SUPPLEMENTAL METHODS

National Comprehensive Cancer Network (NCCN) Guideline Classification

Subjects were considered to fulfill NCCN guidelines for Lynch syndrome (LS) testing[1] if they had a personal history of any of the following: colorectal cancer (CRC) or endometrial cancer (EC) at age <50; ≥2 LS-associated cancers, regardless of age; CRC at any age plus a first-degree relative (FDR) with a LS-associated cancer at age <50; CRC at any age plus ≥2 FDR/second-degree relatives (SDR) with a LS-associated cancer at any age.

Regardless of their personal history, subjects were also considered to fulfill NCCN guidelines for LS testing[1] if they had a family history of any of the following: a FDR/SDR with CRC age <50; a FDR/SDR with EC age <50; a FDR/SDR with ≥2 LS-associated cancers at any age; a FDR/SDR with CRC at any age plus another FDR/SDR/third-degree relative (TDR) with any LS-associated cancer at age <50; a FDR with CRC at any age plus ≥2 FDR/SDR/TDR with any LS-associated cancer at any age. Unless otherwise specified on the test request form, all family history data were assumed to be from the same side of the subject's family.

Subjects were considered to fulfill NCCN guidelines for hereditary breast/ovarian cancer testing for *BRCA1* and *BRCA2* mutations[2] if they had a personal history of any of the following: breast cancer (BC) at age ≤45; male BC at any age; ovarian cancer (OC) at any age; BC at any age plus ≥1 FDR/SDR/TDR with BC at age ≤50; BC at any age plus ≥1 FDR/SDR/TDR with OC or male BC; BC at any age plus ≥2 FDR/SDR/TDR with BC at any age; BC at any age plus ≥2 FDR/SDR/TDR with pancreatic cancer (PC) at any age; PC at any age plus ≥2 FDR/SDR/TDR with BC, OC, or PC.

Regardless of their personal history, subjects were also considered to fulfill NCCN guidelines for hereditary breast/ovarian cancer testing for *BRCA1* and *BRCA2* mutations if they had a family history of any of the following: ≥1 FDR/SDR with OC at any age; ≥1 FDR/SDR with BC at age ≤45; ≥1 FDR/SDR with male BC at any age; ≥1 FDR/SDR with BC plus ≥1 FDR/SDR/TDR with BC, at least one of which was at age ≤50; ≥1 FDR/SDR with BC at any age plus ≥2 FDR/SDR/TDR with BC at any age; ≥1 FDR/SDR with BC plus ≥1 FDR/SDR/TDR with male BC; ≥1 FDR/SDR with BC plus ≥1 FDR/SDR/TDR with OC; ≥1 FDR/SDR with PC plus ≥2 FDR/SDR/TDR with BC, OC, or PC; ≥1 FDR/SDR with PC plus ≥1 FDR/SDR/TDR with a history of two or more of: BC, OC, or PC. Unless otherwise specified on the test request form, all family history data were assumed to be from the same side of the subject's family.

Design of Custom Primer Library for Next-Generation Sequencing (NGS) Target Enrichment

A custom primer library was designed, which includes regions of interest across 25 genes (Table 1) using the RainDance microdroplet PCR system (RainDance Technologies, Inc., Lexington, MA). The library design process began with identifying regions of interest, usually coding exons, for a panel of gene targets. This was done by evaluating results from the Consensus CDS database (http://www.ncbi.nlm.nih.gov/projects/CCDS/CcdsBrowse.cgi) and comparing them with published literature. In the event of multiple reported transcripts, a review of the available literature was performed to determine the transcript(s) most relevant for patient testing. For coding exons, the sequencing regions were flanked by 20 bases of upstream and 10 bases of downstream intronic sequence to allow for evaluation of variants that occur in conserved, proximal splicing elements. Automated primer design of the specified regions was performed by the supplier (RainDance Technologies, Inc., Lexington, MA). This design process included the comparison of putative priming sites to public variant databases and the current genome build (http://www.ncbi.nlm.nih.gov/projects/genome/assembly/grc/human/) to avoid non-specific priming

and common SNPs at primer binding sites. Primer sequences were also checked for predicted primer-primer interactions. Primer designs that pass these filters were arranged into multiplexes of five amplicons to minimize DNA input requirements and the resulting reagents were dropletized into a custom library (RainDance). The RainDance library design involved an iterative process with multiple rounds of testing; the version of the library used in this study contained a combination of multiplexed and single-plex PCR reactions.

Sample Preparation and Next Generation Sequencing

Genomic DNA was extracted from blood (QIAsymphony, Qiagen, Venlo, Netherlands) and was fragmented (SonicMan, Brooks Life Science Systems, Spokane, WA, USA) to approximately 3 kb to facilitate NGS sample preparation. Fragmented DNA was combined with PCR master mix containing the necessary buffers, polymerase and nucleotides. The reaction mix containing DNA was dropletized and merged with droplets containing a custom amplicon primer library using a high-throughput microfluidic emulsion PCR system (RainDance Thunderstorm). This process generated roughly 40,000 droplets corresponding to PCR microreactions per patient sample. The resulting emulsion was amplified for 55 cycles on a Mastercycler Pro thermalcycler (Eppendorf, Hamburg, Germany). The emulsion was broken up according to RainDance protocols and the aqueous PCR products were purified using AMPure XP (Beckman Coulter, Brea, California). Secondary PCR was performed to attach a six nucleotide identifier, specific for each sample within a batch, and recognition sites for NGS. The products were purified, pooled, and then sequenced on an Illumina HiSeq 2500 (Illumina, Inc., San Diego, CA, USA) to generate paired end, 2 x 150 bp reads according to the manufacturer's instructions.

To sequence portions of the *PMS2* and *CHEK2* genes with highly homologous pseudogenes, target enrichment was modified to include long-range PCR. The primary PCR was performed using LA Taq Hot

Start (Takara Bio Inc., Otsu, Japan) on 50ng of genomic DNA to generate gene-specific long range amplicons (LRA). The gene-specific LRA products were diluted 1:10,000 and a second round of PCR was performed to attach an index sequence specific for each sample within a batch and recognition sites for NGS. Equal volumes of each secondary amplicon product for all LRAs were combined per sample. Equal amounts of 96 samples were combined and diluted to 2nM for sequencing on the Illumina MiSeq (Illumina, Inc., San Diego, CA, USA) for 2 x 150 bp paired end sequencing reads.

NGS Data Analysis

DNA sequence reads were assessed using Illumina Sequence Control Software with Real Time Analysis (Illumina, Inc., San Diego, CA, USA). Sequence reads were trimmed at the point where quality scores drop below Q30 using an optimized Burrows-Wheeler Aligner trimming approach and then compared to a list of expected amplicon sequences. For *CHEK2* and *PMS2*, JAligner (Open Source - http://jaligner.sourceforge.net/) was used to determine which DNA target the sequencing read correctly matched; sequencing reads that matched the pseudogene better than the gene target were discarded. Sequence variants were identified by aligning reads using JAligner and comparing to the reference (wildtype) sequence. The average depth of coverage for samples in this study was over 1000X.

Large rearrangement (LR) detection was performed by relative copy number analysis of the NGS data in this study. The number of reads that mapped back to each exon was normalized using the total number of mapped back reads across all genes for that sample. For each run, or group of similar runs, a median normalized and the read count value was determined for each exon. Samples were then evaluated to see if their normalized read count average, across a given exon or partial exon, was ≥1.25X or ≤0.75X the median value. If a relative copy number value was ≥1.25X the median value, the sample was determined to have a heterozygous duplication. If a value was ≤0.75X the median value, the sample was determined

to have a heterozygous deletion.

The median noise of centered normalized read counts for all exons with normal copy number of 2 was determined. The CV (noise in a sample) was calculated as follows:

Median (Absolute Value (1-S) x 2) = Noise in a sample (CV)

S (centered normalized read counts for a given exon and sample) = N/(median N across all samples)

N (normalized read count for a given exon and sample) = c/C

c = Read count for an exon

C = Sum of all c for a sample

PMS2 exons 1-5, 11-15, and *CHEK2* exons 10-14, were excluded from this calculation as they do not hold to the same copy number assumption. If the calculated CV value was <0.08, then the sample was included for NGS data analysis of large rearrangements. If the CV value was ≥0.08, the sample was rejected for NGS LR analysis. By these criteria 90% of the samples in this study were eligible for LR analysis. Positive and negative calls made by the algorithm were reviewed by human analysts.

Pathogenicity Classification

All sequence variations and large rearrangements detected by the 25-gene panel were classified for pathogenicity into the following categories, as previously described: deleterious mutation, suspected deleterious mutation, variants of uncertain clinical significance (VUS), favor polymorphism, and polymorphism.[3, 4] Based on recommendations from the American College of Medical Genetics (ACMG),[4] deleterious mutations included nonsense and frameshift mutations predicted to result in protein truncation, as well as specific missense and intronic alterations that have been previously recognized as deleterious based on supporting linkage, functional, biochemical, and/or statistical evidence. Suspected deleterious mutations included alterations for which available evidence indicated a high likelihood – but

not confirmation – of pathogenicity. Individuals with deleterious or suspected deleterious genomic alterations were collectively defined as having "pathogenic" mutations (Supplemental Table 1).[4-42] Alterations were deemed favor polymorphism or polymorphism if available evidence indicated a low likelihood that such alterations altered normal gene expression and/or function. Alterations were classified as VUS if data were insufficient to support either a deleterious or benign interpretation (Supplemental Table 2 and Supplemental Figure).[4]

Validation of multigene panel

The performance characteristics of the 25-gene panel used in this study have been evaluated in a separate validation study.[43] In this validation study, the sequencing component of the 25-gene panel was validated by comparing the results of NGS with Sanger sequencing on 100 anonymized DNA samples. Samples were sequenced for the coding regions and proximal splice sites of all genes except for *EPCAM*, which is evaluated only for large rearrangements involving the terminal exons. A total of 3923 variants were identified including 3884 single nucleotide substitutions and 39 small insertions or deletions. These results showed 100% concordance between NGS and Sanger sequencing. The validation study gives an estimated analytical sensitivity of >99.92% (lower limit of the 95% confidence interval) and an estimated analytical specificity of >99.99% (lower limit of the 95% confidence interval) for the clinical assay (Minitab version 15, 1 proportion test, Exact method). Reproducibility, both within and between batches, was confirmed by running four samples in triplicate across three separate batches and verifying identical results.

Large rearrangement analysis of the 25-gene panel was also validated in this subsequent study.[43] All genes except for *PMS2* and *CHEK2* were validated for large rearrangement review using both microarray comparative genomic hybridization (CGH) and NGS for large rearrangement dosage analysis. Microarray

CGH was validated by correctly identifying all 51 LR positive samples among 212 anonymized samples. NGS dosage analysis requires higher DNA input volumes and was, therefore, validated on a subset of 49 LR positive samples among 110 anonymized samples. One LR positive sample failed during laboratory processing but NGS dosage analysis correctly identified all 48 LR positive samples that were reviewed as part of the validation and by both large rearrangement detection assays. Large rearrangement review for *PMS2* and *CHEK2* used multiplex ligation-dependent probe amplification (MLPA) and correctly identified all 5 LR positive samples among 110 anonymized samples in the validation set.

SUPPLEMENTAL MATERIAL REFERENCES

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Author names in bold designate shared co-first authors.

Supplemental Table 1: Germline details of 182 individuals found to carry pathogenic mutations with a 25-gene hereditary cancer panel

Subject ID	Gene mutated (Lynch)	HGVS mutation name (Lynch)	Gene mutated (non-Lynch)	HGVS mutation name (non-Lynch)
1095216833	*MLH1*	c.1279C>T (p.Gln427*)		
1095079933	*MLH1*	c.1435_1453del (p.Val479Ilefs*6)		
1086153088	*MLH1*	c.1517T>C (p.Val506Ala)		
0159657500	*MLH1*	c.1746del (p.Phe583Leufs*8)		
1093189976	*MLH1*	c.1852_1854del (p.Lys618del)		
1097149016	*MLH1*	c.1890dupT (p.Asp631*)		
0159893397	*MLH1*	c.1975C>T (p.Arg659*)		
1093190211	*MLH1*	c.1989+3dupC		
1092248139	*MLH1*	c.199G>A (p.Gly67Arg)		
1092247994	*MLH1*	c.208-3C>G		
1093189969	*MLH1*	c.298C>T (p.Arg100*)		
1093189929	*MLH1*	c.350C>T (p.Thr117Met)		
1097162685	*MLH1*	c.380G>T (p.Arg127Ile)		
1092248112	*MLH1*	c.544A>G (p.Arg182Gly)		
1095079935	*MLH1*	c.546-2A>G		
1086153104	*MLH1*	c.589-2A>G		
1095216822	*MLH1*	c.589-2A>G		
1095079895	*MLH1*	c.676C>T (p.Arg226*)		
1086153352	*MLH1*	c.677G>A (p.Arg226Gln)		
1092248097	*MLH1*	c.677G>A (p.Arg226Gln)		
1092248246	*MLH1*	c.677G>A (p.Arg226Gln)		
1086153414	*MLH1*	c.755C>G (p.Ser252*)		
1093189911	*MLH1*	c.790+2T>C		

1097149010	*MLH1*	c.883A>G (p.Ser295Gly)
1095188508	*MLH1*	c.884+4A>G
1092248249	*MLH1*	c.971dupA (p.Arg325Alafs*37)
0159657452	*MLH1*	del exon 13
1095112480	*MLH1*	del exon 5
1095112487	*MLH1*	del exons 1-13
1095188520	*MLH1*	del exons 1-13
1092248006	*MLH1*	dup exons 6-12
1086153113	*MSH2*	c.1076+1G>A
1097162663	*MSH2*	c.1076+1G>A
1086153044	*MSH2*	c.124_127dupTTCT (p.Tyr43Phefs*40)
1095188535	*MSH2*	c.1444A>T (p.Arg482*)
1092248225	*MSH2*	c.1511-1G>A
1097149017	*MSH2*	c.1552_1553del (p.Gln518Valfs*10)
0159657472	*MSH2*	c.1700_1704del (p.Lys567Argfs*3)
0159657450	*MSH2*	c.1760-3C>G
1095112429	*MSH2*	c.1906G>C (p.Ala636Pro)
0159891914	*MSH2*	c.1968C>A (p.Tyr656*)
1097162697	*MSH2*	c.2038C>T (p.Arg680*)
0159893404	*MSH2*	c.2047G>A (p.Gly683Arg)
1097149031	*MSH2*	c.2047G>A (p.Gly683Arg)
1095216850	*MSH2*	c.2210+1G>A
0159657471	*MSH2*	c.2222_2223del (p.Lys741Argfs*8)
1086153380	*MSH2*	c.2291G>A (p.Trp764*)
1093190203	*MSH2*	c.2634+1G>A

1092248052	*MSH2*	c.367-1G>A
1092248028	*MSH2*	c.587del (p.Pro196Glnfs*18)
1086153076	*MSH2*	c.842C>G (p.Ser281*)
1097162637	*MSH2*	c.914_923del (p.Ala305Glufs*23)
1097162639	*MSH2*	c.942+2T>A
1092248051	*MSH2*	c.942+3A>T
1093189908	*MSH2*	c.942+3A>T
1093189960	*MSH2*	c.942+3A>T
1095079883	*MSH2*	c.942+3A>T
1093190258	*MSH2*	del exon 3
1086153348	*MSH2*	del exon 8
0159657410	*MSH2*	del exons 1-6
0159893399	*MSH2*	del exons 1-6
1092248055	*MSH2*	del exons 1-6
1092248221	*MSH2*	del exons 1-6
1092248241	*MSH2*	del exons 1-6
1095079914	*MSH2*	del exons 1-6
1095216857	*MSH2*	del exons 1-6
1097149087	*MSH2*	del exons 1-6
1086153066	*MSH2*	del exons 8-15
1095188549	*MSH2*	dup exon 6
1092248133	*MSH6*	c.10C>T (p.Gln4*)
1095188551	*MSH6*	c.10C>T (p.Gln4*)
1086153366	*MSH6*	c.1444C>T (p.Arg482*)
0159893462	*MSH6*	c.1571dupA (p.Tyr524*)

1097149067	*MSH6*	c.1634_1635del (p.Lys545Argfs*17)
0159893401	*MSH6*	c.2057G>A (p.Gly686Asp)
1097162715	*MSH6*	c.220G>T (p.Gly74*)
1095079885	*MSH6*	c.2906_2907del (p.Tyr969Leufs*5)
1093189924	*MSH6*	c.3261del (p.Phe1088Serfs*2)
0159893435	*MSH6*	c.3439-2A>G
1095112495	*MSH6*	c.3439-2A>G
1092247995	*MSH6*	c.3516_3517del (p.Arg1172Serfs*4)
1095112415	*MSH6*	c.3647-1G>A
1093190249	*MSH6*	c.3699_3702del (p.Lys1233Asnfs*6)
1097162643	*MSH6*	c.3802-14_3809del
1095112405	*MSH6*	c.3934_3937dupGTTA (p.Ile1313Serfs*7)
1086153080	*MSH6*	c.3939_3957dup (p.Ala1320Serfs*5)
1092248186	*MSH6*	c.3939_3957dup (p.Ala1320Serfs*5)
1095112465	*MSH6*	c.3939_3957dup (p.Ala1320Serfs*5)
1095216873	*MSH6*	c.3939_3957dup (p.Ala1320Serfs*5)
1086153089	*MSH6*	c.3959_3962del (p.Ala1320Glufs*6)
1092248103	*MSH6*	c.3959_3962del (p.Ala1320Glufs*6)
1092248204	*MSH6*	c.4001+1G>A
0159657467	*MSH6*	c.694C>T (p.Gln232*)
1095112417	*MSH6*	c.694C>T (p.Gln232*)
1095079855	*PMS2*	c.137G>T (p.Ser46Ile)
1092248016	*PMS2*	c.1831dupA (p.Ile611Asnfs*2)
1092248177	*PMS2*	c.1840A>T (p.Lys614*)
1092248205	*PMS2*	c.1840A>T (p.Lys614*)

1092248226	*PMS2*	c.1882C>T (p.Arg628*)
1092248166	*PMS2*	c.1A>T (p.Met1?)
1095112410	*PMS2*	c.214G>T (p.Gly72*)
1092248021	*PMS2*	c.251-2A>T
1097149076	*PMS2*	c.736_741delins11 (p.Pro246Cysfs*3)
1095112490	*PMS2*	c.746_753del (p.Asp249Valfs*2)
0159657458	*PMS2*	c.765C>A (p.Tyr255*)
1093189907	*PMS2*	c.861_864del (p.Arg287Serfs*19)
1092248136	*PMS2*	del exons 8-11
1093189949	*PMS2*	del exons 9-10
1095112488	*EPCAM*	del exons 1-9
1093189941	*EPCAM*	del exons 2-9
0159893426	*EPCAM*	del exons 6-9
0159893400	*APC*	c.3927_3931del (p.Glu1309Aspfs*4)
1092248247	*APC*	c.268A>T (p.Lys90*)
1093190205	*APC*	c.531+5G>A
1095188522	*APC*	c.70C>T (p.Arg24*)
1097162691	*APC*	c.667C>T (p.Gln223*)
0159657480	*MUTYH* (biallelic)	c.325C>T (p.Arg109Trp) AND c.1187G>A (p.Gly396Asp)
1095112444	*MUTYH* (biallelic)	c.1187G>A (p.Gly396Asp) homozygous
1095216806	*MUTYH* (biallelic)	c.1187G>A (p.Gly396Asp) homozygous
0159657488	*BRCA1*	del exon 21
0159893405	*BRCA1*	c.5266dupC (p.Gln1756Profs*74)
1092248015	*BRCA1*	c.5266dupC (p.Gln1756Profs*74)
1092248047	*BRCA1*	c.5066T>G (p.Met1689Arg)

1093189935	*BRCA1*	c.5266dupC (p.Gln1756Profs*74)
1093189938	*BRCA1*	c.5096G>A (p.Arg1699Gln)
0159893382	*BRCA2*	c.4647_4650del (p.Lys1549Asnfs*18)
1092248111	*BRCA2*	c.9382C>T (p.Arg3128*)
1092248179	*BRCA2*	c.5946del (p.Ser1982Argfs*22)
1093189918	*BRCA2*	c.4631dupA (p.Asn1544Lysfs*4)
1093190286	*BRCA2*	c.7008-2A>G
1095079936	*BRCA2*	c.846_847del (p.Ile283Trpfs*11)
1095112479	*BRCA2*	c.1296_1297del (p.Asn433Glnfs*18)
1095216848	*BRCA2*	c.2971_2983del (p.Asn991Aspfs*3)
1097162706	*BRCA2*	c.5946del (p.Ser1982Argfs*22)
1086153128	*ATM*	c.8371_8374del (p.Tyr2791Glyfs*14)
1092248032	*ATM*	c.1066-3_1072del
1092248117	*ATM*	c.5414G>A (p.Trp1805*)
1093189919	*ATM*	c.1564_1565del (p.Glu522Ilefs*43)
1095188583	*ATM*	c.8395_8404del (p.Phe2799Lysfs*4)
1095214818	*ATM*	c.8824C>T (p.Gln2942*)
1095216856	*ATM*	c.7271T>G (p.Val2424Gly)
1097149023	*ATM*	c.6015dupC (p.Glu2007Argfs*11)
1086153125	*BARD1*	c.1690C>T (p.Gln564*)
1092248067	*BRIP1*	c.2392C>T (p.Arg798*)
1093190284	*BRIP1*	c.484C>T (p.Arg162*)
1092247990	*CHEK2*	c.444+1G>A
1092248022	*CHEK2*	c.1100del (p.Thr367Metfs*15)
1092248202	*CHEK2*	c.444+1G>A

1093189977			*CHEK2*	c.909-2A>G
1095112448			*CHEK2*	c.1100del (p.Thr367Metfs*15)
1092248056			*NBN*	c.657_661del (p.Lys219Asnfs*16)
1095079919			*PALB2*	c.758dupT (p.Ser254Ilefs*3)
1093190269			*RAD51C*	c.890_899del (p.Leu297Hisfs*2)
1093190213	*MSH2*	c.1576del (p.Thr526Profs*17)	*ATM*	del exons 61-62
0159657434	*MSH6*	c.3261dupC (p.Phe1088Leufs*5)	*STK11*	c.375-1C>T
1092248195	*MSH2*	del exons 1-6	*MUTYH* (monoallelic)	c.1187G>A (p.Gly396Asp)
0159657427			*MUTYH* (monoallelic)	c.1187G>A (p.Gly396Asp)
0159657441			*MUTYH* (monoallelic)	c.1187G>A (p.Gly396Asp)
0159657473			*MUTYH* (monoallelic)	c.1187G>A (p.Gly396Asp)
0159893402			*MUTYH* (monoallelic)	c.536A>G (p.Tyr179Cys), c.494A>G (p.Tyr165Cys)
0159893438			*MUTYH* (monoallelic)	c.1187G>A (p.Gly396Asp)
1092248023			*MUTYH* (monoallelic)	c.1187G>A (p.Gly396Asp)
1092248192			*MUTYH* (monoallelic)	c.1187G>A (p.Gly396Asp)
1092248228			*MUTYH* (monoallelic)	c.1147del (p.Ala385Profs*23)
1092248266			*MUTYH* (monoallelic)	c.536A>G (p.Tyr179Cys)
1093189965			*MUTYH* (monoallelic)	c.1187G>A (p.Gly396Asp)
1093189978			*MUTYH* (monoallelic)	c.536A>G (p.Tyr179Cys)
1093190192			*MUTYH* (monoallelic)	c.1187G>A (p.Gly396Asp)
1093190226			*MUTYH* (monoallelic)	c.1187G>A (p.Gly396Asp)
1095079872			*MUTYH* (monoallelic)	c.1227_1228dupGG (p.Glu410Glyfs*43)
1095079891			*MUTYH* (monoallelic)	c.933+3A>C
1095079913			*MUTYH* (monoallelic)	c.1187G>A (p.Gly396Asp)
1095112413			*MUTYH* (monoallelic)	c.1187G>A (p.Gly396Asp)

1095112432	*MUTYH* (monoallelic)	c.1187G>A (p.Gly396Asp)
1095112475	*MUTYH* (monoallelic)	c.1187G>A (p.Gly396Asp)
1095112499	*MUTYH* (monoallelic)	c.1187G>A (p.Gly396Asp)
1095188584	*MUTYH* (monoallelic)	c.1187G>A (p.Gly396Asp)
1095216832	*MUTYH* (monoallelic)	c.1187G>A (p.Gly396Asp)
1095216898	*MUTYH* (monoallelic)	c.536A>G (p.Tyr179Cys)
1097149007	*MUTYH* (monoallelic)	c.536A>G (p.Tyr179Cys)
1097149020	*MUTYH* (monoallelic)	c.734G>A (p.Arg245His)
1097149054	*MUTYH* (monoallelic)	c.1227_1228dupGG (p.Glu410Glyfs*43)

Supplemental Table 2: Germline variants of uncertain significance detected by a multigene hereditary cancer panel in 1260 individuals undergoing Lynch syndrome testing

Subject ID	Gene	HGVS alteration name
0159657411	*BMPR1A*	c.1327C>T (p.Arg443Cys)
0159657412	*BRIP1*	c.3293C>A (p.Ala1098Asp)
0159657416	*PALB2*	c.2135C>T (p.Ala712Val)
0159657420	*BARD1*	c.1738G>A (p.Glu580Lys)
0159657423	*BARD1*	c.33G>T (p.Gln11His)
0159657425	*ATM*	c.7313C>T (p.Thr2438Ile)
0159657425	*ATM*	c.334G>A (p.Ala112Thr)
0159657425	*ATM*	c.320G>A (p.Cys107Tyr)
0159657425	*BARD1*	c.1738G>A (p.Glu580Lys)
0159657425	*CDH1*	c.833-16C>G
0159657425	*CDH1*	c.595A>T (p.Thr199Ser)
0159657425	*NBN*	c.37+5G>A
0159657425	*PTEN*	c.802-51_802-14del
0159657425	*RAD51D*	c.904-11T>A
0159657429	*ATM*	c.5675-10T>G
0159657429	*CHEK2*	c.1343T>G (p.Ile448Ser)
0159657429	*NBN*	c.1489A>G (p.Thr497Ala)
0159657430	*NBN*	c.1882G>A (p.Glu628Lys)
0159657432	*TP53*	c.139C>T (p.Pro47Ser)
0159657434	*NBN*	c.1690G>A (p.Glu564Lys)
0159657438	*ATM*	c.2289T>A (p.Phe763Leu)
0159657438	*MSH6*	c.972A>C (p.Lys324Asn)
0159657442	*MLH1*	c.479C>T (p.Ala160Val)
0159657448	*BARD1*	c.1242G>A (p.Met414Ile)
0159657450	*BMPR1A*	c.1327C>T (p.Arg443Cys)
0159657451	*MSH6*	c.3257C>T (p.Pro1086Leu)
0159657454	*RAD51C*	c.376G>A (p.Ala126Thr)
0159657455	*MLH1*	c.-42C>T
0159657456	*ATM*	c.4388T>G (p.Phe1463Cys)
0159657456	*ATM*	c.1810C>T (p.Pro604Ser)
0159657457	*MSH6*	c.743G>C (p.Arg248Pro)
0159657459	*SMAD4*	c.586A>G (p.Ser196Gly)
0159657460	*PMS2*	c.595C>T (p.Arg199Cys)
0159657460	*RAD51D*	c.973G>A (p.Gly325Ser)
0159657462	*ATM*	c.2096A>G (p.Glu699Gly)
0159657462	*NBN*	c.797C>T (p.Pro266Leu)
0159657462	*PTEN*	c.802-51_802-14del
0159657462	*RAD51D*	c.695G>A (p.Arg232Gln)
0159657467	*MSH6*	c.1180T>G (p.Ser394Ala)
0159657469	*NBN*	c.1489A>G (p.Thr497Ala)
0159657470	*PMS2*	c.1715C>T (p.Ala572Val)
0159657470	*RAD51C*	c.706-13C>G
0159657473	*ATM*	c.1229T>C (p.Val410Ala)
0159657474	*APC*	c.6639G>A (p.Met2213Ile)
0159657475	*MUTYH*	c.998-9C>T
0159657476	*ATM*	c.6067G>A (p.Gly2023Arg)
0159657483	*ATM*	c.8915A>G (p.Gln2972Arg)

0159657485	*RAD51C*	c.146-8A>G
0159657487	*CDH1*	c.2413G>A (p.Asp805Asn)
0159657487	*PMS2*	c.2350G>A (p.Asp784Asn)
0159657488	*MSH2*	c.1480T>C (p.Ser494Pro)
0159657491	*APC*	c.1825G>A (p.Val609Ile)
0159657494	*MLH1*	c.1942C>T (p.Pro648Ser)
0159657496	*BRIP1*	c.1198G>T (p.Asp400Tyr)
0159657497	*PALB2*	c.2606C>T (p.Ser869Phe)
0159657501	*CDKN2A (P16)*	c.430C>T (p.Arg144Cys)
0159657503	*PMS2*	c.1864A>G (p.Met622Val)
0159893377	*ATM*	c.5071A>C (p.Ser1691Arg)
0159893378	*ATM*	c.1229T>C (p.Val410Ala)
0159893378	*MSH6*	c.3854T>C (p.Phe1285Ser)
0159893381	*ATM*	c.6995T>C (p.Leu2332Pro)
0159893381	*CDH1*	c.833-16C>G
0159893382	*ATM*	c.6095G>A (p.Arg2032Lys)
0159893384	*ATM*	c.3175G>A (p.Ala1059Thr)
0159893386	*APC*	c.6724A>G (p.Ser2242Gly)
0159893395	*BMPR1A*	c.1327C>T (p.Arg443Cys)
0159893397	*CDKN2A (P14ARF)*	dup exons 1-2
0159893397	*CDKN2A (P16)*	dup exons 1-3
0159893399	*MSH6*	c.3961A>G (p.Arg1321Gly)
0159893401	*TP53*	c.704A>G (p.Asn235Ser)
0159893402	*APC*	c.5528C>T (p.Pro1843Leu)
0159893403	*ATM*	c.1229T>C (p.Val410Ala)
0159893403	*ATM*	c.5612C>T (p.Thr1871Ile)
0159893403	*CHEK2*	c.470T>C (p.Ile157Thr)
0159893404	*PTEN*	c.802-51_802-14del
0159893404	*PTEN*	c.1061C>A (p.Pro354Gln)
0159893408	*MSH2*	c.174C>G (p.Phe58Leu)
0159893416	*CHEK2*	c.751A>T (p.Ile251Phe)
0159893416	*PALB2*	c.1696C>T (p.Arg566Cys)
0159893417	*MSH6*	c.1474_1476del (p.Met492del)
0159893421	*CDKN2A (P16)*	c.-33G>C
0159893424	*CDH1*	c.1603A>T (p.Ile535Phe)
0159893427	*BRIP1*	c.2220G>T (p.Gln740His)
0159893428	*APC*	c.4919G>A (p.Arg1640Gln)
0159893429	*BARD1*	c.1738G>A (p.Glu580Lys)
0159893429	*CDH1*	c.833-16C>G
0159893430	*BARD1*	c.2282G>A (p.Ser761Asn)
0159893430	*MSH2*	c.1847C>G (p.Pro616Arg)
0159893432	*NBN*	c.683T>G (p.Ile228Arg)
0159893433	*TP53*	c.665C>T (p.Pro222Leu)
0159893434	*BRCA1*	c.3535A>C (p.Lys1179Gln)
0159893436	*APC*	c.2993G>T (p.Gly998Val)
0159893438	*ATM*	c.7122A>C (p.Glu2374Asp)
0159893439	*ATM*	c.544G>C (p.Val182Leu)
0159893439	*NBN*	c.1882G>A (p.Glu628Lys)
0159893441	*APC*	c.4711_4713del (p.Asp1571del)
0159893444	*CDKN2A (P16)*	c.361C>G (p.Leu121Val)

0159893444	*PTEN*	c.802-51_802-14del
0159893445	*APC*	c.6363_6365dupTGC (p.Ala2122dup)
0159893445	*CDK4*	c.431A>G (p.Glu144Gly)
0159893445	*CDKN2A (P16)*	c.150+6T>C
0159893446	*CDK4*	c.460G>C (p.Val154Leu)
0159893450	*CDH1*	c.2494G>A (p.Val832Met)
0159893450	*TP53*	c.31G>C (p.Glu11Gln)
0159893451	*MSH6*	c.1402C>T (p.Arg468Cys)
0159893453	*TP53*	c.97-6C>T
0159893457	*BMPR1A*	c.1513G>A (p.Ala505Thr)
0159893458	*CDH1*	c.1888C>G (p.Leu630Val)
0159893458	*NBN*	c.1690G>A (p.Glu564Lys)
0159893464	*SMAD4*	c.1573A>G (p.Ile525Val)
0159893467	*ATM*	c.2608A>G (p.Asn870Asp)
1086153037	*MUTYH*	c.724G>T (p.Val242Leu)
1086153040	*BRCA2*	dup exons 23-24
1086153055	*BARD1*	c.1738G>A (p.Glu580Lys)
1086153055	*PMS2*	c.847A>C (p.Ser283Arg)
1086153058	*RAD51C*	c.506T>C (p.Val169Ala)
1086153058	*SMAD4*	c.542C>G (p.Thr181Ser)
1086153059	*ATM*	c.6067G>A (p.Gly2023Arg)
1086153060	*BARD1*	c.1795G>A (p.Glu599Lys)
1086153062	*BMPR1A*	c.1066C>T (p.Pro356Ser)
1086153066	*ATM*	c.1229T>C (p.Val410Ala)
1086153066	*ATM*	c.4066A>G (p.Asn1356Asp)
1086153075	*APC*	c.6637A>G (p.Met2213Val)
1086153076	*MUTYH*	c.998-18G>A
1086153076	*PMS2*	c.1717A>T (p.Thr573Ser)
1086153078	*PMS2*	c.1723A>G (p.Asn575Asp)
1086153078	*PMS2*	c.2386G>A (p.Val796Ile)
1086153086	*BRCA2*	c.898G>A (p.Val300Ile)
1086153092	*CDKN2A (P16)*	c.434T>C (p.Ile145Thr)
1086153100	*CDKN2A (P14ARF)*	c.217A>C (p.Ser73Arg)
1086153101	*ATM*	c.5821G>C (p.Val1941Leu)
1086153106	*APC*	c.1481G>A (p.Ser494Asn)
1086153106	*MSH6*	c.3284G>A (p.Arg1095His)
1086153107	*BARD1*	c.26_40del (p.Asn9_Arg13del)
1086153108	*ATM*	c.8228C>T (p.Thr2743Met)
1086153108	*CHEK2*	c.470T>C (p.Ile157Thr)
1086153112	*MSH6*	c.1858G>A (p.Gly620Ser)
1086153112	*MUTYH*	c.1508G>A (p.Gly503Glu)
1086153113	*BARD1*	c.2008A>G (p.Lys670Glu)
1086153113	*BRCA1*	c.2522G>C (p.Arg841Pro)
1086153114	*CHEK2*	c.428A>G (p.His143Arg)
1086153116	*CHEK2*	c.538C>T (p.Arg180Cys)
1086153122	*ATM*	c.544G>C (p.Val182Leu)
1086153122	*PMS2*	c.123_131del (p.Leu42_Glu44del)
1086153130	*MUTYH*	c.1518+5G>A
1086153131	*SMAD4*	c.788-1G>C
1086153325	*CDKN2A (P16)*	c.-33G>C

1086153332	*MLH1*	c.52C>T (p.Arg18Cys)
1086153345	*STK11*	c.1193C>T (p.Ala398Val)
1086153348	*ATM*	c.8965C>G (p.Gln2989Glu)
1086153353	*APC*	c.3511C>T (p.Arg1171Cys)
1086153356	*RAD51C*	c.376G>A (p.Ala126Thr)
1086153363	*NBN*	c.797C>T (p.Pro266Leu)
1086153363	*TP53*	c.139C>T (p.Pro47Ser)
1086153365	*ATM*	c.290T>C (p.Ile97Thr)
1086153365	*BRIP1*	c.2236A>G (p.Ile746Val)
1086153365	*NBN*	c.797C>T (p.Pro266Leu)
1086153365	*RAD51C*	c.922G>T (p.Ala308Ser)
1086153365	*STK11*	c.894C>A (p.Phe298Leu)
1086153375	*CDKN2A (P14ARF)*	c.361G>A (p.Ala121Thr)
1086153375	*PALB2*	c.94C>G (p.Leu32Val)
1086153376	*MLH1*	c.144A>C (p.Gln48His)
1086153376	*TP53*	c.139C>T (p.Pro47Ser)
1086153377	*ATM*	c.6315G>C (p.Arg2105Ser)
1086153399	*ATM*	c.6995T>C (p.Leu2332Pro)
1086153399	*BARD1*	c.620A>G (p.Lys207Arg)
1086153399	*NBN*	c.505C>T (p.Arg169Cys)
1086153399	*SMAD4*	c.880A>G (p.Met294Val)
1086153399	*TP53*	c.97-6C>T
1086153401	*RAD51C*	c.376G>A (p.Ala126Thr)
1086153404	*BRCA2*	c.292T>G (p.Leu98Val)
1086153404	*RAD51C*	c.211A>T (p.Asn71Tyr)
1086153414	*ATM*	c.1229T>C (p.Val410Ala)
1086153418	*ATM*	c.5071A>C (p.Ser1691Arg)
1086153418	*MSH6*	c.3226C>G (p.Arg1076Gly)
1092247687	*CHEK2*	c.1343T>G (p.Ile448Ser)
1092247989	*MSH2*	c.2006G>A (p.Gly669Asp)
1092247992	*BARD1*	c.1569-13C>G
1092247997	*BARD1*	c.1347A>G (p.Gln449Gln)
1092247998	*ATM*	c.3925G>A (p.Ala1309Thr)
1092248000	*BMPR1A*	c.1327C>T (p.Arg443Cys)
1092248002	*ATM*	c.6919C>T (p.Leu2307Phe)
1092248002	*ATM*	c.1744T>C (p.Phe582Leu)
1092248002	*BMPR1A*	c.1243G>A (p.Glu415Lys)
1092248002	*BRCA2*	c.7444A>G (p.Thr2482Ala)
1092248004	*CDH1*	c.670C>T (p.Arg224Cys)
1092248004	*CDH1*	c.2440-6C>G
1092248004	*MSH2*	c.499G>C (p.Asp167His)
1092248005	*APC*	c.5635G>T (p.Ala1879Ser)
1092248006	*PTEN*	c.892C>G (p.Gln298Glu)
1092248007	*APC*	c.1606G>A (p.Glu536Lys)
1092248007	*NBN*	c.797C>T (p.Pro266Leu)
1092248007	*RAD51D*	c.983C>T (p.Thr328Ile)
1092248008	*BRIP1*	c.2372A>G (p.Asp791Gly)
1092248008	*CDK4*	c.779T>A (p.Val260Glu)
1092248010	*ATM*	c.544G>C (p.Val182Leu)
1092248012	*APC*	c.6679G>T (p.Gly2227Cys)

1092248012	*BRIP1*	c.890A>G (p.Lys297Arg)
1092248022	*CDH1*	c.2440-6C>G
1092248024	*RAD51D*	c.904-3C>T
1092248025	*RAD51C*	c.376G>A (p.Ala126Thr)
1092248031	*CDKN2A (P16)*	c.170C>T (p.Ala57Val)
1092248031	*MSH6*	c.3930G>C (p.Glu1310Asp)
1092248034	*NBN*	c.278C>T (p.Ser93Leu)
1092248043	*PALB2*	c.1846G>C (p.Asp616His)
1092248044	*PMS2*	c.1717A>T (p.Thr573Ser)
1092248044	*PMS2*	c.1714G>A (p.Ala572Thr)
1092248045	*CDH1*	c.1888C>G (p.Leu630Val)
1092248045	*PALB2*	c.3035C>T (p.Thr1012Ile)
1092248045	*PALB2*	c.1273G>A (p.Val425Met)
1092248045	*RAD51C*	c.635G>A (p.Arg212His)
1092248047	*MSH2*	c.1697_1709delinsTTCT (p.Asn566_Tyr570delinsIleLeu)
1092248048	*ATM*	c.7187C>G (p.Thr2396Ser)
1092248054	*BRCA2*	c.8222A>G (p.Lys2741Arg)
1092248060	*CHEK2*	c.470T>C (p.Ile157Thr)
1092248061	*ATM*	c.4709T>C (p.Val1570Ala)
1092248065	*RAD51C*	c.790G>A (p.Gly264Ser)
1092248068	*APC*	c.6679G>T (p.Gly2227Cys)
1092248068	*NBN*	c.321-17C>G
1092248069	*STK11*	c.1211C>T (p.Ser404Phe)
1092248070	*MSH6*	c.3299C>T (p.Thr1100Met)
1092248072	*PALB2*	c.2135C>T (p.Ala712Val)
1092248074	*ATM*	c.1810C>T (p.Pro604Ser)
1092248076	*CDKN2A (P16)*	c.-2G>A
1092248077	*BRIP1*	c.584T>C (p.Leu195Pro)
1092248080	*BRCA2*	c.6158C>A (p.Ser2053Tyr)
1092248082	*ATM*	c.8228C>T (p.Thr2743Met)
1092248082	*MUTYH*	c.700G>A (p.Val234Met)
1092248084	*ATM*	c.4375G>A (p.Gly1459Arg)
1092248085	*CDK4*	c.684-4A>T
1092248086	*APC*	c.4332A>T (p.Gln1444His)
1092248086	*CDH1*	c.88C>A (p.Pro30Thr)
1092248087	*RAD51C*	c.790G>A (p.Gly264Ser)
1092248088	*SMAD4*	c.554C>A (p.Pro185Gln)
1092248091	*CDK4*	c.155G>A (p.Ser52Asn)
1092248097	*STK11*	c.1211C>T (p.Ser404Phe)
1092248099	*ATM*	c.5071A>C (p.Ser1691Arg)
1092248101	*APC*	c.1391A>G (p.His464Arg)
1092248102	*BARD1*	c.2306C>T (p.Ser769Phe)
1092248102	*PMS2*	c.58C>T (p.Arg20Trp)
1092248103	*CDH1*	c.2440-6C>G
1092248115	*ATM*	c.5071A>C (p.Ser1691Arg)
1092248115	*NBN*	c.511A>G (p.Ile171Val)
1092248117	*APC*	c.7797A>C (p.Lys2599Asn)
1092248119	*ATM*	c.8147T>C (p.Val2716Ala)
1092248125	*ATM*	c.280A>G (p.Met94Val)
1092248127	*STK11*	c.970C>G (p.Pro324Ala)

1092248130	*CDH1*	c.2635G>A (p.Gly879Ser)
1092248130	*PMS2*	c.614A>C (p.Gln205Pro)
1092248131	*CHEK2*	c.1567C>T (p.Arg523Cys)
1092248134	*APC*	c.3299C>T (p.Ser1100Phe)
1092248135	*CHEK2*	c.410G>A (p.Arg137Gln)
1092248138	*NBN*	c.1882G>A (p.Glu628Lys)
1092248140	*MUTYH*	c.985G>A (p.Val329Met)
1092248146	*SMAD4*	c.-3C>G
1092248148	*NBN*	c.-2C>A
1092248149	*CHEK2*	c.14C>T (p.Ser5Leu)
1092248151	*ATM*	c.6860G>C (p.Gly2287Ala)
1092248159	*NBN*	c.1262T>C (p.Leu421Ser)
1092248162	*BRIP1*	c.2220G>T (p.Gln740His)
1092248164	*BRIP1*	c.584T>C (p.Leu195Pro)
1092248167	*CDKN2A (P16)*	c.-25C>T
1092248181	*ATM*	c.1810C>T (p.Pro604Ser)
1092248181	*BMPR1A*	c.1235T>C (p.Val412Ala)
1092248181	*CDKN2A (P16)*	c.-25C>T
1092248183	*NBN*	c.1882G>A (p.Glu628Lys)
1092248184	*BRIP1*	c.584T>C (p.Leu195Pro)
1092248184	*CHEK2*	c.593-20_593-18del
1092248185	*NBN*	c.1489A>G (p.Thr497Ala)
1092248185	*PALB2*	c.100C>T (p.Arg34Cys)
1092248185	*PTEN*	c.802-51_802-14del
1092248192	*MSH2*	c.167A>T (p.Glu56Val)
1092248193	*ATM*	c.7475T>G (p.Leu2492Arg)
1092248197	*ATM*	c.1229T>C (p.Val410Ala)
1092248197	*BRIP1*	c.584T>C (p.Leu195Pro)
1092248198	*MLH1*	c.1709A>G (p.Asn570Ser)
1092248205	*NBN*	c.1035C>T (p.Gly345Gly)
1092248206	*BRCA2*	c.8990A>G (p.Tyr2997Cys)
1092248206	*NBN*	c.797C>T (p.Pro266Leu)
1092248206	*PMS2*	c.497T>C (p.Leu166Pro)
1092248217	*STK11*	c.721G>A (p.Ala241Thr)
1092248223	*ATM*	c.5071A>C (p.Ser1691Arg)
1092248225	*APC*	c.5690A>C (p.His1897Pro)
1092248225	*BRIP1*	c.1433A>G (p.His478Arg)
1092248227	*BRIP1*	c.2236A>G (p.Ile746Val)
1092248234	*BMPR1A*	c.749T>C (p.Met250Thr)
1092248238	*NBN*	c.613A>G (p.Ile205Val)
1092248239	*PALB2*	c.2816T>G (p.Leu939Trp)
1092248246	*NBN*	c.1354A>C (p.Thr452Pro)
1092248246	*PTEN*	c.802-51_802-14del
1092248251	*SMAD4*	c.455-6A>G
1092248252	*ATM*	c.496+4T>C
1092248255	*CHEK2*	c.853A>T (p.Ile285Phe)
1092248263	*ATM*	c.3925G>A (p.Ala1309Thr)
1092248265	*BRIP1*	c.1444A>G (p.Ile482Val)
1092248267	*ATM*	c.1810C>T (p.Pro604Ser)
1092248267	*ATM*	c.2494C>T (p.Arg832Cys)

1092248268	*MLH1*	c.1565G>A (p.Arg522Gln)
1092248269	*BRCA2*	c.5537T>C (p.Ile1846Thr)
1092248270	*ATM*	c.544G>C (p.Val182Leu)
1092248270	*NBN*	c.797C>T (p.Pro266Leu)
1092248272	*RAD51D*	c.29C>T (p.Pro10Leu)
1093187329	*PALB2*	c.94C>G (p.Leu32Val)
1093187353	*CDKN2A (P16)*	c.430C>T (p.Arg144Cys)
1093187353	*PMS2*	c.1211C>G (p.Pro404Arg)
1093189904	*RAD51D*	c.695G>A (p.Arg232Gln)
1093189905	*CHEK2*	c.538C>T (p.Arg180Cys)
1093189909	*ATM*	c.6067G>A (p.Gly2023Arg)
1093189909	*CDH1*	c.2590G>A (p.Glu864Lys)
1093189910	*CDH1*	c.1223C>T (p.Ala408Val)
1093189911	*MSH6*	dup exon 8
1093189912	*MSH6*	dup exon 8
1093189917	*CHEK2*	c.1091T>C (p.Ile364Thr)
1093189917	*MLH1*	c.482C>T (p.Thr161Met)
1093189917	*MSH6*	c.3802-19_3802-16dupAATA
1093189922	*ATM*	c.544G>C (p.Val182Leu)
1093189922	*MUTYH*	c.1301C>T (p.Thr434Met)
1093189923	*BRIP1*	c.338C>T (p.Thr113Ile)
1093189923	*CDKN2A (P16)*	c.-33G>C
1093189924	*CDKN2A (P16)*	c.430C>T (p.Arg144Cys)
1093189924	*NBN*	c.1489A>G (p.Thr497Ala)
1093189926	*BRIP1*	c.3411_3412delinsCT (p.Asp1138delinsTyr)
1093189926	*CDKN2A (P16)*	c.430C>T (p.Arg144Cys)
1093189933	*ATM*	c.3154-4G>A
1093189934	*CDH1*	c.2440-6C>G
1093189934	*CDKN2A (P16)*	c.-25C>T
1093189939	*BRCA2*	c.6109G>A (p.Glu2037Lys)
1093189939	*BRIP1*	c.1684A>G (p.Ile562Val)
1093189939	*SMAD4*	c.424+5G>A
1093189940	*APC*	c.4336G>A (p.Ala1446Thr)
1093189945	*ATM*	c.6919C>T (p.Leu2307Phe)
1093189946	*ATM*	c.544G>C (p.Val182Leu)
1093189950	*APC*	c.4364A>G (p.Asn1455Ser)
1093189950	*MLH1*	c.83C>T (p.Pro28Leu)
1093189951	*ATM*	c.6572+4T>C
1093189951	*STK11*	c.-1C>T
1093189956	*ATM*	c.6995T>C (p.Leu2332Pro)
1093189962	*CDKN2A (P16)*	c.170C>G (p.Ala57Gly)
1093189962	*CHEK2*	c.922-1G>A
1093189966	*CDK4*	c.122A>G (p.Asn41Ser)
1093189967	*ATM*	c.4388T>G (p.Phe1463Cys)
1093189967	*ATM*	c.1810C>T (p.Pro604Ser)
1093189970	*APC*	c.854A>G (p.Asp285Gly)
1093189971	*BRIP1*	c.728T>C (p.Ile243Thr)
1093189971	*MLH1*	c.53G>C (p.Arg18Pro)
1093189974	*APC*	c.5424_5426del (p.Asn1808del)
1093189974	*CDKN2A (P16)*	c.-33G>C

1093189976	*PMS2*	c.1268C>G (p.Ala423Gly)
1093189977	*BRIP1*	c.584T>C (p.Leu195Pro)
1093189981	*ATM*	c.2401G>A (p.Gly801Ser)
1093189986	*ATM*	c.4424A>G (p.Tyr1475Cys)
1093189992	*RAD51C*	c.376G>A (p.Ala126Thr)
1093189994	*ATM*	c.6114C>G (p.His2038Gln)
1093189997	*BRIP1*	c.2564G>A (p.Arg855His)
1093189999	*ATM*	c.1073A>G (p.Asn358Ser)
1093189999	*ATM*	c.186-7C>T
1093189999	*BRIP1*	c.413T>C (p.Leu138Ser)
1093189999	*NBN*	c.797C>T (p.Pro266Leu)
1093189999	*NBN*	c.37+5G>A
1093189999	*PTEN*	c.-7C>T
1093189999	*RAD51D*	c.695G>A (p.Arg232Gln)
1093190193	*BMPR1A*	c.1420G>C (p.Val474Leu)
1093190196	*BRCA2*	dup exons 23-24
1093190197	*CDH1*	c.833-16C>G
1093190197	*NBN*	c.797C>T (p.Pro266Leu)
1093190201	*APC*	c.2258A>T (p.His753Leu)
1093190201	*CHEK2*	c.246_260del (p.Asp82_Glu86del)
1093190203	*ATM*	c.544G>C (p.Val182Leu)
1093190203	*CDKN2A (P16)*	c.430C>T (p.Arg144Cys)
1093190203	*NBN*	c.2146A>G (p.Asn716Asp)
1093190205	*CHEK2*	c.470T>C (p.Ile157Thr)
1093190206	*ATM*	c.4375G>A (p.Gly1459Arg)
1093190211	*APC*	c.7468G>A (p.Asp2490Asn)
1093190211	*BRIP1*	c.3444C>A (p.Asp1148Glu)
1093190213	*ATM*	c.2396C>T (p.Ala799Val)
1093190220	*BARD1*	c.841C>T (p.Pro281Ser)
1093190223	*STK11*	dup exons 1-9
1093190228	*ATM*	c.370A>G (p.Ile124Val)
1093190228	*NBN*	c.1317A>G (p.Ile439Met)
1093190230	*ATM*	c.6067G>A (p.Gly2023Arg)
1093190232	*ATM*	c.6974C>T (p.Ala2325Val)
1093190232	*CHEK2*	c.1420C>T (p.Arg474Cys)
1093190232	*CHEK2*	c.1260-8A>T
1093190232	*CHEK2*	c.1333T>C (p.Tyr445His)
1093190232	*CHEK2*	c.1525C>T (p.Pro509Ser)
1093190232	*CHEK2*	c.1348G>A (p.Glu450Lys)
1093190232	*MLH1*	c.52C>T (p.Arg18Cys)
1093190232	*RAD51D*	c.577-13C>A
1093190234	*ATM*	c.6067G>A (p.Gly2023Arg)
1093190239	*BARD1*	c.1738G>A (p.Glu580Lys)
1093190245	*BRCA2*	c.6973G>A (p.Val2325Ile)
1093190245	*PALB2*	c.49-6_49-4del
1093190245	*RAD51D*	c.835G>A (p.Asp279Asn)
1093190246	*BRIP1*	c.890A>G (p.Lys297Arg)
1093190246	*STK11*	c.1211C>T (p.Ser404Phe)
1093190251	*CHEK2*	c.1461G>C (p.Gln487His)
1093190252	*BRIP1*	c.3079G>A (p.Glu1027Lys)

1093190257	*NBN*	c.511A>G (p.Ile171Val)
1093190262	*APC*	c.5894A>G (p.His1965Arg)
1093190263	*BRIP1*	c.3746A>G (p.Lys1249Arg)
1093190265	*CDKN2A (P16)*	c.-33G>C
1093190267	*MUTYH*	c.37-7G>A
1093190269	*CDH1*	c.2440-6C>G
1093190269	*SMAD4*	c.1106A>G (p.Asn369Ser)
1093190273	*CDKN2A (P16)*	c.-2G>A
1093190279	*ATM*	c.2096A>G (p.Glu699Gly)
1093190280	*PMS2*	c.572A>G (p.Tyr191Cys)
1093190280	*PTEN*	c.802-51_802-14del
1093190280	*STK11*	c.970C>G (p.Pro324Ala)
1093190284	*ATM*	c.2608A>G (p.Asn870Asp)
1093190284	*ATM*	c.3993+5G>T
1093190284	*NBN*	c.797C>T (p.Pro266Leu)
1093190284	*TP53*	c.139C>T (p.Pro47Ser)
1093190285	*NBN*	c.1882G>A (p.Glu628Lys)
1093190285	*TP53*	c.851C>T (p.Thr284Ile)
1093190286	*ATM*	c.544G>C (p.Val182Leu)
1093190286	*PMS2*	c.2347G>A (p.Val783Ile)
1093190287	*BRCA2*	c.8360G>A (p.Arg2787His)
1095079850	*BRIP1*	c.550G>T (p.Asp184Tyr)
1095079852	*BARD1*	c.33G>T (p.Gln11His)
1095079854	*CDKN2A (P16)*	c.-33G>C
1095079855	*ATM*	c.5267C>G (p.Thr1756Arg)
1095079857	*MUTYH*	c.1037C>T (p.Ser346Leu)
1095079860	*BRIP1*	c.2236A>G (p.Ile746Val)
1095079862	*BRIP1*	c.1961G>T (p.Gly654Val)
1095079865	*RAD51C*	c.890T>C (p.Leu297Pro)
1095079866	*BARD1*	c.33G>T (p.Gln11His)
1095079869	*MSH2*	c.797C>T (p.Ala266Val)
1095079871	*APC*	c.5026A>G (p.Arg1676Gly)
1095079871	*APC*	c.7399C>A (p.Pro2467Thr)
1095079872	*CDH1*	c.88C>A (p.Pro30Thr)
1095079872	*CHEK2*	c.14C>T (p.Ser5Leu)
1095079878	*BRCA2*	c.6413T>A (p.Val2138Asp)
1095079878	*CDKN2A (P16)*	c.146T>C (p.Ile49Thr)
1095079879	*ATM*	c.3961A>G (p.Met1321Val)
1095079889	*ATM*	c.4424A>G (p.Tyr1475Cys)
1095079892	*PALB2*	c.1250C>A (p.Ser417Tyr)
1095079896	*PTEN*	c.802-51_802-14del
1095079899	*APC*	c.7570A>G (p.Lys2524Glu)
1095079904	*PMS2*	c.2108C>T (p.Thr703Met)
1095079907	*BRIP1*	c.890A>G (p.Lys297Arg)
1095079908	*CDKN2A (P16)*	c.-25C>T
1095079908	*MSH2*	c.114C>G (p.Asp38Glu)
1095079908	*PTEN*	c.802-51_802-14del
1095079908	*TP53*	c.139C>T (p.Pro47Ser)
1095079909	*ATM*	c.5071A>C (p.Ser1691Arg)
1095079913	*MLH1*	c.100G>A (p.Glu34Lys)

1095079914	*BMPR1A*	c.1141C>G (p.Leu381Val)
1095079917	*MUTYH*	c.700G>A (p.Val234Met)
1095079933	*APC*	c.2586C>G (p.Asn862Lys)
1095079933	*CDH1*	c.892G>A (p.Ala298Thr)
1095079934	*MSH2*	c.1897A>G (p.Ile633Val)
1095079934	*RAD51C*	c.376G>A (p.Ala126Thr)
1095079935	*NBN*	c.511A>G (p.Ile171Val)
1095079937	*CHEK2*	c.470T>C (p.Ile157Thr)
1095079939	*MSH2*	c.2105T>G (p.Val702Gly)
1095079941	*SMAD4*	c.1573A>G (p.Ile525Val)
1095079944	*CDH1*	c.2635G>A (p.Gly879Ser)
1095111827	*PALB2*	c.2816T>G (p.Leu939Trp)
1095112405	*APC*	dup exon 13
1095112405	*ATM*	dup exon 23
1095112405	*CDKN2A (P14ARF)*	c.217A>C (p.Ser73Arg)
1095112406	*MUTYH*	c.322A>G (p.Lys108Glu)
1095112412	*APC*	c.8276G>A (p.Arg2759His)
1095112412	*TP53*	c.845G>A (p.Arg282Gln)
1095112413	*BRIP1*	c.584T>C (p.Leu195Pro)
1095112417	*MSH6*	c.1180T>G (p.Ser394Ala)
1095112420	*PTEN*	c.235G>A (p.Ala79Thr)
1095112424	*ATM*	c.3787A>G (p.Ser1263Gly)
1095112425	*NBN*	c.37+5G>A
1095112426	*APC*	c.7352C>A (p.Thr2451Asn)
1095112426	*ATM*	c.2804C>T (p.Thr935Met)
1095112426	*SMAD4*	c.455-6A>G
1095112428	*CDK4*	c.409G>A (p.Val137Ile)
1095112430	*CDK4*	c.684-4A>T
1095112437	*APC*	c.8141G>A (p.Arg2714His)
1095112440	*ATM*	c.2932T>C (p.Ser978Pro)
1095112444	*BRIP1*	c.2220G>T (p.Gln740His)
1095112447	*MSH2*	c.1595T>C (p.Val532Ala)
1095112454	*NBN*	c.283G>C (p.Asp95His)
1095112458	*NBN*	c.511A>G (p.Ile171Val)
1095112461	*PALB2*	c.3307G>A (p.Val1103Met)
1095112463	*BRIP1*	c.1655T>C (p.Ile552Thr)
1095112463	*PALB2*	c.3191A>T (p.Tyr1064Phe)
1095112466	*APC*	c.848G>A (p.Arg283Gln)
1095112468	*ATM*	c.5653A>G (p.Thr1885Ala)
1095112471	*APC*	c.3161A>C (p.His1054Pro)
1095112475	*PALB2*	c.-5G>T
1095112476	*ATM*	c.8268+6T>A
1095112479	*CHEK2*	c.1607C>T (p.Pro536Leu)
1095112479	*PMS2*	c.1247C>G (p.Ser416Cys)
1095112480	*BRIP1*	c.430G>A (p.Ala144Thr)
1095112484	*BRIP1*	c.890A>G (p.Lys297Arg)
1095112485	*MSH6*	c.261-14C>A
1095112485	*MUTYH*	c.1255G>A (p.Ala419Thr)
1095112488	*APC*	c.3535T>A (p.Tyr1179Asn)
1095112488	*BMPR1A*	c.1433G>A (p.Arg478His)

1095112488	*NBN*	c.37+5G>A
1095112491	*ATM*	c.1229T>C (p.Val410Ala)
1095112492	*CHEK2*	c.1217G>A (p.Arg406His)
1095112493	*ATM*	c.3137T>C (p.Leu1046Pro)
1095112496	*BARD1*	c.1409A>G (p.Asn470Ser)
1095188497	*PTEN*	c.802-51_802-14del
1095188500	*CHEK2*	c.1525C>T (p.Pro509Ser)
1095188501	*BRIP1*	c.584T>C (p.Leu195Pro)
1095188508	*ATM*	c.6995T>C (p.Leu2332Pro)
1095188508	*TP53*	c.139C>T (p.Pro47Ser)
1095188510	*APC*	c.6512G>A (p.Gly2171Glu)
1095188512	*PMS2*	c.883C>T (p.Arg295Trp)
1095188513	*BRIP1*	c.139C>G (p.Pro47Ala)
1095188515	*MUTYH*	c.925C>T (p.Arg309Cys)
1095188515	*PALB2*	c.2674G>A (p.Glu892Lys)
1095188516	*TP53*	c.1150A>G (p.Met384Val)
1095188519	*PMS2*	c.1420G>T (p.Ala474Ser)
1095188520	*ATM*	c.5071A>C (p.Ser1691Arg)
1095188521	*RAD51D*	c.568G>A (p.Ala190Thr)
1095188521	*RAD51D*	c.481-7G>A
1095188532	*APC*	c.4088A>G (p.Lys1363Arg)
1095188541	*ATM*	c.186-7C>T
1095188541	*BRIP1*	c.2236A>G (p.Ile746Val)
1095188546	*APC*	c.2204C>T (p.Ala735Val)
1095188546	*ATM*	c.496+4T>C
1095188546	*MUTYH*	c.1276C>T (p.Arg426Cys)
1095188546	*RAD51C*	c.790G>A (p.Gly264Ser)
1095188547	*MSH6*	c.124C>T (p.Pro42Ser)
1095188547	*MSH6*	c.1932G>C (p.Arg644Ser)
1095188547	*RAD51D*	c.146C>T (p.Ala49Val)
1095188550	*STK11*	c.1283C>G (p.Ser428Trp)
1095188551	*CDH1*	c.88C>A (p.Pro30Thr)
1095188556	*BARD1*	c.709C>G (p.Gln237Glu)
1095188564	*ATM*	c.1744T>C (p.Phe582Leu)
1095188566	*MSH2*	c.490G>A (p.Gly164Arg)
1095188574	*ATM*	c.1810C>T (p.Pro604Ser)
1095188579	*ATM*	c.131A>T (p.Asp44Val)
1095188581	*STK11*	c.1211C>T (p.Ser404Phe)
1095188586	*ATM*	c.6988C>G (p.Leu2330Val)
1095188588	*BMPR1A*	c.1355A>G (p.Glu452Gly)
1095188588	*PALB2*	c.194C>T (p.Pro65Leu)
1095214770	*CDKN2A (P16)*	c.-33G>C
1095214770	*MLH1*	c.-43C>T
1095214818	*TP53*	c.97-6C>T
1095216809	*PMS2*	c.2174C>T (p.Ala725Val)
1095216810	*CHEK2*	c.410G>A (p.Arg137Gln)
1095216811	*MLH1*	c.1937A>G (p.Tyr646Cys)
1095216813	*NBN*	c.2146A>G (p.Asn716Asp)
1095216822	*CDKN2A (P16)*	c.206A>G (p.Glu69Gly)
1095216822	*CHEK2*	c.14C>T (p.Ser5Leu)

1095216825	*BRIP1*	c.890A>G (p.Lys297Arg)
1095216826	*PMS2*	c.163+4A>G
1095216830	*APC*	c.6236A>C (p.Asp2079Ala)
1095216830	*BRIP1*	c.2285G>A (p.Arg762His)
1095216833	*ATM*	c.7592T>C (p.Met2531Thr)
1095216833	*BARD1*	c.2282G>A (p.Ser761Asn)
1095216836	*BMPR1A*	c.1432C>T (p.Arg478Cys)
1095216850	*NBN*	c.2146A>G (p.Asn716Asp)
1095216850	*PMS2*	c.1556A>G (p.Tyr519Cys)
1095216850	*PMS2*	c.1559C>T (p.Ala520Val)
1095216851	*NBN*	c.832T>G (p.Ser278Ala)
1095216852	*NBN*	c.1882G>A (p.Glu628Lys)
1095216871	*ATM*	c.7912T>G (p.Trp2638Gly)
1095216871	*BRIP1*	c.430G>A (p.Ala144Thr)
1095216871	*MUTYH*	c.74G>A (p.Gly25Asp)
1095216871	*MUTYH*	c.53C>T (p.Pro18Leu)
1095216875	*APC*	c.4072G>A (p.Ala1358Thr)
1095216879	*ATM*	c.6067G>A (p.Gly2023Arg)
1095216882	*MSH2*	c.1760-7T>C
1095216883	*BRIP1*	c.790C>T (p.Arg264Trp)
1095216895	*CDH1*	c.1234G>A (p.Val412Ile)
1095216897	*RAD51C*	c.376G>A (p.Ala126Thr)
1095216899	*BARD1*	c.1738G>A (p.Glu580Lys)
1095216899	*NBN*	c.2146A>G (p.Asn716Asp)
1095216899	*PTEN*	c.802-51_802-14del
1097148993	*ATM*	c.7522G>A (p.Gly2508Arg)
1097148993	*CDH1*	c.2635G>A (p.Gly879Ser)
1097148999	*PMS2*	c.857A>G (p.Asp286Gly)
1097149000	*APC*	c.2222A>G (p.Asn741Ser)
1097149002	*ATM*	c.2289T>A (p.Phe763Leu)
1097149005	*MLH1*	c.739T>C (p.Ser247Pro)
1097149006	*CDH1*	c.2512A>G (p.Ser838Gly)
1097149006	*PALB2*	c.2903C>G (p.Ala968Gly)
1097149010	*ATM*	c.1810C>T (p.Pro604Ser)
1097149010	*ATM*	c.2494C>T (p.Arg832Cys)
1097149010	*RAD51D*	c.695G>A (p.Arg232Gln)
1097149011	*NBN*	c.456G>A (p.Met152Ile)
1097149013	*ATM*	c.6067G>A (p.Gly2023Arg)
1097149014	*APC*	c.7808A>G (p.Glu2603Gly)
1097149016	*BRIP1*	c.550G>T (p.Asp184Tyr)
1097149017	*TP53*	c.139C>T (p.Pro47Ser)
1097149018	*TP53*	c.97-6C>T
1097149019	*BRIP1*	c.3262C>T (p.His1088Tyr)
1097149019	*CHEK2*	c.470T>C (p.Ile157Thr)
1097149020	*MSH6*	c.1822A>G (p.Ile608Val)
1097149025	*BRCA2*	c.3581G>A (p.Gly1194Asp)
1097149030	*APC*	c.3323A>G (p.Asn1108Ser)
1097149030	*RAD51D*	c.629C>T (p.Ala210Val)
1097149031	*STK11*	c.336G>C (p.Gln112His)
1097149032	*BARD1*	c.668A>G (p.Glu223Gly)

1097149034	*ATM*	c.186-7C>T
1097149034	*BRIP1*	c.3378A>C (p.Glu1126Asp)
1097149043	*BMPR1A*	c.499A>G (p.Met167Val)
1097149045	*PALB2*	c.400G>A (p.Asp134Asn)
1097149046	*MSH6*	c.3758T>C (p.Val1253Ala)
1097149047	*APC*	c.6985A>G (p.Ile2329Val)
1097149048	*BARD1*	c.1738G>A (p.Glu580Lys)
1097149052	*BMPR1A*	c.1327C>T (p.Arg443Cys)
1097149054	*MSH6*	c.2926C>T (p.Arg976Cys)
1097149054	*NBN*	c.1489A>G (p.Thr497Ala)
1097149054	*PTEN*	c.802-51_802-14del
1097149055	*MSH6*	c.949A>G (p.Lys317Glu)
1097149056	*APC*	c.835G>T (p.Gly279Cys)
1097149056	*ATM*	c.1229T>C (p.Val410Ala)
1097149056	*BRIP1*	c.10A>G (p.Met4Val)
1097149056	*PALB2*	c.2027T>C (p.Ile676Thr)
1097149057	*ATM*	c.4414T>G (p.Leu1472Val)
1097149057	*BRCA2*	c.9442G>T (p.Ala3148Ser)
1097149059	*BMPR1A*	c.1439G>T (p.Arg480Leu)
1097149060	*BRIP1*	c.2236A>G (p.Ile746Val)
1097149060	*NBN*	c.797C>T (p.Pro266Leu)
1097149061	*ATM*	c.1744T>C (p.Phe582Leu)
1097149062	*PTEN*	c.349A>C (p.Asn117His)
1097149065	*PALB2*	c.821C>T (p.Thr274Ile)
1097149066	*BRIP1*	c.550G>T (p.Asp184Tyr)
1097149066	*TP53*	c.704A>G (p.Asn235Ser)
1097149067	*CHEK2*	c.538C>T (p.Arg180Cys)
1097149070	*CDH1*	c.892G>A (p.Ala298Thr)
1097149070	*MLH1*	c.1637A>G (p.Lys546Arg)
1097149072	*PALB2*	c.2201C>A (p.Thr734Asn)
1097149074	*BRIP1*	c.370A>G (p.Thr124Ala)
1097149083	*CDKN2A (P16)*	c.-2G>A
1097149085	*BRCA1*	c.655G>A (p.Asp219Asn)
1097149086	*MLH1*	c.1761G>A (p.Met587Ile)
1097149086	*NBN*	c.1489A>G (p.Thr497Ala)
1097162627	*PALB2*	c.653A>T (p.Glu218Val)
1097162628	*ATM*	c.2362A>C (p.Ser788Arg)
1097162628	*ATM*	c.6088A>G (p.Ile2030Val)
1097162633	*ATM*	c.544G>C (p.Val182Leu)
1097162633	*NBN*	c.797C>T (p.Pro266Leu)
1097162638	*MUTYH*	c.998-9C>T
1097162639	*ATM*	c.6919C>T (p.Leu2307Phe)
1097162639	*RAD51C*	c.146-8A>G
1097162641	*APC*	c.277C>G (p.Leu93Val)
1097162643	*ATM*	c.8968G>A (p.Glu2990Lys)
1097162646	*MSH6*	c.3762_3764del (p.Glu1254del)
1097162647	*ATM*	c.3077+4G>A
1097162647	*ATM*	c.544G>C (p.Val182Leu)
1097162650	*PALB2*	c.2135C>T (p.Ala712Val)
1097162653	*BMPR1A*	c.1204G>T (p.Val402Leu)

1097162653	*MUTYH*	c.56G>A (p.Arg19Gln)
1097162655	*BRIP1*	c.752G>A (p.Arg251His)
1097162656	*BMPR1A*	c.1327C>T (p.Arg443Cys)
1097162656	*CDKN2A (P16)*	c.-25C>T
1097162661	*CHEK2*	c.1076A>G (p.Glu359Gly)
1097162662	*ATM*	c.5071A>C (p.Ser1691Arg)
1097162668	*BARD1*	c.2191C>T (p.Arg731Cys)
1097162668	*BARD1*	c.1601C>T (p.Thr534Ile)
1097162668	*MSH2*	c.1690A>G (p.Thr564Ala)
1097162669	*MSH6*	c.3731T>C (p.Leu1244Ser)
1097162670	*RAD51C*	c.376G>A (p.Ala126Thr)
1097162672	*BRIP1*	c.1000G>T (p.Ala334Ser)
1097162675	*MLH1*	c.1649T>C (p.Leu550Pro)
1097162677	*MSH6*	c.3190G>C (p.Ala1064Pro)
1097162677	*TP53*	c.139C>T (p.Pro47Ser)
1097162678	*PALB2*	c.2135C>T (p.Ala712Val)
1097162680	*ATM*	c.1810C>T (p.Pro604Ser)
1097162680	*ATM*	c.4388T>G (p.Phe1463Cys)
1097162684	*ATM*	c.1229T>C (p.Val410Ala)
1097162686	*NBN*	c.2146A>G (p.Asn716Asp)
1097162686	*STK11*	c.894C>A (p.Phe298Leu)
1097162687	*BRCA2*	c.5270A>G (p.Tyr1757Cys)
1097162691	*BRCA2*	c.5284T>G (p.Tyr1762Asp)
1097162695	*MUTYH*	c.1417G>A (p.Ala473Thr)
1097162696	*APC*	c.6068G>T (p.Arg2023Ile)
1097162699	*ATM*	c.4091A>G (p.Asp1364Gly)
1097162699	*CHEK2*	c.320-5T>A
1097162700	*ATM*	c.3925G>A (p.Ala1309Thr)
1097162704	*PTEN*	c.802-51_802-14del
1097162709	*MSH2*	c.2211-5T>G
1097162710	*ATM*	c.6067G>A (p.Gly2023Arg)
1097162712	*APC*	c.4018_4020dupTCT (p.Ser1341dup)
1097162713	*CDH1*	c.2104G>A (p.Glu702Lys)
1097162716	*MUTYH*	c.481G>C (p.Asp161His)
1097162717	*ATM*	c.5890A>G (p.Lys1964Glu)