation codon 451	co	USA		[Syngal et al., 1999]	N	1
HNP-043	Ex 8	del 32bp at 1354	in frame del codon 452-462		Italy		[Bapat et al., 1999]	Y	1
DP1	Ex 8	A-T nt 1358	creates novel splice site	co/ut	Canada		[Millar et al., 1999]	N	1

3106	Ex 8	T-G nt 1373	Leu458STOP	co/MT	USA	[Liu et al., 1994]	Y	1
28	Ex 8	T-G nt 1373	Leu458STOP	co	Sweden	[Wahlberg et al., 1997]	Y	1
Family 26	Ex 8	complex inversion and substitution	nonsense segment	co	Australia	[Scott et al., 2001]	N	1
HNP-049	Ex 8	Ex 8 deleted	deletion Ex 8 cod 426-462		Newfoundland	[Bapat et al., 1999]	N	1
HNP-047	Ex 8	Ex 8 deleted	deletion Ex 8 cod 426-462		Italy	[Bapat et al., 1999]	N	1
19	IVS 8	g-a nt 1386+1			Germany	[Holinski-Feder et al., 2001]	N	1
C54	IVS 8	g-t nt 1387-1	splice defect	co	Finland	[Wu et al., 1997]	Y	1
52	IVS 8	g-t nt 1387-1	splice defect	co	Finland	[Aaltonen et al., 1998]	N	1
Case 26	IVS 8	g-t nt 1387–8		co	USA	[Samowitz et al., 2001]	N	3/4
Family 27	Ex 9	del G nt 1408	frameshift and STOP codon 470	co	Australia	[Scott et al., 2001]	N	1
20	Ex 9	del G nt 1408	frameshift and STOP codon 470		Germany	[Holinski-Feder et al., 2001]	N	1
CFS137	Ex 9	C-G nt 1418	Ser473STOP	co/es	Italy	[Fornasarig et al., 2000]	N	1
43	Ex 9	A-T nt 1435	Ser479Cys	co	Italy	[Calistri et al., 2000]	N	2/3
I-219	Ex 9	ins A nt 1444	frameshift from codon 481	co/en/st/l	Italy	[Wijnen et al., 1995]	Y	1
27,18	Ex 9	del AATG nt 1455	frameshift from codon 486	co	Hong Kong	[Chan et al., 1999]	N	1
21	Ex 9	C-T nt 1477	Gln493Stop		USA	de la Chapelle, unpub.	Y	1
R-RMS15	Ex 9	del A nt 1497	frameshift from codon 499 – truncation	co/ut/leu	Italy	[Genuardi et al., 1998]	N	1
386	Ex 9	ins C nt 1501	frameshift from codon 501		Czech Republic	Krepelova, unpub.	Y	1
Family 28	Ex 9	G-C nt 1510	splice defect	co	Australia	[Scott et al., 2001]	N	1

1515*	IVS 9	g-t nt 1511-10		co/HNPCC	Switzerland		[Buerstedde et al., 1995]	Y	3/4
	IVS 9	t-a nt 1511-9			Norway	0.2	[Borresen et al., 1995]	Y	3/4
	IVS 9	t-a nt 1511-9			Scotland/USA	0.852	[Farrington et al., 1998]	N	3/4
	IVS 9	t-a nt 1511-9			Spain		Caldes, unpub.	Y	3/4
21	IVS 9	t-a nt 1510*(1511)*-9			Germany		[Holinski-Feder et al., 2001]	N	3/4
cases & controls	IVS 9	a-t nt 1511–9			USA		[Samowitz et al., 2001]	N	**3/4**
NLB-172	IVS 9	a-g nt 1501(*1511*)-2	splice defect	co/HNPCC	Netherlands		Wijnen, unpub.	Y	1
SNU-YC13	Ex 10	G-T nt 1516	Asp506Tyr	co	Korea		[Han et al., 1996]	Y	2
38	Ex 10	del CA nt 1550	frameshift from codon 517	co/HNPCC	Finland		[Nystrom-Lahti et al., 1996]	Y	1
80	Ex 10	del CA nt 1550	frameshift from codon 517	co	Sweden		[Liu et al., 1998a]	Y	1
19*	Ex 10	C-T nt 1552	Gln518STOP	co	Portugal		[Fidalgo et al., 2000]	N	1
	Ex 10	T-C nt 1563	Tyr521Tyr		Czech Republic		Krepelova, unpub.	Y	4
1846	Ex 10	del A nt 1575	frameshift from codon 525		Switzerland/Ital		Heinimann, unpub.	Y	1
MTS-K2(n)	Ex 10	del A nt 1576	frameshift from codon 525	MT/en	Germany		[Kruse et al., 1998]	Y	1
h1	Ex 10	del C nt 1577	frameshift from codon 525		England		Bunyan, unpub.	Y	1
Case 1/H23/H8	Ex 10	del A, codon 529	frameshift --> termination codon 542	co	Sweden		[Nilbert et al., 1999]	N	1
NL-13	Ex 10	ins G nt 1595	frameshift from codon 531	co/sk/ov/st/cx	Netherlands		[Wijnen et al., 1995]	Y	1
R-RM5	Ex 10	ins GA	truncation, codon 546-547	co	Italy		[Genuardi et al., 1998]	N	1
F719	IVS10	a-g nt 1661+10			Italy		[Montera et al., 2000]	N	4

5 kindreds	IVS 10	a-g nt 1661+10		co/HNPCC	Switzerland		[Buerstedde et al., 1995]	N	4
Family 29	IVS 10	a-g, 12 from exon 10 (*g-a nt 1661+12*)		co	Australia		[Scott et al., 2001]	N	4
	IVS 10	a-g nt 1661+12			Scotland/USA	0.62	[Farrington et al., 1998]	N	4
	IVS 10	g-a nt 1661+12			Netherlands	0.25	[Wijnen et al., 1994]	Y	4
	IVS 10	g-a nt 1661+12			Sweden	0.47	[Wahlberg et al., 1997]	Y	4
	IVS 10	g-a nt 1661+12			Sweden		[Wahlberg et al., 1999]	Y	4
	IVS 10	g-a nt 1661+12			Spain		Caldes, unpub.	Y	4
22	IVS 10	g-a nt 1661+12			Germany		[Holinski-Feder et al., 2001]	N	4
cases & controls	IVS 10	g-a nt 1661+12			USA		[Samowitz et al., 2001]	N	4
1 Kindred	IVS 10	a-t nt 1662-25			Scotland		Farrington/Dunlop, unpub.	N	4
-	IVS 10	t-a nt 1662-9			Scotland/USA	0.07	[Farrington et al., 1998]	N	3/4
5 Kindreds	IVS 10	t-a nt 1662-9			Switzerland		[Heinimann et al., 1999]	N	3/4
3 Kindreds	IVS 10	t-a nt 1662-9			USA		[Cunningham et al., 2001]	N	3/4
12	Ex 11*(10)?*	A-C *(nt 1660)?*	Ser554Arg or possible splice defect		Germany		[Muller-Koch et al., 2001]	N	1/2
	Ex 11	T-C nt 1666	Leu556Leu		Netherlands	0.01	[Wijnen et al., 1995]	Y	4
	Ex 11	T-C nt 1666	Leu556Leu		Germany	0.01	[Wehner et al., 1997]	Y	4
Case 27	Ex 11	T-C nt 1666	Leu556Leu	co	USA		[Samowitz et al., 2001]	N	4
Family 30	Ex 11	T-C nt 1669 (*1666*)	Leu557 (*556*) Leu		Australia		[Scott et al., 2001]	N	4

	Ex 11	del T nt 1674	frameshift – STOP codon 559		Europe		Deffenbaugh, unpub.	Y	1
132	Ex 11	del T nt 1677	frameshift from codon 559	MT/ga/br	Germany		[Kruse et al., 1998]	Y	1
27	Ex 11	del A nt 1683	frameshift from codon 561	co	Germany		[Wehner et al., 1997]	Y	1
23	Ex 11	del A nt 1683	frameshift from codon 561	co	Germany		[Holinski-Feder et al., 2001]	N	1
S11	Ex 11	A-T nt 1685	Glu562Val	co	England		[Beck et al., 1997a]	N	2
NL-221	Ex 11	del AA nt 1696	frameshift from codon 565	co/HNPCC	Netherlands		[Wijnen et al., 1997]	Y	1
167	Ex 11	del AAACA nt 1699	frameshift from codon 566	MT/en	Germany		[Kruse et al., 1996]	Y	1
24	Ex 11	del AAACA nt 1699	frameshift from codon 566	co	Germany		[Holinski-Feder et al., 2001]	N	1
GC12845, GC12313	Ex 11	del AAACA nt 1699	frameshift from codon 566		UK		West, unpub.	Y	1
Case 7	Ex 11	del nt 1700-1704	frameshift from codon 567	co/ut/ly	USA		[Samowitz et al., 2001]	N	1
339	Ex 11	del GA nt 1705	frameshift from codon 568	co	Japan		[Nomura et al., 2000]	N	1
K-70/HNP168	Ex 11	del GA, codon 568-569	frameshift		Japan		[Bai et al., 1999]	N	1
GDLX-39#III-7	Ex 11	A-G nt 1737	Lys579Lys	co	Italy	0/100	[Curia et al., 1999]	N	4
	Ex 11	A-G nt 1737	Lys579Lys		Netherlands	0.01	[Wijnen et al., 1995]	Y	4
Case 28	Ex 11	A-G nt 1737	Lys579Lys	co	USA		[Samowitz et al., 2001]	N	4
1642	Ex 11	G-T nt 1738	Glu580STOP	Co/HNPCC	Switzerland		[Heinimann et al., 1999]	Y	1
B/4	Ex 11	G-T nt 1738	Glu580STOP	gl/co	Hong-Kong		[Leung et al., 1998;Chan et al., 1999]	Y	1
	Ex 11	T-C nt 1755	Ser585Ser		Finland	0.005	[Wahlberg et al., 1997]	Y	4
	IVS 11	g-a nt 1759+1	Splice defect		Latin American / Caribbean		Deffenbaugh, unpub.	Y	1

1807	IVS 11	t-c nt 1759 + 2	Splice defect		Switzerland		Heinimann, unpub.	Y	1
C/2	Ex 12	del G nt 1760	frameshift from codon 588	gl/co	Hong-Kong		[Leung et al., 1998;Chan et al., 1999]	Y	1
MD1325	Ex 12	93 bp deletion nt 1783-1875	in frame del codon 595-625	co	Scotland		Farrington/Dunlop, unpub.	N	1/2
MD666	Ex 12	del AAT nt 1786	in frame del Asn codon 596	co/ut	Scotland		[Liu et al., 1996]	Y	1
MD1332,MD1370	Ex 12	del AAT nt 1786	in frame del Asn codon 596	co/ut	Scotland		Farrington/Dunlop, unpub.	N	1
1898 or 1A	Ex 12	del AAT nt 1786	in frame del Asn codon 596	re	USA		[Moslein et al., 1996]	Y	1
1383*	Ex 12	del AAT nt 1786	in frame del Asn codon 596	co/HNPCC	Switzerland		[Buerstedde et al., 1995]	Y	1
	Ex 12	del AAT nt 1786	in frame del Asn codon 596	co/HNPCC	Switzerland		[Heinimann et al., 1999]		1
	Ex 12	del AAT nt 1786	in frame del Asn codon 596	co/re	Switzerland		[Mary et al., 1994]	Y	1
1342	Ex 12	del AAT nt 1786	in frame del Asn codon 596	co/pa	Norway		[Borresen et al., 1995]	Y	1
N-534, N-554	Ex 12	del AAT nt 1786	in frame del Asn codon 596	co/HNPCC	Norway		[Wijnen et al., 1997]	Y	1
FF50	Ex 12	del AAT nt 1786	in frame del Asn codon 596	co/ut	France		[Wang et al., 1999]	N	1
HNPCC-MO-29	Ex 12		Missense codon 596		Italy		Ponz de Leon, unpub.	Y	2/3
R-MD4	Ex 12	A-G nt 1787	Asn596Ser		Italian	<0.004	[Viel et al., 1997]	Y	2/3
GC9160	Ex 12	C-T nt 1801	Gln601STOP		UK		West, unpub.	Y	1
MD814	Ex 12	C-T nt 1801	Gln601STOP	co/ut/MT	Scotland		[Liu et al., 1995b]	Y	1
1	Ex 12	C-T nt 1801	Gln601STOP	MT	UK		[Kruse et al., 1996]	Y	1
138	Ex 12	G-A nt 1808	Asp603Asn	co	Finland		[Salovaara et al., 2000]	N	3/4

133	Ex 12	del T nt 1809	frameshift from codon 602	co/MT/pr	Germany		[Kruse et al., 1996]	Y	1
25	Ex 12	del T nt 1809	frameshift from codon 602	co	Germany		[Holinski-Feder et al., 2001]	N	1
26	Ex 12	C-T nt 1826	Ala609Val		Germany		[Holinski-Feder et al., 2001]	N	3/4
	Ex 12	del T nt 1827	634STOP				Deffenbaugh, unpub.	Y	1
K-41/H110	Ex 12	T-G nt 1857	Tyr619STOP	co	Japan		[Lu et al., 1996]	N	1
25	Ex 12	ins TG nt 1860	frameshift from codon 619	co/HNPCC	Finland		[Nystrom-Lahti et al., 1996]	Y	1
110	Ex 12	C-T nt 1861	Arg621STOP	co/bl/urt	USA		[Weber et al., 1997]	Y	1
4	Ex 12	C-T nt 1861	Arg621STOP	co/en/st/ov	Russia		[Maliaka et al., 1996]	Y	1
J	Ex 12	C-T nt 1865	Pro622Leu		New Zealand		[Leach et al., 1993]	Y	2/3
-	Ex 12	G-C nt 1906	Ala636Pro		France		Bressac-de Pailerets, unpub.	Y	2/3
54	Ex 12	G-C nt 1906	Ala636Pro		USA		de la Chapelle, unpub.	Y	2/3
Cx10	Ex 12	C-T nt 1915	His639Tyr – creates splice site		USA		[Leach et al., 1993]	N	1/2
MD579	Ex 12	C-T nt 1915	del codons 588-820	co/ut/ov	Scotland		[Farrington et al., 1998]	Y	1
MD435	Ex 12	C-T nt 1915	del codons 588-820	co/ut/ov/br	Scotland		Farrington/Dunlop, unpub.	N	1
JV	Ex 12	C-T nt 1915	out of frame del codon 638-669	co	USA		[Liu et al., 1994]	Y	1
12*,4*,8*,11*, 15*,17*	Ex 12	A-G nt 1916	His639Arg		Japan		[Nakahara et al., 1997]	N	3
	Ex 12	T-G nt 1921	Cys641Gly		Scotland/USA	0.007	[Farrington et al., 1998]	N	3/4
4*,8*,15*	Ex 12	G-A nt 1939	Glu647Lys		Japan		[Nakahara et al., 1997]	N	2
OZ-6	Ex 12	C-G nt 1968	Try656STOP	co/ut	Australia		[Kohonen-Corish et al.,	Y	1

							1996]		
Family 31	Ex 12	G-C (*C-G*) nt 1968	stop codon	co	Australia		[Scott et al., 2001]	N	1
2	Ex 12	del AG nt 1985	frameshift from codon 661	co/MT/en/ov/ ur/bl	UK		[Kolodner et al., 1994]	Y	1
h110	Ex 12	del AG nt 1985	frameshift from codon 661		England		Bunyan, unpub.	Y	1
DP4	IVS 12	del 11bp nt 2005+2	splice defect	co/ut	Canada		[Millar et al., 1999]	N	2/3
	IVS 12	t-c nt 2006-6			Norway	0.14		N	3/4
	IVS 12	t-c nt 2006-6			USA		[Leach et al., 1993]	Y	3/4
	IVS 12	t-c nt 2006-6			Germany	0.06	[Wehner et al., 1997]	Y	3/4
	IVS 12	t-c nt 2006-6			Scotland/USA	0.063	[Farrington et al., 1998]	N	3/4
	IVS 12	t-c nt 2006-6			Switzerland		[Heinimann et al., 1999]	N	3/4
	IVS 12	t-g nt 2006-6			Sweden	0.23	[Wahlberg et al., 1997]	Y	3/4
27	IVS 12	t-c nt 2006-6			Germany		[Holinski-Feder et al., 2001]	N	3/4
cases & controls	IVS 12	t-g nt 2006-6			USA		[Samowitz et al., 2001]	N	3/4
	IVS 12	g-a, -6 *(-4*) upstream of exon 13			Sweden		[Wahlberg et al., 1999]	Y	3/4
1033*, 1121*	IVS 12	t-c –4 (-*6*) from exon 13 splice acceptor		co/HNPCC	Switzerland		[Buerstedde et al., 1995]	Y	2/3
MTS-K8(n)	Ex 13	del T nt 2015	frameshift from codon 671	MT/pa	Germany		[Kruse et al., 1998]	N	1
MD830	Ex 13	C-T nt 2038	Arg680STOP	co	Scotland		[Farrington et al., 1998]	Y	1
MD1440	Ex 13	C-T nt 2038	Arg680STOP	co	Scotland		Farrington/Dunlop, unpub.	N	1

N-414	Ex 13	C-T nt 2038	Arg680STOP	co/HNPCC	Norway		[Wijnen et al., 1997]	Y	1
	Ex 13	C-T nt 2038	Arg680STOP				Deffenbaugh, unpub.	Y	1
403	Ex 13	C-T nt 2038	Arg680STOP	co	USA		[Cunningham et al., 2001]	N	1
Case 29	Ex 13	G-A nt 2047	Gly683Arg	co	USA		[Samowitz et al., 2001]	N	2
Case 30	Ex 13	G-T nt 2048	Gly683Val	co	USA		[Samowitz et al., 2001]	N	2/3
Family 33	Ex 13	duplication 66 bp	reiteration	co	Australia		[Scott et al., 2001]	N	2
92	Ex 13	G-A nt 2064	Met688Ile	co	Japan		[Nomura et al., 2000]	N	3/4
SNU-H1024	Ex 13	G-A nt 2064	Met688Ile	co	Korea		[Yuan et al., 1998]	N	3/4
Case 31	Ex 13	T-C nt 2072	Ile691Arg	co	USA		[Samowitz et al., 2001]	N	2
56	Ex 13	T-C nt 2090 (*2089*)	Cys697Arg		Germany		[Raedle et al., 1999]	Y	2
FF46	Ex 13	T-C nt 2090 (*2089*)	Cys697 Arg	co/ut/leu	France		[Wang et al., 1999]	N	2
62	Ex 13	G-T nt 2090	Cys697Phe		Germany		[Wehner et al., 1997]	Y	2/3
28	Ex 13	G-T nt 2090	Cys697Phe		Germany		[Holinski-Feder et al., 2001]	N	2/3
21957/ 3A	Ex 13	del G nt 2113	frameshift from codon 704		USA		[Moslein et al., 1996]	Y	1
	Ex 13	C-T nt 2131	Arg711STOP		Argentina		Fodde, unpub.	Y	1
	Ex 13	G-C nt 2139	Gly713Gly		Finland	0.04	[Nystrom-Lahti et al., 1996]	Y	4
IMS6	Ex 13	C-T nt 2152	Gln718STOP		Australia		Bennett, unpub.	Y	1
	Ex 13	A-G nt 2154	Gln718Gln		Lithuania		Avizienyte, unpub.	Y	4
Family 32	Ex 13	A-G nt 2157	Gln719Gln	co	Australia		[Scott et al., 2001]	N	4
21907 or 4A	Ex 13	del T nt 2204	frameshift from codon 734		USA		[Moslein et al., 1996]	Y	1
TM		Ex 13 deleted	out of frame del Ex 13 codon 669-	co/HNPCC	USA/Finland		[Liu et al., 1994]	N	1

			737						
300	Ex 13	Ex 13 deleted	out of frame del Ex 13 codon 669-737	co/HNPCC	USA		[Jeon et al., 1996]	N	1
	IVS 13	t-c nt2211-6*(+6 or IVS12)*		co	USA		[Cunningham et al., 2001]	N	3/4
GC15825	IVS13	a-t nt 2211-2	Splice site		UK		West, unpub.	Y	1
MD579	IVS 13	g-t nt 2211 - 1	Splice defect, with mutation in Exon 12 – del codon 588-820	co/ut/ov	Scotland		[Farrington et al., 1998]	Y	1
MD435	IVS 13	g-t nt 2211-1	Splice defect, with mutation in Exon 12 – del codon 588-820	co/ut/ov/br	Scotland		Farrington/Dunlop, unpub.	N	1
BIL-IGR924	Ex 14	G-A nt 2251	Gly751Arg		France		[Dieumegard et al., 2000]	Y	2/3
1827	Ex 14	del C nt 2261	frameshift from codon 754		Switzerland		Heinimann, unpub.	Y	1
I-357	Ex 14	del C nt 2294	frameshift from codon 764	co	Italy		[Pensotti et al., 1997]	Y	1
	Ex 14	A-G nt 2308	Ile770Val		Scotland/USA	0.07	[Farrington et al., 1998]	N	3/4
NL-203	Ex 14	del C nt 2345	frameshift from codon 781	co/en/pr/bl/st	Netherlands		[Wijnen et al., 1995]	Y	1
96	Ex 14	del C nt 2345	frameshift from codon 781		Germany		[Raedle et al., 1999]	Y	1
22	Ex 14	ins TT nt 2360	frameshift from codon 787	co	Portugal		[Fidalgo et al., 2000]	N	1
HNP3/HNP129/ K-32	Ex 14	T-G nt 2432	Leu811STOP	co/ga/ov	Japan		[Miyaki et al., 1995]	Y	1
CFS69	Ex 15	C-T nt 2470	Gln824STOP	co/ut	Italy		[Fornasarig et al., 2000]	N	1
A-AV17	Ex 15	C-T nt 2470	Gln824STOP	co/ut	Italy		[Genuardi et al., 1998]	N	1
PLO-SG644	Ex 15	ins 14bp nt 2484	frameshift from codon 828		France		[Dieumegard et al., 2000]	Y	1

NL-57	Ex 15	G-A nt 2500	Ala834Thr	co/HNPCC	Netherlands		[Wijnen et al., 1997].	Y	2/3
	Ex 15	G-A nt 2500	Ala834Thr		Netherlands	0/100	[Wijnen et al., 1998a]	N	2/3
CFS112	Ex 15	G-A nt 2500	Ala834Thr	co	Italy		[Fornasarig et al., 2000]	N	2/3
h26	Ex 15	del CTAATTT nt 2501	frameshift from codon 833		England		Bunyan, unpub.	Y	1
Family 34	Ex 15	del TAATTTC nt 2502	stop codon	co	Australia		[Scott et al., 2001]	N	1
Case 31	Ex 15	A-G nt 2503	Asn835Asp	co	USA		[Samowitz et al., 2001]	N	3/4
SC	Ex 15	7-bp deletion	frameshift from codon 834	co	England		[Beck et al., 1997a]	N	1
31	Ex 15	7-bp deletion	frameshift from codon 834	co	England		[Coleman et al., 2001]	N	1
HNPCC-3	Ex 15		frameshift from codon 834	co	England		[Beck et al., 1997c]	N	1
K-30/HNP130	Ex 15	del T nt 2506	frameshift from codon 835	co	Japan		[Lu et al., 1996]	N	1
PT	Ex 15		frameshift from codon 843	co	England		[Beck et al., 1997a]	N	1
G726	Ex 15		Gly845STOP (*Lys845STOP*)	co	Italy		[Montera et al., 2000]	N	1
350*	Ex 15	A-G nt 2533	Lys845Glu	co	Japan		[Nomura et al., 2000]	N	2/3
MAR-SG63	Ex 15	C-T nt 2536	Gln846STOP		France		Bressac-de Paillerets, unpub.	Y	1
MAT-SG417	Ex 15	del C nt 2543	frameshift from codon 847		France		Bressac-de Paillerets, unpub.	Y	1
FF16	Ex 15	C-T nt 2581	Gln861STOP	co/d/ga/urt	France		[Wang et al., 1999]	N	1
37735	Ex 15	del 5 bp nt 2592	frameshift		UK		Norbury, unpub.	Y	1
HNP5	Ex 15	del AG nt 2629	frameshift from codon 876	co	Japan		[Miyaki et al., 1995]	Y	1
HNP-054	Ex 15	del AG nt 2633	frameshift from codon 877		French Canada		[Bapat et al., 1999]	Y	1
DP2, DP3	Ex 15	del AG nt 2633	frameshift from codon 877	co/ut	Canada		[Millar et al., 1999]	N	1

SNU-H1027	Ex 15	del GA	frameshift	co	Korea	[Yuan et al., 1998]	N	1
610	Ex 15	Ex 15 deleted	out of frame del Ex 15 codon 820-878	co/HNPCC	USA	[Luce et al., 1995]	N	1
DH	Ex 8-15	Ex 8-15 deleted	out of frame del Ex 8-15 codon 426-878	co/HNPCC	USA/Finland	[Liu et al., 1994]	N	1
JG	IVS 15	g-a nt 2634+1	out of frame del Ex 15 codon 820-878	co/HNPCC	USA/Finland	[Liu et al., 1994]	Y	1
30/96	Ex 16	del A nt 2647	frameshift from codon 883		Italy	[Percesepe et al., 2001]	N	1
K2064	Ex 16	del C nt 2662	frameshift from codon 887	co	USA	[Swensen et al., 1997]	N	1
C003	Ex 16	C-G nt 2714	Thr905Arg	co	England	[Froggatt et al., 1996]	Y	2/3
Case 32,33	Ex 16	T-C nt 2766	Phe922Phe	co	USA	[Samowitz et al., 2001]	N	4
	Ex 16	G-C nt 2800 (*2802*)	Thr933Thr (*934*)		Switzerland	[Heinimann et al., 1999]	N	4

* = more than one gene variant identified in the index case

Key to ‘Cancer’ Column:

am = ampulla of vater, b = brain, bd=bile duct, bl = bladder, bo = bone, br = breast, bs = basal cell, co = colon, cx = cervix, d = duodenum, en = endometrium, es = esophagus, ga = gastric, gb = gall bladder, gl = glioma, je = jejunum, k = kidney, l = lung, leu = leukaemia, ly = lymphoma, lv = liver, MT = Muir-Torre, ov = ovarian, pa = pancreas, ph = pharynx, pr = prostate, rh = rhabdomyosarcoma, re = rectal, rp = renal pelvic, sa = sarcoma, se = seminoma, si = small intestine, sk = skin, sp = spine, th = thyroid,TS = Turcots’, uc = ulcerative colitis, ur = urether, urt = urinary tract, ut = uterine,

Abbreviations:

del = deletion, Ex = exon, ins = insertion, HNPCC = Hereditary Non-Polyposis Colorectal Cancer, IVS = Intervening Sequence, N / A = Not Available, nt = mucleotide, unpub. = unpublished, UTR = Untranslated Region

Reference List

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