Supplementary Online Content

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This supplementary material has been provided by the authors to give readers additional information about their work.

eAppendix 1. Pancreatic Cancer Patient Research Registry

The Mayo Clinic Biospecimen Resource for Pancreas Research is an ongoing patient registry, established in 2000, under an Institutional Review Board approved protocol. We employ an "ultra-rapid" case ascertainment method to prospectively recruit research participants, which denotes identification of potential research participants at the time of first visit to Mayo Clinic, often prior to a confirmed diagnosis of pancreatic cancer. Support for the resource has been continuous from both Mayo Clinic intramural funds and the NCI-funded Mayo Clinic SPORE in Pancreatic Cancer (P50 CA102701).

All patients being evaluated for pancreatic mass or pancreatic cancer at Mayo Clinic campuses (Rochester, MN; Jacksonville, FL; or Phoenix, AZ) are eligible for participation in the Resource. Potential participants are identified by electronic search of appointment schedules and are recruited from Gastroenterology, Oncology, and Surgery clinics. Patients are usually invited in person by a study coordinator to learn about and participate in the registry protocol (~34% are approached by mail). Participation consists of completing of a consent form, risk factor and family history surveys, providing a 50 ml venous blood sample (typically 40 ml EDTA, 10 ml heparinized, and 10 ml without anti-coagulant) during the clinic visit, and authorizing study team access to medical records. The risk factor survey collected information on race and ethnicity. Race was self-identified by checking categories (American Indian/Alaskan Native; Asian/Asian American; Black/African-American, Native Hawaiian/Other Pacific Islander; White Caucasian; Multiracial (specify)). Similarly, ethnic group was self-identified by checking "Hispanic/Latino" or "Non-Hispanic/Non-Latino." Dr. Gloria Petersen, an experienced genetic epidemiologist trained in Medical Genetics and who is an American Board of Medical Genetics and Genomics -certified Ph.D. Medical Geneticist, designed the questionnaire to carefully obtain family history details in a standardized fashion. Detailed family history structure including third degree relatives was obtained and pedigrees are generated. All participant pedigrees are reviewed personally by Dr. Petersen and her study team on a semi-monthly basis to ensure that family histories obtained are consistent, with followup to the participant/family for clarification as needed. If the patient is unwilling to provide a blood sample, saliva samples are sought for genomic DNA extraction. Participants are also asked to complete follow-up surveys at 6 and 12 months. Upon returning a signed consent, participants recruited by mail complete surveys and blood sampling kits are used for biospecimen collection at their convenience in their home town or city. If components of the protocol are incomplete, reminder letters are mailed to all participants or potential participants 3 weeks and 6 weeks after the initial invitation. If there is no further response, participants are designated as non-responders.

Participant tracking, clinical data, biospecimen data, and surveys are maintained in a secure, password-protected custom Sybase database, accessible to authorized study personnel. All surveys are scanned or double entered, with built-in error checks. Medical records from clinical providers outside of Mayo Clinic are requested. Data related to health history, diagnosis, and outcomes are obtained from authorized medical records and coded into the database. Records are manually abstracted using a standardized protocol by a gastrointestinal cancer specialist/medical oncologist. Abstracted information includes type of chemotherapy and/or radiotherapy, disease staging, response to treatment and duration of therapy, and common toxicities. Records are considered as "complete" when medical record documentation about the number, type, and duration of all therapies, with documentation of cessation of therapy has been abstracted.

The study sample consisted of participants who were recruited from October 12, 2000 through March 31, 2016. In this period, we approached 6,074 patients with pancreatic adenocarcinoma. Of these, 3985 (65.6%) provided informed consent, and 3210 provided research blood specimens, including 3030 patients constituting the analytic sample. Among those who did not participate in the registry, 633 declined to participate, whereas 1448 did not respond to our invitation to be in the study (it is our experience that the majority of these participants are very ill or coping with the diagnosis of a lethal cancer, and could not provide a stated response to the research invitation); eight participants consented to be in the registry but did not provide a research blood specimen. Approximately 85.5% (2,591) of the 3,030 participants in the analytic sample were consented to

participate in the registry within 30 days of their diagnosis, with 64.7% (1,961) consented within two days. The study sample is 43.1% female, and the racial/ethnic composition is 96.6% White, 2.2% African American, 0.36% Native American, 0.4% Asian, and 0.1% Native Hawaiian/Pacific Islander; and 1.4% Hispanic ethnicity. This composition is congruent with the race and ethnicity classifications (African/African American; Hispanic, East Asian and South Asian, and Non-Hispanic White) used in the study. These classifications were used in order to match principal component derived classifications of reference controls in gnomAD. The racial/ethnic composition of the study is similar to the U.S. Census Bureau American Community Survey (2011-2015)(https://www.census.gov/programs-surveys/acs/data.html) for the Midwest region around Mayo Clinic, which indicates that the population is 50% female; 92.6% White, 1.9% African American, 0.42% Native American, 2.5% Asian, and 0.03% Native Hawaiian/Pacific Islander. Around 4% are Hispanic ethnicity.

The biospecimen resource adheres to the principles and practices recommended by the NCI Best Practice for Biospecimen Resources (https://biospecimens.cancer.gov/practices/). Mayo Clinic has highly efficient systems developed for handling biospecimens for clinical analysis, and these are adapted for research studies. Date and time stamps, and prandial status are recorded for the specimens. Blood samples drawn at Mayo Clinic are processed within 4 hours at the institutionally-supported Biospecimens Accessioning and Processing (BAP) core lab (http://www.mayo.edu/research/centers-programs/cancer-research/shared-resources-core-facilities-services/biospecimens-accessioning-processing). Blood samples drawn outside of Mayo Clinic are expressed shipped directly to the BAP lab and processed, typically within 24 hours. From a 50 ml research blood sample, 4 to 5 aliquots of 1.5 ml serum, 4 to 5 aliquots of 1.5 ml plasma, two buffy coats, and genomic DNA extracted from another buffy coat sample are stored. Samples are all processed using robotic technology by standardized methods and stored rapidly in a Biostore (NEXUS) -80°C freezer and liquid nitrogen tanks (-120^{o}C) until needed.

Genomic DNA derived from blood samples is stored for use in future research projects. Germline genetic testing is explicitly mentioned in the consent form; patients are aware this is a possible research analysis to be performed on their samples. The research registry had medical record reports of 41 participants who had received clinical genetic testing (unrelated to our research). These were presumably disclosed by a genetics professional. Among these 41, eleven had a positive genetic test result, and those results were congruent with the testing performed by the research laboratory in this report. Among the participants who had genetic analysis performed by research protocol, all were deceased, so no test results were disclosed.

A subset of pancreatic cancer cases reported in this study were previously subjected to germline genetic testing. A total of 341 patients with a family history of pancreatic cancer from the Mayo Clinic research registry, including 309 from the current study, were tested for germline pathogenic mutations in *BRCA1*, *BRCA2*, *PALB2*, and *CDKN2A*, to assess the prevalence of mutations in these genes in the high-risk setting[1]. A separate study evaluated the prevalence of pathogenic mutations among 25 cancer susceptibility genes on the “MyRisk” panel in 302 family history positive patients, including 273 from the current study[2]. Whole genome sequencing of germline DNA from 187 patients with a family history of pancreatic cancer, including 155 from the current study, was conducted to discover novel pancreatic cancer susceptibility genes[3]. In addition, 1537 randomly selected patients with pancreatic cancer that were collected from 10/12/2000-2/18/2009, including 1512 from the current study, were tested for germline pathogenic *CDKN2A* mutations[4]. Finally, 96 patients sequentially enrolled between 9/5/2013 and 6/17/2014, including 95 from the current study, were tested for pathogenic mutations in 22 genes as a pilot study to estimate the frequency of mutations in these genes in a sequential series not specifically selected for family history[5]. In total, only 96 sequentially recruited and 302 with a family history of pancreatic cancer were screened for a varying number of genes, leaving 2639 patients in the current study that never had a full panel of cancer susceptibility gene mutation testing.

eAppendix 2. Reference Controls

gnomAD reference controls

The Genome Aggregation Database (gnomAD) contains sequencing data with 123136 exomes and 15496 genomes from unrelated individuals sequenced as part of various disease-specific and population genetic studies. The raw sequence data was reprocessed through the same pipeline, and jointly variant-called to increase consistency across projects. The gnomAD data set contains individuals sequenced using multiple exome capture methods and sequencing chemistries. The resulting variation in coverage is incorporated into the variant frequency calculations for each variant. GnomAD was quality controlled and analyzed using the Hail open-source framework for scalable genetic analysis. GnomAD provides allele frequencies separately for several races and ethnic groups including non-Finnish European (NFE), which excludes Ashkenazi Jewish and Finnish European individuals; African/African American (AFR); admixed American (AMR); East Asian (EAS); and South Asian (SAS). While the gnomAD dataset overlaps with approximately 30000 common ExAC_non_TCGA controls, the substantially increased number of gnomAD controls along with updated variant calling algorithms over ExAC identified gnomAD as a valuable alternative reference control dataset.

Exome Aggregation Consortium (ExAC) reference controls

The Exome Aggregation Consortium (ExAC) contains exome sequence data from 60706 unrelated individuals sequenced as part of various disease-specific and population genetic studies. All of the raw data from these projects was reprocessed through a common pipeline. Principal component analysis (PCA) was performed to identify population clusters corresponding to individuals of European, African, South Asian, East Asian, and admixed American. Europeans were separated into individuals of Finnish and non-Finnish ancestry (NFE). ExAC also contained cancer cases from The Cancer Genome Atlas (TCGA). Exclusion of sequence data from the TCGA cases yielded ExAC_ non_TCGA (the cancer genome atlas project) reference controls. In addition, reference controls of European non-Finnish ancestry excluding TCGA cases (ExAC_NFE_non-TCGA) were used for association studies involving non-hispanic white cases.

eAppendix 3. Custom Sequencing of Mayo Clinic Pancreatic Cancer Cases

A QIAseq custom panel of 1733 primers accounting for all coding regions and consensus splice sites from 37 cancer genes potentially involved in germline susceptibility to cancer and 126 breast cancer associated SNPs was designed for germline genetic testing of patients for inherited mutations in these predisposition genes. For this study, the focus was on results from 21 cancer predisposition genes (eTable 1) commonly found on other commercial hereditary cancer genetic testing panels for common cancer including breast, ovarian, colorectal, endometrial, and pancreatic cancer[6-9]. The QIAseq protocol was optimized for high-throughput robotic processing of DNA samples. Two pilot projects were conducted to assess the quality of multiplex PCR and sequencing using the custom panel.

The first pilot study involved 48 DNA samples extracted from blood specimens with known mutations in several genes on the panel including two large genomic rearrangements in *BRCA1*. DNA samples were amplified with the 1300 primers in two 24 sample batches using dual barcoding. Individual amplicon pools were evaluated by quantitative PCR, eGel, and Qubit analysis, pooled, and subjected to sequencing on the MiSeq. Informatics analysis was performed as described below and was blinded to mutation status of samples and mutation descriptions. All samples sequenced equivalently with on average 75% of reads mapping to target sequences. In addition, >96% of targets had >400 reads in all samples. All 48 mutations were identified including the *BRCA1* rearrangements. No false positive known or likely pathogenic mutations were identified. In a second pilot study, 48 samples from patients with pancreatic cancer, in two batches of 24, were selected based on results from prior genetic testing[1-3,5] and were subjected to multiplex PCR using the custom QIAseq panel followed by sequencing on a HiSeq4000. >99% of targets had >20 reads in all samples. Informatics analysis was blinded to mutation content. All pre-selected mutations were identified. On the basis of these studies, the custom QIAseq panel was considered validated for analysis of DNA samples from the sequential series of patients with pancreatic cancer.

A total of 3046 genomic DNA samples extracted from peripheral blood lymphocyte samples were processed to prepare DNA libraries for Next generation sequencing. Libraries were generated using QIAseq custom panel. One sample failed library preparation. Sample libraries were sequenced in pools of 768 per lane of a HiSeq4000 (Illumina) with 150bp paired-end reads corresponding to a median coverage of 200X.

eAppendix 4. Bioinformatics Analysis of Sequencing Data

Sequencing adapters, gene-specific primers, and the QIASeq common sequencing element were removed from the sequence data as well as hard trimming the first 24bp from each read[10]. Reads were aligned with bwa-mem[11]. Realignment, recalibration, Haplotype calling, Depth of Coverage, and GenotypeGVCF walkers were run from GATK v3.4-46[12].

Nucleotide reads of greater than 20X was set as the Quality Control (QC) threshold for coverage. A total of 3041 samples (99.8%) had sequencing coverage above 20X for >90% of target nucleotides. Samples were excluded if 90% of the target regions were not covered at or above 90X, due to high levels of homozygosity, or if identity by descent was suggestive of cryptic relationships.

Data analysis was performed for the resulting 3030 samples. Copy number variations were detected with PatternCNV v1.1.3[13]. Annotations were provided through the BioR toolkit[14] leveraging dbNSFP v3.0[15], ClinVar[16], CAVA[17], and population frequencies from ExAC[18] (but with TCGA samples removed). Variants were viewed and filtered with VCF-Miner[19]. Bam files of classified pathogenic variants were viewed by IGV. All loss of function variants (nonsense, frameshift, consensus splice sites (+/-1 or 2), and any intronic or missense variants defined as pathogenic or likely pathogenic in ClinVar by two or more clinical laboratories (Ambry Genetics, SCRP, InVitae, GeneDX, Counsyl, and InSiGHT) were validated by Sanger sequencing. All suspected mosaic somatic variants (allele ratio>80:20), and truncating variants in the last 55bp of the penultimate exon or last exon that potentially avoid nonsense mediated mRNA decay and do not influence known functional domains were excluded. Similarly variants located after established cutoffs for protein function (e.g. *BRCA2* p.Tyr3208X) were excluded. Variants reported with reduced penetrance (e.g. *CHEK2* c.Ile157Thr), and variants with minor allele frequency (MAF)>0.3%, other than common founder mutations (e.g. *CHEK2* c.1100delC) were excluded.

eAppendix 5. Cleaning and Filtering of Reference Control Sequencing Data

gnomAD data cleaning and filtering:

- Restricted to gnomAD exome data
- Deleterious variant classification rules:
    1. Restricted to variants with AF<0.003, except known deleterious founder variants (e.g. *CHEK2* c.1100delC)
    2. Include LOF (loss-of-function) variants (nonsense, frameshift, +/-1,2 splice) unless Clinvar classified as benign or VUS in majority of clinical cancer genetics testing laboratories (Ambry, SCRP, InVitae, GeneDx, Emory and InSiGHT)
    3. Exclude missense variants and +/-≥3 splice unless Clinvar classified as pathogenic or likely pathogenic by two or more of aforementioned clinical genetics groups.
    4. Exclude deleterious variants with known low risk: *PMS2* c.736_741del6ins11, *TP53* p.Arg283His, 5'UTR_EX1del, p.Arg181His, p.Arg156His, CHEK2 p.Ile157Thr.
    5. Exclude deleterious variants not influenced by Nonsense mediated RNA decay (NMD) (Thresholds: *BRCA2* c.9924; *BARD1* c.1947, *BRIP1* c.2851, *RAD51D* c.849)
    6. Mark variants in *PMS2* pseudogene region (Exon9 and exon 11-15), calculate variant frequency and odds ratios without these variants.
    7. Review variants with Allele Count (AC) ≥15 by IGV and by frequency in control data from dbSNP.
    8. Stratify by populations: AFR, AMR, EAS, NFE, and SAS
- AN (allele number) was calculated as average of all variants within the coding region of a gene of interest.

ExAC data cleaning and filtering:

- Restricted to ExAC_non_TCGA exome data
- Deleterious variant classification rules:
    - Same as in gnomAD rules 1-6
    - Exclude ExAC non-PASS recurrent variants with allele count in ExAC>8 and tested in <20000 ExAC alleles
    - Exclude ExAC non-PASS variants with multiple repetitive sequences called multiple times. Example: *MSH2*_c.942+2_942+6del5, *MSH2*_c.942+2_942+4delTAA, MSH2_c.942+2_942+5delTAAA, *MSH2*_c.942+2_942+3delTA, MSH2_c.942+2_942+8del7 *MSH2*_c.942+2_942+7del6.
- Stratify by populations: AFR, AMR, EAS, NFE, SAS
- AN (allele number) was calculated as average of all variants within the coding region of a gene of interest.

eAppendix 6. Statistical Analysis

Association analysis: For the overall analysis of gene specific odds ratios, the relative frequency of the race and ethnicity populations in the pancreatic cancer cases was estimated. The corresponding gnomAD reference control populations were weighted such that the relative population frequency was the same in the cases and weighted controls. Weighted logistic regression was used to estimate the odds ratio of an association between pathogenic mutations within a gene and pancreatic cancer. Confidence intervals were estimated by the profile likelihood method. All analyses were performed in R (version 4.3.2). All tests were two-sided.

For comparisons within individual populations, gene specific odds ratios for each of the genes of interest with mutations were estimated based on the combined rare, pathogenic allele count across the gene relative to the number of alleles tested (2-fold the number of individuals tested). Confidence intervals for the odds ratios were estimated based on inverting the Fisher's exact test[20]. The method to find the shortest continuous interval consistent with the Fisher's exact test statistical significance level guarantees the p-value from the test with be in agreement with the corresponding confidence interval not including the null value within the interior of the interval. The confidence intervals were generated in R (version 4.3.2) using the *exact2x2* package[21]. To test the hypothesis that age of diagnosis of pancreatic cancer differs between mutation carriers and non-carriers, the cumulative distribution of the age of diagnosis within each subgroup and the Kolmogorov-Smirnov test was used. The test allows for the identification of any differences in the age of diagnosis distribution and was therefore considered more powerful than only testing a shift in the average age of diagnosis between carriers and non-carriers.

Associations between mutations in each gene and patient clinical characteristics were assessed using logistic regression within the pancreatic cancer cases. The odds a patient with pancreatic cancer is a mutation carrier was modeled as a function of the clinical characteristic, e.g. family history of pancreatic cancer, adjusting for the age of cancer diagnosis. The coefficient in the regression model is interpreted as an adjusted enrichment parameter, and can be used to identify subsets of patients with a higher likelihood of having a mutation in a specific gene. This is referred to as a case-case analysis since it compares pancreatic cancer cases with mutations to pancreatic cancer cases without mutations. The patient characteristics tested were personal history of any prior cancer, family history of pancreatic cancer, family history of breast cancer, and family history of colorectal cancer.

Survival analysis: The patient population for survival analysis was restricted to the subset of 2698 adenocarcinoma cases recruited at Mayo Clinic within 3 months (≤ 92 days) of an initial diagnosis. Duration of overall survival was calculated from the date of pancreatic cancer diagnosis at Mayo Clinic until the earliest of the following: death date, or last known alive date. Updates to patient vital status and the corresponding date were obtained from multiple institutional resources, and personal/family correspondence. Death information was confirmed via external vital statistics database services. Patients not known to be deceased at the time of this analysis were censored (vital status coded as not deceased) with overall survival duration calculated using the date they were last known to be alive. Date of pancreatic cancer diagnosis was defined as the date of pathological diagnosis (97%), or clinical diagnosis (3%) for those without pathology.

The primary survival analysis was a comparison of outcome for carriers versus non-carriers of pathogenic mutations in the set of 6 genes statistically significantly associated with pancreatic cancer (ATM, BRCA1, BRCA2, CDKN2A, MLH1, TP53). Copy number variants within these genes were included in the analysis. Since the number of mutation carriers within each gene was relatively small, the pre-defined analysis plan involved pooling all carriers of mutations in the genes into a single group. Median survival for mutation carriers and non-carriers was estimated using Kaplan-Meier methods, with log-rank tests used to test for significance in the survival distribution. Multivariable Cox models adjusting for age at diagnosis, gender and pancreas cancer stage (Surgically Resectable, Locally Advanced, Metastatic) were also considered, with Likelihood Ratio

tests used to test for statistical significance of the carrier status on the survival hazard. Follow-up was estimated using the Kaplan-Meier method for the censoring distribution[22].

eTable 1. Genes in the Custom Qiaseq Panel Evaluated for Presence of Mutations in Patients With Pancreatic Cancer

Study Genes[a]	Gene	refseq	chr	Molecular function of encoded protein[b]
1	*ATM*	NM_000051.3	11	A cell cycle checkpoint kinase that phosphorylates Tp53 and many other targets and is required for the cellular response to DNA damage and genome instability
1	*BARD1*	NM_000465.3	2	Interacts with the N-terminal region of BRCA1 and plays a central role in the control of cell cycle in response to DNA damage
1	*BRCA1*	NM_007294.3	17	A nuclear phosphoprotein that plays a role in maintaining genome stability, acts as a tumor suppressor, and plays a role in transcription, DNA repair of double-stranded breaks, and recombination
1	*BRCA2*	NM_000059.3	13	Involved in maintenance of genome stability through homologous recombination repair of double-stranded DNA breaks
1	*BRIP1*	NM_032043.2	17	Interacts with the BRCT repeats of BRCA1 and influences the normal double-strand break repair function of BRCA1
1	*CDH1*	NM_004360.4	16	A classical member of the cadherin superfamily; loss of function contributes to cancer progression by reducing cell-cell contact and increasing proliferation, invasion, and/or metastasis
1	*CDKN2A*	NM_000077.4	9	Regulates G1 phase of the cell cycle and progression to S phase through CDK4 and Tp53
1	*CHEK2*	NM_007194.3	22	A tumor suppressor that phosphorylates BRCA1 and other DNA repair and cell cycle regulators
1	*FANCC*	NM_000136.2	9	Belongs to Fanconi anemia complementation group (FANC) C, Fanconi anemia is a genetically heterogeneous recessive disorder characterized by cytogenetic instability, hypersensitivity to DNA crosslinking agents, increased chromosomal breakage, and defective DNA repair.
1	*MLH1*	NM_000249.3	3	Hetero-dimerizes with mismatch repair endonuclease PMS2 to form MutL alpha, part of the DNA mismatch repair system
1	*MRE11A*	NM_005591.3	11	A nuclear protein involved in homologous recombination, telomere length maintenance, and DNA double-strand break repair; forms a complex with the RAD50 and NBN proteins.
1	*MSH2*	NM_000251.2	2	A human homolog of the E.coli mismatch repair gene mutS, that plays an essential role in DNA mismatch repair (MMR). Implicated in hereditary nonpolyposis colon cancer (HNPCC)
1	*MSH6*	NM_000179.2	2	Plays an essential role in repairing DNA; helps fix errors during DNA replication in preparation for cell division; joins with MSH2 to form a protein complex that identifies errors in DNA during replication
1	*NBN*	NM_002485.4	8	A member of the MRE11/RAD50/NBN double-strand break repair complex; involved in DNA double-strand break repair and DNA damage-induced checkpoint activation
1	*NF1*	NM_001042492.2	17	Encoded protein Neurofibromin acts as a tumor suppressor and functions as a negative regulator of the ras signal transduction pathway
1	*PALB2*	NM_024675.3	16	Binds to and co-localizes with BRCA2 in nuclear foci and likely permits the stable intra-nuclear localization and accumulation of BRCA2
1	*PMS2*	NM_000535.6	7	A key component of the mismatch repair system that functions to correct DNA mismatches and small insertions and deletions that can occur during DNA replication and homologous recombination
0	*PTEN*	NM_000314.6	10	Negatively regulates intracellular levels of phosphatidylinositol-3,4,5-trisphosphate in cells and functions as a tumor suppressor by negatively regulating AKT/PKB signaling pathway
1	*RAD51C*	NM_058216.2	17	A member of the RAD51 family similar to bacterial RecA and Saccharomyces cerevisiae Rad51. interacts with RAD51 paralogs and is important for Holliday junction resolution during homologous recombination repair of DNA
0	*RAD51D*	NM_001142571	17	A member of the RAD51 family; forms complex with other members of RAD51 family to catalyze homologous pairing between single- and double-stranded DNA, and plays a role in the early stage of recombinational repair of DNA
1	*TP53*	NM_000546.5	17	A tumor suppressor protein that responds to diverse cellular stresses to regulate expression of target genes, thereby inducing cell cycle arrest, apoptosis, senescence, DNA repair, or changes in metabolism

Abbreviations: chr, chromosome.

[a] Pancreatic study: 1, genes used in pancreatic cancer analysis; 0, genes not used in analysis due to absence of mutations.

[b] Information is adapted from NCBI (National Center for Biotechnology Information) Gene: http://www.ncbi.nlm.nih.gov/gene.

eTable 2. Mutations in Individual Genes Among Cases With Pancreatic Cancer and gnomAD Controls of Different Racial and Ethnic Populations

	Cases								gnomAD Controls							
Gene	AFR	AMR	SAS/ EAS	NFE	Total AFR	Total AMR	Total SAS/ EAS	Total NFE	AFR	AMR	SAS/ EAS	NFE	Total AFR	Total AMR	Total SAS/ EAS	Total NFE
ATM	3	0	1	65	50	42	11	2896	18	57	80	231	6998	16639	23711	52144
BARD1	0	0	0	4	50	42	11	2896	7	8	15	56	7632	16780	23997	55752
BRCA1	0	0	0	18	50	42	11	2896	9	22	45	132	7580	16627	23839	55479
BRCA2	0	2	0	55	50	42	11	2896	20	63	58	172	7589	16430	23506	55213
BRIP1	1	0	0	4	50	42	11	2896	19	17	39	119	7626	16760	23972	55656
CDH1	0	0	0	1	50	42	11	2896	1	0	6	8	7569	16744	23948	55550
CDKN2A	0	0	0	9	50	42	11	2896	1	1	0	13	7641	16776	23994	55709
CHEK2	0	0	0	33	50	42	11	2896	14	32	51	475	7639	16780	23995	55754
FANCC	0	0	0	8	50	42	11	2896	12	12	16	89	7250	16578	23685	54596
MLH1	0	0	0	4	50	42	11	2896	2	4	8	11	7435	16585	23722	55060
MRE11A	0	0	0	2	50	42	11	2896	4	11	28	53	7618	16766	23964	55693
MSH2	0	0	0	1	50	42	11	2896	0	1	3	12	7454	16685	23838	55349
MSH6	0	0	0	6	50	42	11	2896	6	13	26	56	7449	16440	23497	54802
NBN	0	0	0	4	50	42	11	2896	10	12	16	87	7406	16700	23863	54886
NF1	0	0	0	4	50	42	11	2896	4	5	2	20	7622	16775	23990	55740
PALB2	0	1	0	11	50	42	11	2896	15	24	18	96	7588	16764	23976	55583
PMS2	0	0	0	2	50	42	11	2896	4	17	13	52	7637	16766	23970	55697
RAD51C	0	0	0	3	50	42	11	2896	2	5	36	51	7640	16770	23997	55663
TP53	0	0	0	6	50	42	11	2896	1	2	5	17	7376	16540	23640	54419

Abbreviations: AFR, African American; AMR, Hispanic; SAS, South Asian; EAS, East Asian; NFE, non-Finnish European; gnomAD, Genome Aggregation Database.

eTable 3. Allele Count (AC) of Mutations in Mayo Clinic Cases With Pancreatic Cancer and Two Samples of Reference Controls

Gene	Nucleotide	Protein	AC in Mayo Clinic pancreatic cancer cases	AC in gnomAD controls	AC in ExAC controls
ATM	c.*7delG		0	0	1
ATM	c.1027_1030delGAAA		0	2	2
ATM	c.1039G>T	p.Glu347X	0	1	1
ATM	c.103C>T	p.Arg35X	0	5	1
ATM	c.1139_1142dupACAG		1	2	0
ATM	c.1201C>T	p.Gln401X	0	1	1
ATM	c.1236-2A>T		0	0	3
ATM	c.1236-2dupA		0	0	1
ATM	c.1290_1291delTG		0	1	0
ATM	c.1333delC		1	0	0
ATM	c.1339C>T	p.Arg447X	0	2	1
ATM	c.1348delG		0	0	1
ATM	c.1369C>T	p.Arg457X	0	1	0
ATM	c.1402_1403delAA		0	10	6
ATM	c.140C>G	p.Ser47X	0	1	1
ATM	c.1476dupT		0	1	0
ATM	c.1501C>T	p.Gln501X	1	1	0
ATM	c.1564_1565delGA		3	11	3
ATM	c.1596C>A	p.Cys532X	0	0	1
ATM	c.15dupT		0	1	0
ATM	c.1607+1G>T		0	0	1
ATM	c.170G>A	p.Trp57X	1	0	0
ATM	c.1741_1742delTT		0	1	0
ATM	c.1880dupT		0	0	1
ATM	c.1898+1G>C		0	0	1
ATM	c.1978delA		1	0	0
ATM	c.2012_2013dupTA		0	1	1
ATM	c.2023C>T	p.Gln675X	1	3	3
ATM	c.2028	2029insTG	0	0	1
ATM	c.2131_2132dupAA		0	1	0
ATM	c.217_218delGA		0	1	1
ATM	c.2176delC		0	1	1
ATM	c.2192dupA		0	1	0
ATM	c.2201_2202insC		0	1	0
ATM	c.2250G>A	p.=	3	10	1
ATM	c.2251-10T>G		1	0	1
ATM	c.2284_2285delCT		0	2	0
ATM	c.2295_2296delTA		0	1	0
ATM	c.2358_2359delCT		0	1	1
ATM	c.237delA		0	1	0
ATM	c.2413C>T	p.Arg805X	1	10	5
ATM	c.2465T>G	p.Leu822X	0	1	0
ATM	c.2502dupA		1	1	0
ATM	c.2508dupA		0	1	1
ATM	c.2554C>T	p.Gln852X	0	2	2
ATM	c.2690delT		0	1	1
ATM	c.2693T>G	p.Leu898X	0	1	0
ATM	c.2734C>T	p.Gln912X	0	1	1
ATM	c.2806	2809dupCTAG	0	0	1
ATM	c.2840dupA		0	1	0
ATM	c.2880delC		1	1	1
ATM	c.2906_2909dupTTCT		0	1	0

ATM	c.2921+1G>A		1	0	2
ATM	c.2958_2961delTTGT		0	1	0
ATM	c.2T>C	p.Met1	0	1	0
ATM	c.3077+1G>A		0	0	1
ATM	c.3077+1G>T		0	0	1
ATM	c.3078-1G>A		1	3	1
ATM	c.3154-2A>G		1	0	0
ATM	c.3173G>A	p.Trp1058X	0	1	0
ATM	c.3214G>T	p.Glu1072X	0	1	0
ATM	c.3245_3247delinsTGAT		5	0	0
ATM	c.3281delA		0	2	2
ATM	c.3284G>A	p.Arg1095Lys	0	1	1
ATM	c.3292C>T	p.Gln1098X	0	1	0
ATM	c.331+5G>A		0	0	1
ATM	c.332-1G>A		0	0	1
ATM	c.3372C>G	p.Tyr1124X	0	1	0
ATM	c.3475delG		0	1	1
ATM	c.3510dupA		0	1	0
ATM	c.3542_3543delAA		0	1	1
ATM	c.3546_3547delGA		0	1	1
ATM	c.3576G>A	p.=	0	4	1
ATM	c.362T>A	p.Leu121X	0	1	0
ATM	c.3712_3716del5		1	0	0
ATM	c.378delT		1	0	0
ATM	c.3802delG		4	10	2
ATM	c.381delA		0	1	0
ATM	c.3837delG		0	1	0
ATM	c.387delA		0	1	1
ATM	c.3894dupT		0	1	0
ATM	c.392C>G	p.Ser131X	0	1	0
ATM	c.3931C>T	p.Gln1311X	0	1	1
ATM	c.3993+1G>A		1	0	3
ATM	c.3994-2A>G		1	0	4
ATM	c.3G>A	p.Met1	0	1	0
ATM	c.4019_4029del11		0	1	0
ATM	c.4046_4064del19		0	1	1
ATM	c.4083_4087del5		0	1	0
ATM	c.4106C>A_p.Ser1369X		0	1	0
ATM	c.4176T>A_p.Tyr1392X		0	1	1
ATM	c.4373delG		0	1	0
ATM	c.4394T>C	p.Leu1465Pro	0	1	1
ATM	c.4396C>T	p.Arg1466X	0	4	1
ATM	c.43delC		0	1	1
ATM	c.4437-1G>C		0	1	1
ATM	c.4493T>G	p.Leu1498X	0	1	0
ATM	c.450_453delTTCT		0	1	0
ATM	c.4507C>T	p.Gln1503X	0	3	0
ATM	c.4525_4526insAGTA		0	1	0
ATM	c.4603C>T	p.Gln1535X	0	1	1
ATM	c.4625dupT		0	1	0
ATM	c.4668T>G	p.Tyr1556X	0	3	3
ATM	c.467G>A	p.Trp156X	1	0	0
ATM	c.4732C>T	p.Gln1578X	0	1	1
ATM	c.4776+2_4776+13del12		0	0	1
ATM	c.4842_4843insCT		0	1	0
ATM	c.4844_4848del5		0	1	1
ATM	c.4852C>T	p.Arg1618X	0	1	1
ATM	c.4909+1G>A		0	0	1

ATM	c.4941dupA		0	0	1
ATM	c.50_51delAT		0	1	1
ATM	c.5188C>T	p.Arg1730X	0	2	1
ATM	c.5228C>T	p.Thr1743Ile	0	4	2
ATM	c.5383_5384delTG		0	1	0
ATM	c.540delA		0	2	0
ATM	c.5443delG		0	1	0
ATM	c.5487_5488insGTTAA		0	1	0
ATM	c.5549delT		0	1	0
ATM	c.5554C>T	p.Gln1852X	0	1	1
ATM	c.5623C>T	p.Arg1875X	2	4	0
ATM	c.5632_5635delTCGC		0	3	1
ATM	c.565_566insAAAAA		0	1	0
ATM	c.565dupA		0	1	0
ATM	c.5674G>T	p.Glu1892X	1	0	0
ATM	c.5692C>T	p.Arg1898X	0	2	1
ATM	c.5697C>A	p.Cys1899X	0	0	1
ATM	c.5712dupA		1	2	0
ATM	c.572_585del14		0	0	3
ATM	c.5870_5871delAT		0	1	0
ATM	c.5908C>T	p.Gln1970X	0	7	0
ATM	c.5919-2A>G		0	0	1
ATM	c.5932G>T	p.Glu1978X	2	11	1
ATM	c.5979_5983del5		0	2	1
ATM	c.6013delinsAA		1	0	0
ATM	c.6082C>T	p.Gln2028X	0	1	0
ATM	c.6095G>A	p.Arg2032Lys	0	7	3
ATM	c.6100C>T	p.Arg2034X	3	1	0
ATM	c.6154G>A	p.Glu2052Lys	0	15	7
ATM	c.6198+1G>A		0	0	1
ATM	c.6200C>A	p.Ala2067Asp	0	1	0
ATM	c.6404_6405insTT		0	1	1
ATM	c.6415G>T	p.Glu2139X	0	1	0
ATM	c.6573-2A>G		0	0	2
ATM	c.6596_6597delCT		0	0	1
ATM	c.6635_6636delTC		0	1	1
ATM	c.6650_6657del8		0	1	1
ATM	c.666_678del13		0	1	0
ATM	c.6677dupT		0	1	0
ATM	c.6750delT		0	1	0
ATM	c.67C>T	p.Arg23X	0	5	2
ATM	c.6908dupA		0	2	2
ATM	c.695_699del5		0	1	0
ATM	c.6997dupA		0	2	0
ATM	c.7000_7003delTACA		1	2	1
ATM	c.7032G>A	p.Trp2344X	0	3	0
ATM	c.7072C>T	p.Gln2358X	0	1	0
ATM	c.7096G>T	p.Glu2366X	0	1	0
ATM	c.70A>T	p.Lys24X	0	1	0
ATM	c.7223C>A	p.Ser2408X	0	1	0
ATM	c.7271T>G	p.Val2424Gly	0	11	2
ATM	c.7327C>T	p.Arg2443X	1	1	0
ATM	c.7388dupT		0	1	0
ATM	c.741dupT		1	0	0
ATM	c.742C>T	p.Arg248X	0	2	1
ATM	c.7449G>A	p.Trp2483X	0	3	0
ATM	c.7456C>T	p.Arg2486X	0	3	0
ATM	c.7463G>A	p.Cys2488Tyr	1	0	0

ATM	c.748C>T	p.Arg250X	0	1	1
ATM	c.7517_7520delGAGA		0	2	0
ATM	c.7542T>G	p.Tyr2514X	0	1	1
ATM	c.7570G>C	p.Ala2524Pro	1	0	0
ATM	c.7629_7629+4del5		0	1	1
ATM	c.7630-2A>C		5	0	0
ATM	c.7638_7646del9	p.Arg2547_Ser2549del	2	5	1
ATM	c.7705_7706delGA		0	5	1
ATM	c.7777C>T	p.Gln2593X	0	1	1
ATM	c.7788G>A	p.=	0	2	1
ATM	c.7792C>T	p.Arg2598X	0	0	1
ATM	c.7875_7876delinsGC	p.Asp2625_Ala2626delinsGluPro	1	0	0
ATM	c.7880delA		0	1	0
ATM	c.790delT		3	2	0
ATM	c.7913G>A	p.Trp2638X	0	1	1
ATM	c.7921C>T	p.Gln2641X	0	1	1
ATM	c.7989_7991delTGT	p.Val2664del	0	1	0
ATM	c.8014delG		0	1	0
ATM	c.802C>T	p.Gln268X	0	1	1
ATM	c.8049_8056dup8		1	1	0
ATM	c.8122G>A	p.Asp2708Asn	0	1	0
ATM	c.8147T>C	p.Val2716Ala	0	6	1
ATM	c.8185C>T	p.Gln2729X	0	0	1
ATM	c.8213T>G	p.Leu2738X	0	1	0
ATM	c.8224_8225delAA		0	1	0
ATM	c.824delT		0	2	0
ATM	c.8264_8268del5		1	2	1
ATM	c.8264dupA		1	0	0
ATM	c.8266A>T	p.Lys2756X	0	4	1
ATM	c.8278delC		0	1	0
ATM	c.8281_8284delTCTC		0	1	1
ATM	c.8284C>T	p.Gln2762X	0	1	1
ATM	c.8292_8293delTG		0	1	0
ATM	c.8293G>A	p.Gly2765Ser	0	2	1
ATM	c.8303_8306delAATG		0	1	0
ATM	c.8307G>A	p.Trp2769X	0	2	1
ATM	c.8321delT		1	0	0
ATM	c.8395_8404del10		0	4	1
ATM	c.8397delT		0	1	0
ATM	c.8418+1G>A		0	0	1
ATM	c.8418+5_8418+8delGTGA		0	1	1
ATM	c.8435_8436delCT		0	3	1
ATM	c.8440delG		0	1	0
ATM	c.8494C>T	p.Arg2832Cys	0	7	0
ATM	c.850C>T	p.Gln284X	0	1	1
ATM	c.8545C>T	p.Arg2849X	1	1	0
ATM	c.8549T>A	p.Leu2850X	0	1	0
ATM	c.8565T>G	p.Ser2855Arg	0	1	0
ATM	c.8615_8616delAT		0	2	0
ATM	c.8638G>T	p.Glu2880X	0	1	1
ATM	c.8655dupT		1	4	5
ATM	c.8672delG		0	1	1
ATM	c.8683G>T	p.Glu2895X	0	2	1
ATM	c.8732C>T	p.Thr2911Ile	1	0	0
ATM	c.8737_8738delGA		1	0	0
ATM	c.8744_8745dupTG		0	1	0
ATM	c.875C>T	p.Pro292Leu	0	2	2
ATM	c.8786+1G>A		0	0	1

ATM	c.8787-2_8793del9		0	1	1
ATM	c.8814_8824del11		0	2	1
ATM	c.8833_8834delCT		0	1	0
ATM	c.8873_8874delTT		0	1	1
ATM	c.8876_8879delACTG		0	3	1
ATM	c.8977C>T	p.Arg2993X	0	3	1
ATM	c.8987+2T>A		0	0	1
ATM	c.901+1G>A		0	0	2
ATM	c.9021dupA		1	0	0
ATM	c.902-1G>T		0	1	0
ATM	c.9022C>T	p.Arg3008Cys	0	4	2
ATM	c.9023G>A	p.Arg3008His	0	1	1
ATM	c.9040C>T	p.Gln3014X	1	0	0
ATM	c.9079dupA		0	1	0
ATM	c.9139C>T	p.Arg3047X	0	3	0
ATM	c.9156G>A	p.Trp3052X	0	1	0
ATM	c.939C>A	p.Tyr313X	0	1	0
ATM	c.943_944delTT		0	2	2
ATM	c.980delG		0	1	0
ATM	c.992delA		0	1	0
BARD1	c.115_116delGC		0	1	0
BARD1	c.1196T>G	p.Leu399X	0	1	0
BARD1	c.1205C>A	p.Ser402X	0	2	0
BARD1	c.1216C>T	p.Arg406X	0	4	3
BARD1	c.1314+1G>A		0	1	1
BARD1	c.1329delT		0	1	1
BARD1	c.1338C>A	p.Tyr446X	0	2	1
BARD1	c.1385G>A	p.Trp462X	0	1	0
BARD1	c.1487C>G	p.Ser496X	0	1	0
BARD1	c.1539dupA		0	1	0
BARD1	c.1652C>G	p.Ser551X	1	4	1
BARD1	c.1662_1665delTAGC		0	1	1
BARD1	c.1678-1G>T		0	0	1
BARD1	c.1690C>T	p.Gln564X	0	5	4
BARD1	c.1819delG		0	1	1
BARD1	c.1886G>A	p.Trp629X	0	1	1
BARD1	c.1904-2_1904-1delAG		0	1	1
BARD1	c.1905G>A	p.Trp635X	0	1	0
BARD1	c.1921C>T	p.Arg641X	1	4	1
BARD1	c.1935_1954dup20		0	14	6
BARD1	c.2001+1G>T		0	1	1
BARD1	c.2001+2T>C		0	0	1
BARD1	c.2136dupC		0	1	1
BARD1	c.2148_2149delCA		0	1	0
BARD1	c.2199C>A	p.Cys733X	0	1	1
BARD1	c.2203C>T	p.Gln735X	0	2	1
BARD1	c.2229dupT		0	1	0
BARD1	c.2290dupA		0	1	1
BARD1	c.2300_2301delTG		0	6	5
BARD1	c.258T>A	p.Cys86X	0	0	1
BARD1	c.298C>T	p.Gln100X	0	1	0
BARD1	c.334C>T	p.Arg112X	0	2	1
BARD1	c.448C>T	p.Arg150X	1	2	1
BARD1	c.457_460dupAAAG		0	1	0
BARD1	c.496C>T	p.Gln166X	0	1	0
BARD1	c.513dupA		0	0	1
BARD1	c.526C>T	p.Gln176X	0	0	1
BARD1	c.539_540delAT		0	1	1

BARD1	c.547delG		0	1	1
BARD1	c.55G>T	p.Glu19X	0	4	1
BARD1	c.601delA		0	1	0
BARD1	c.623dupA		0	2	2
BARD1	c.627_628delAA		0	1	0
BARD1	c.632T>A	p.Leu211X	1	0	0
BARD1	c.659T>G	p.Leu220X	0	1	0
BARD1	c.70_71insGT		0	3	0
BARD1	c.733C>T	p.Gln245X	0	1	0
BARD1	c.860_861delAG		0	1	0
BARD1	c.941T>A	p.Leu314X	0	1	1
BARD1	c.998_999delCT		0	1	0
BRCA1	c.*63C>T		0	0	1
BRCA1	c.1039delC		0	0	1
BRCA1	c.1054G>T	p.Glu352X	0	2	1
BRCA1	c.1082_1092del11		0	3	3
BRCA1	c.1175_1214del40		0	1	0
BRCA1	c.1190delA		0	1	1
BRCA1	c.135-1G>T		0	0	1
BRCA1	c.1360_1361delAG		0	2	2
BRCA1	c.1504_1508del5		0	2	2
BRCA1	c.1556delA		2	0	0
BRCA1	c.1579_1580delAA		0	1	0
BRCA1	c.1650dupT		0	1	1
BRCA1	c.1687C>T	p.Gln563X	0	6	4
BRCA1	c.1772delT		0	1	1
BRCA1	c.1793T>G	p.Leu598X	0	2	2
BRCA1	c.181T>G	p.Cys61Gly	0	7	3
BRCA1	c.1823_1826delAGAA		0	1	0
BRCA1	c.1839_1840delGA		0	1	1
BRCA1	c.1842_1843dupGT		0	1	1
BRCA1	c.1881_1884delCAGT		0	1	0
BRCA1	c.188T>A	p.Leu63X	0	0	1
BRCA1	c.190T>G	p.Cys64Gly	0	1	0
BRCA1	c.1961dupA		0	3	2
BRCA1	c.1977_1978delAG		0	1	1
BRCA1	c.2035A>T	p.Lys679X	1	0	0
BRCA1	c.211A>G	p.Arg71Gly	0	1	0
BRCA1	c.212+1G>A		1	1	0
BRCA1	c.2215_2216insCT		0	1	0
BRCA1	c.2269delG		0	1	2
BRCA1	c.2296_2297delAG		0	1	0
BRCA1	c.2338C>T	p.Gln780X	0	1	0
BRCA1	c.2405_2406delTG		0	1	1
BRCA1	c.2409delT		0	1	1
BRCA1	c.2433delC		0	2	1
BRCA1	c.2457delC		0	2	0
BRCA1	c.246delT		0	1	0
BRCA1	c.2475delC		1	1	0
BRCA1	c.2603C>G	p.Ser868X	0	1	0
BRCA1	c.2685_2686delAA		0	1	0
BRCA1	c.2706_2707dupAT		0	2	0
BRCA1	c.2709_2710delTG		1	0	0
BRCA1	c.2767_2770delGTTA		0	1	0
BRCA1	c.2796_2799delTGGT		0	1	0
BRCA1	c.2836_2837delAT		0	2	1
BRCA1	c.301+1G>A		0	1	1
BRCA1	c.301+1G>C		0	1	1

BRCA1	c.301+1G>T		0	1	0
BRCA1	c.3037_3038delGA		0	1	1
BRCA1	c.3048_3052dup5		0	4	4
BRCA1	c.3083delG		0	1	0
BRCA1	c.3143delG		0	1	0
BRCA1	c.3228_3229delAG		0	1	0
BRCA1	c.3331C>T	p.Gln1111X	0	1	0
BRCA1	c.3400G>T	p.Glu1134X	1	0	0
BRCA1	c.3442delG		0	1	0
BRCA1	c.346delG		0	1	1
BRCA1	c.3477_3480delAAAG		0	1	1
BRCA1	c.3481_3491del11		1	0	0
BRCA1	c.3485delA		0	1	0
BRCA1	c.34C>T	p.Gln12X	0	0	1
BRCA1	c.3531dupT		0	1	0
BRCA1	c.3598C>T	p.Gln1200X	0	1	0
BRCA1	c.3607C>T	p.Arg1203X	0	3	0
BRCA1	c.3626delT		0	1	1
BRCA1	c.3627dupA		0	2	1
BRCA1	c.3644_3648del5		0	1	0
BRCA1	c.3648dupA		0	1	1
BRCA1	c.3661G>T	p.Glu1221X	0	1	1
BRCA1	c.3710_3711dupTA		0	1	0
BRCA1	c.3748G>T	p.Glu1250X	0	2	1
BRCA1	c.3756_3759delGTCT		0	3	0
BRCA1	c.3770_3771delAG		0	2	1
BRCA1	c.3858_3861delTGAG		0	2	0
BRCA1	c.390C>A	p.Tyr130X	0	1	1
BRCA1	c.3931_3934delAACA		0	1	0
BRCA1	c.3999_4008del10		0	1	1
BRCA1	c.4015G>T	p.Glu1339X	0	1	0
BRCA1	c.4035delA		0	4	2
BRCA1	c.4065_4068delTCAA		2	2	1
BRCA1	c.4088C>G	p.Ser1363X	0	1	0
BRCA1	c.4096+1G>A		0	3	3
BRCA1	c.4165_4166delAG		0	1	1
BRCA1	c.4182_4183dupTC		0	1	0
BRCA1	c.4183C>T	p.Gln1395X	0	1	0
BRCA1	c.4222C>T	p.Gln1408X	0	1	0
BRCA1	c.4243delG		0	1	0
BRCA1	c.4300dupA		0	1	0
BRCA1	c.4327C>T	p.Arg1443X	0	5	1
BRCA1	c.4382_4388dup7		1	0	0
BRCA1	c.441G>C	p.Leu147Phe	0	3	2
BRCA1	c.4484G>T	p.Arg1495Met	0	1	1
BRCA1	c.4485-1G>A		0	0	2
BRCA1	c.4508C>A	p.Ser1503X	0	2	1
BRCA1	c.4689C>G	p.Tyr1563X	1	2	0
BRCA1	c.4745delA		0	1	0
BRCA1	c.4868C>G	p.Ala1623Gly	0	1	1
BRCA1	c.4936delG		0	1	0
BRCA1	c.4964_4982del19		0	0	1
BRCA1	c.5068A>T	p.Lys1690X	0	1	0
BRCA1	c.5074+1G>A		0	1	0
BRCA1	c.5074G>C	p.Asp1692His	0	2	1
BRCA1	c.5080G>T	p.Glu1694X	0	1	0
BRCA1	c.5095C>T	p.Arg1699Trp	0	3	0
BRCA1	c.5096G>A	p.Arg1699Gln	1	6	3

BRCA1	c.5102	5103delTG	0	0	1
BRCA1	c.5123C>A	p.Ala1708Glu	0	5	3
BRCA1	c.5136G>A	p.Trp1712X	0	1	0
BRCA1	c.5137delG		0	1	0
BRCA1	c.514C>T	p.Gln172X	0	1	1
BRCA1	c.514delC		1	0	0
BRCA1	c.5177_5180delGAAA		0	2	1
BRCA1	c.5251C>T	p.Arg1751X	0	2	0
BRCA1	c.5266dupC		3	15	9
BRCA1	c.5324T>G	p.Met1775Arg	0	3	0
BRCA1	c.5332+1G>A		0	1	1
BRCA1	c.5333-2A>C		0	0	1
BRCA1	c.5363G>A	p.Gly1788Asp	0	1	1
BRCA1	c.5366delC		0	0	1
BRCA1	c.5391delA		0	1	1
BRCA1	c.5468-1G>T		0	0	1
BRCA1	c.5485dupG		0	1	1
BRCA1	c.5497G>A	p.Val1833Met	0	1	0
BRCA1	c.5503C>T	p.Arg1835X	1	3	0
BRCA1	c.68_69delAG		0	16	23
BRCA1	c.791_794delGTTC		0	2	1
BRCA1	c.81-1G>A		0	0	1
BRCA1	c.815_824dup10		0	1	0
BRCA1	c.895_896delGT		0	1	1
BRCA1	c.923delG		0	1	0
BRCA1	c.929delA		0	1	0
BRCA1	c.962G>A	p.Trp321X	0	1	1
BRCA2	c.*14C>T		0	0	3
BRCA2	c.*8C>T		0	0	1
BRCA2	c.1039C>T	p.Gln347X	0	1	0
BRCA2	c.1097T>G	p.Leu366X	0	1	0
BRCA2	c.1189_1190insTTAG		0	1	0
BRCA2	c.1265delA		1	1	0
BRCA2	c.1310_1313delAAGA		0	1	1
BRCA2	c.133G>T	p.Glu45X	0	1	0
BRCA2	c.1399A>T	p.Lys467X	0	1	1
BRCA2	c.145G>T	p.Glu49X	0	1	0
BRCA2	c.1672delA		0	1	1
BRCA2	c.1755_1759del5		0	1	0
BRCA2	c.1796_1800del5		0	1	0
BRCA2	c.1813dupA		2	0	1
BRCA2	c.1887_1893del7		0	1	0
BRCA2	c.2059_2063del5		0	1	0
BRCA2	c.2224C>T	p.Gln742X	0	0	1
BRCA2	c.2279_2283del5		0	1	1
BRCA2	c.2287delC		1	0	0
BRCA2	c.2319delT		0	1	1
BRCA2	c.2330dupA		0	1	0
BRCA2	c.2339C>G	p.Ser780X	0	1	0
BRCA2	c.2376C>A	p.Tyr792X	0	1	0
BRCA2	c.2442delC		0	1	1
BRCA2	c.2588dupA		0	0	2
BRCA2	c.2589delT		0	1	1
BRCA2	c.2653_2656delGACA		0	2	1
BRCA2	c.2655_2656delCA		0	1	0
BRCA2	c.2658_2659dupTG		0	1	0
BRCA2	c.26delC		1	0	0
BRCA2	c.2743_2747del5		0	1	1

BRCA2	c.2786dupT		1	0	0
BRCA2	c.2808_2811delACAA		0	3	0
BRCA2	c.2830A>T	p.Lys944X	1	2	0
BRCA2	c.2926delT		0	1	0
BRCA2	c.2957_2958insG		0	1	0
BRCA2	c.2T>G	p.Met1	0	2	0
BRCA2	c.2T>G	p.Met1?	0	0	1
BRCA2	c.3046G>T	p.Glu1016X	0	1	0
BRCA2	c.306delA		0	1	0
BRCA2	c.3076_3077delAA		0	1	0
BRCA2	c.3100_3101delAT		0	3	0
BRCA2	c.3103G>T	p.Glu1035X	0	1	0
BRCA2	c.3109C>T	p.Gln1037X	0	1	0
BRCA2	c.3167_3170delAAAA		0	1	1
BRCA2	c.3170_3174del5		0	3	3
BRCA2	c.3264dupT		0	5	1
BRCA2	c.3436G>T	p.Glu1146X	0	1	0
BRCA2	c.3455T>G	p.Leu1152X	0	1	1
BRCA2	c.3481_3482dupGA		0	1	0
BRCA2	c.3500_3501delTA		0	1	0
BRCA2	c.3545_3546delTT		0	5	4
BRCA2	c.3599_3600delGT		0	5	4
BRCA2	c.3708dupA		0	1	0
BRCA2	c.3744_3747delTGAG		2	1	0
BRCA2	c.3751dupA		0	1	0
BRCA2	c.3785C>G	p.Ser1262X	0	1	1
BRCA2	c.3847_3848delGT		2	9	9
BRCA2	c.3871C>T	p.Gln1291X	0	1	1
BRCA2	c.3975_3978dupTGCT		0	1	0
BRCA2	c.4003G>T	p.Glu1335X	0	1	1
BRCA2	c.4037_4038delCT		0	0	1
BRCA2	c.4160_4161insGGAAG		0	1	1
BRCA2	c.4162_4166del5		0	1	1
BRCA2	c.4245delG		0	1	1
BRCA2	c.4256_4257delAA		0	1	0
BRCA2	c.4258delG		0	2	1
BRCA2	c.425G>A	p.Ser142Asn	0	8	5
BRCA2	c.4325C>A	p.Ser1442X	0	1	0
BRCA2	c.4398_4402del5		0	1	1
BRCA2	c.4409_4410delTA		0	1	0
BRCA2	c.4414_4415delAA		0	1	1
BRCA2	c.4415_4418delAGAA		0	1	1
BRCA2	c.4471_4474delCTGA		0	1	0
BRCA2	c.4478_4481delAAAG		1	4	0
BRCA2	c.4525C>T	p.Gln1509X	0	0	1
BRCA2	c.4544dupA		0	1	0
BRCA2	c.4563_4564delAT		0	2	0
BRCA2	c.4593dupA		0	0	1
BRCA2	c.4631delA		0	1	0
BRCA2	c.4631dupA		0	1	1
BRCA2	c.4691dupC		0	1	0
BRCA2	c.475+1G>A		0	0	1
BRCA2	c.4794_4797delCAAT		0	1	0
BRCA2	c.4808dupA		0	2	0
BRCA2	c.4821_4822delTG		0	2	1
BRCA2	c.4829_4830delTG		0	1	2
BRCA2	c.4859T>G	p.Leu1620X	0	1	0
BRCA2	c.4876_4877delAA		0	2	0

BRCA2	c.4889C>G	p.Ser1630X	0	2	1
BRCA2	c.4914dupA		0	1	0
BRCA2	c.4936delG		0	1	1
BRCA2	c.4965C>G	p.Tyr1655X	1	1	0
BRCA2	c.5073dupA		0	0	1
BRCA2	c.5079dupT		0	1	0
BRCA2	c.5110_5113delAGAA		0	1	0
BRCA2	c.5130_5133delTGTA		0	1	0
BRCA2	c.5146_5149delTATG		0	1	1
BRCA2	c.5213_5216delCTTA		1	1	1
BRCA2	c.5217_5223del7		0	1	1
BRCA2	c.523C>T	p.Gln175X	0	0	1
BRCA2	c.5241_5242insTA		0	1	1
BRCA2	c.5290_5291delTC		0	1	0
BRCA2	c.5303_5304delTT		0	1	0
BRCA2	c.5350_5351delAA		2	0	0
BRCA2	c.5542delA		0	1	1
BRCA2	c.5576_5579delTTAA		0	4	3
BRCA2	c.5604	5605delCA	0	0	1
BRCA2	c.5616_5620del5		0	1	2
BRCA2	c.5621_5624delTTAA		2	1	0
BRCA2	c.5631delC		0	1	1
BRCA2	c.5645C>A	p.Ser1882X	2	3	3
BRCA2	c.5681dupA		0	1	0
BRCA2	c.5682C>G	p.Tyr1894X	0	1	0
BRCA2	c.5722_5723delCT		1	1	0
BRCA2	c.5754_5755delTA		0	1	1
BRCA2	c.5773C>T	p.Gln1925X	0	1	1
BRCA2	c.5796_5797delTA		0	0	1
BRCA2	c.5828delC		0	1	0
BRCA2	c.5855T>A	p.Leu1952X	0	1	0
BRCA2	c.5857G>T	p.Glu1953X	0	3	0
BRCA2	c.5864C>A	p.Ser1955X	1	3	0
BRCA2	c.5946delT		4	7	21
BRCA2	c.5978T>G	p.Leu1993X	0	1	1
BRCA2	c.6024dupG		0	1	0
BRCA2	c.6033_6034delTT		0	1	0
BRCA2	c.6037A>T	p.Lys2013X	1	0	0
BRCA2	c.6065C>G	p.Ser2022X	0	1	1
BRCA2	c.6079dupA		0	1	1
BRCA2	c.6082_6086del5		0	1	1
BRCA2	c.6124C>T	p.Gln2042X	0	1	1
BRCA2	c.6267delG		0	1	0
BRCA2	c.6269delA		0	1	0
BRCA2	c.6275_6276delTT		2	7	2
BRCA2	c.6373dupA		1	0	0
BRCA2	c.6466_6469delTCTC		0	1	1
BRCA2	c.6468_6469dupTC		0	1	0
BRCA2	c.6485_6486delAA		0	1	0
BRCA2	c.6486_6489delACAA		0	2	1
BRCA2	c.6535_6536insA		0	1	1
BRCA2	c.658_659delGT		1	10	3
BRCA2	c.6591_6592delTG		0	1	0
BRCA2	c.6600_6601delTT		0	1	0
BRCA2	c.6643delT		0	1	0
BRCA2	c.6644_6647delACTC		1	1	0
BRCA2	c.6673_6703dup31		0	1	0
BRCA2	c.6724_6725delGA		0	1	0

BRCA2	c.6757_6758delCT		0	1	0
BRCA2	c.6761_6762delTT		2	0	0
BRCA2	c.6787dupG		0	1	0
BRCA2	c.68-2A>G		0	0	1
BRCA2	c.6841+1G>A		0	0	1
BRCA2	c.6849delC		0	1	0
BRCA2	c.6859_6863del5		0	1	0
BRCA2	c.6899_6906dup8		0	1	1
BRCA2	c.6944_6947delTAAA		0	2	0
BRCA2	c.6998dupT		0	1	0
BRCA2	c.7007G>C	p.Arg2336Pro	0	1	0
BRCA2	c.7024C>T	p.Gln2342X	0	1	0
BRCA2	c.7025_7026delAA		1	0	0
BRCA2	c.7063G>T	p.Glu2355X	0	1	1
BRCA2	c.7069_7070delCT		5	5	4
BRCA2	c.7147dupT		0	1	1
BRCA2	c.7208_7211delCCAA		0	1	0
BRCA2	c.7235C>T	p.Thr2412Ile	0	1	1
BRCA2	c.7379_7382delACAA		0	1	0
BRCA2	c.7419_7420delTG		0	1	0
BRCA2	c.7422dupA		1	0	0
BRCA2	c.7433dupT		0	1	1
BRCA2	c.7435+2T>A		1	0	0
BRCA2	c.7480C>T	p.Arg2494X	0	4	1
BRCA2	c.755_758delACAG		1	2	0
BRCA2	c.7556dupC		0	1	0
BRCA2	c.7558C>T	p.Arg2520X	1	3	1
BRCA2	c.7673_7674dupAG		0	1	1
BRCA2	c.7697dupA		0	0	1
BRCA2	c.7757G>A	p.Trp2586X	0	2	0
BRCA2	c.7805+1G>A		0	0	1
BRCA2	c.7878G>C	p.Trp2626Cys	0	1	1
BRCA2	c.7933A>T	p.Arg2645X	0	1	0
BRCA2	c.7976+1G>A		1	0	0
BRCA2	c.7976G>A	p.Arg2659Lys	0	1	0
BRCA2	c.7977-1G>C		0	1	0
BRCA2	c.8023A>G	p.Ile2675Val	0	1	0
BRCA2	c.8167G>C	p.Asp2723His	3	0	0
BRCA2	c.8168A>C	p.Asp2723Ala	0	1	1
BRCA2	c.8169T>A	p.Asp2723Glu	0	1	0
BRCA2	c.8243G>A	p.Gly2748Asp	1	2	0
BRCA2	c.8247_8248delGA		1	0	0
BRCA2	c.8377G>A	p.Gly2793Arg	0	2	1
BRCA2	c.8463dupT		0	1	1
BRCA2	c.8478C>A	p.Tyr2826X	0	1	1
BRCA2	c.8488-2A>G		0	0	1
BRCA2	c.8537_8538delAG		0	3	0
BRCA2	c.8594dupT		1	0	0
BRCA2	c.8754G>A	p.=	0	1	0
BRCA2	c.8755-1G>A		1	0	0
BRCA2	c.8953+1G>T		1	0	0
BRCA2	c.9004G>A	p.Glu3002Lys	1	0	0
BRCA2	c.9016_9017delTA		0	1	0
BRCA2	c.9026_9030del5		0	1	0
BRCA2	c.9054_9055delTA		0	1	0
BRCA2	c.9097delA		0	0	6
BRCA2	c.9097dupA		0	0	9
BRCA2	c.9100C>T	p.Gln3034X	1	0	0

BRCA2	c.9117G>A	p.=	1	0	0
BRCA2	c.9127G>T	p.Glu3043X	0	0	1
BRCA2	c.9154C>T	p.Arg3052Trp	0	3	1
BRCA2	c.9227G>A	p.Gly3076Glu	0	1	1
BRCA2	c.9235delG		0	8	1
BRCA2	c.9253dupA		0	2	0
BRCA2	c.9285C>G	p.Asp3095Glu	0	1	0
BRCA2	c.9286dupG		0	1	0
BRCA2	c.9294C>G	p.Tyr3098X	0	2	0
BRCA2	c.92G>A	p.Trp31X	0	1	1
BRCA2	c.9371A>T	p.Asn3124Ile	0	2	1
BRCA2	c.9382C>T	p.Arg3128X	0	3	1
BRCA2	c.9403delC		1	0	0
BRCA2	c.9435_9436delGT		0	1	0
BRCA2	c.9546delT		0	1	0
BRCA2	c.956dupA		0	2	1
BRCA2	c.9580_9581delCC		0	1	1
BRCA2	c.961C>T	p.Gln321X	1	0	0
BRCA2	c.9666delT		0	1	1
BRCA2	c.9672dupA		0	1	0
BRCA2	c.9682delA		0	1	0
BRCA2	c.9689delT		0	1	1
BRCA2	c.9699_9702delTATG		0	17	12
BRCA2	c.9770_9773delAAGA		0	1	1
BRCA2	c.9808delG		0	1	0
BRCA2	c.9867delT		1	0	0
BRCA2	c.9891	9894dupATTT	0	0	1
BRCA2	c.9924C>G	p.Tyr3308X	0	2	0
BRCA2	c.994dupA		0	1	1
BRIP1	c.1045G>C	p.Ala349Pro	0	3	2
BRIP1	c.1066C>T	p.Arg356X	0	2	0
BRIP1	c.1067delG		0	0	1
BRIP1	c.1069G>T	p.Glu357X	0	0	1
BRIP1	c.1070	1071insTTAC	0	0	1
BRIP1	c.1126_1127delCA		0	1	1
BRIP1	c.1141-2A>G		0	0	1
BRIP1	c.1162C>T	p.Gln388X	0	1	0
BRIP1	c.1236delA		0	1	0
BRIP1	c.1240C>T	p.Gln414X	0	1	1
BRIP1	c.128_131delTGTT		0	1	0
BRIP1	c.1315C>T	p.Arg439X	0	3	0
BRIP1	c.133G>T	p.Glu45X	1	0	0
BRIP1	c.1343G>A	p.Trp448X	0	6	4
BRIP1	c.1372G>T	p.Glu458X	0	0	1
BRIP1	c.1425_1429del5		0	1	1
BRIP1	c.1473+1G>T		0	0	1
BRIP1	c.1488_1500del13		0	1	1
BRIP1	c.1510dupA		0	1	1
BRIP1	c.1551_1552insACAG		0	1	0
BRIP1	c.1661delA		0	0	1
BRIP1	c.1702_1703delAA		0	1	0
BRIP1	c.1741C>T	p.Arg581X	0	5	1
BRIP1	c.1776G>A	p.Trp592X	0	0	1
BRIP1	c.1794+1G>A		0	2	1
BRIP1	c.1795-2A>T		0	0	2
BRIP1	c.1853_1854insG		0	1	0
BRIP1	c.1871C>A	p.Ser624X	0	4	0
BRIP1	c.1888dupA		0	1	1

BRIP1	c.193C>T	p.Gln65X	0	3	4
BRIP1	c.1970delG		0	2	2
BRIP1	c.2010dupT		0	2	2
BRIP1	c.2012delA		0	1	0
BRIP1	c.2053C>T	p.Gln685X	0	1	0
BRIP1	c.2108_2109insCC		0	3	2
BRIP1	c.2133delT		1	0	0
BRIP1	c.2164_2167delACAG		0	1	1
BRIP1	c.2223_2225dupGTA	p.Val741_Tyr742insX	0	1	0
BRIP1	c.2250delA		0	1	0
BRIP1	c.2255_2256delAA		0	2	0
BRIP1	c.2273dupT		0	2	0
BRIP1	c.2313	2319del7	0	0	1
BRIP1	c.2377C>T	p.Gln793X	0	1	1
BRIP1	c.2392C>T	p.Arg798X	1	40	10
BRIP1	c.2400C>G	p.Tyr800X	1	6	2
BRIP1	c.241delG		0	1	0
BRIP1	c.24T>A	p.Tyr8X	0	0	1
BRIP1	c.2581dupT		0	1	0
BRIP1	c.2684_2687delCCAT		0	3	2
BRIP1	c.2732dupT		0	12	6
BRIP1	c.2765T>G	p.Leu922X	0	3	1
BRIP1	c.2823dupA		0	1	1
BRIP1	c.2830C>T	p.Gln944X	0	1	1
BRIP1	c.285delT		0	1	0
BRIP1	c.2867C>G	p.Ser956X	0	1	1
BRIP1	c.290_293delACAA		0	1	1
BRIP1	c.2906-1G>A		0	0	1
BRIP1	c.2947delA		0	1	0
BRIP1	c.2947dupA		0	3	1
BRIP1	c.2990_2993delCAAA		1	8	2
BRIP1	c.2992_2993delAA		0	1	0
BRIP1	c.2992_2995delAAGA		0	2	0
BRIP1	c.3004_3005insTGACAGCT		0	1	1
BRIP1	c.3005G>A	p.Trp1002X	0	1	1
BRIP1	c.3072delG		0	2	0
BRIP1	c.3086delG		0	1	1
BRIP1	c.3196delT		0	5	3
BRIP1	c.3208delT		0	1	0
BRIP1	c.3240dupT		0	2	1
BRIP1	c.3260dupA		0	1	1
BRIP1	c.3390_3393delCTAT		0	8	6
BRIP1	c.3401delC		0	2	1
BRIP1	c.3440dupA		0	5	3
BRIP1	c.3525dupT		0	2	1
BRIP1	c.394dupA		0	2	0
BRIP1	c.484C>T	p.Arg162X	0	2	1
BRIP1	c.535_536delGA		0	1	0
BRIP1	c.57T>A	p.Tyr19X	0	1	0
BRIP1	c.633delT		0	4	3
BRIP1	c.674delA		0	1	1
BRIP1	c.68dupC		0	1	0
BRIP1	c.691_692insTC		0	1	0
BRIP1	c.909delT		0	1	0
BRIP1	c.918+1G>A		0	4	0
BRIP1	c.93+2T>G		0	0	1
BRIP1	c.932delA		0	1	1
BRIP1	c.939T>G	p.Tyr313X	0	1	0

CDH1	c.*20C>T		0	0	13
CDH1	c.1003C>T	p.Arg335X	0	2	0
CDH1	c.1118C>T	p.Pro373Leu	0	6	0
CDH1	c.1488_1494del7		0	2	0
CDH1	c.1921C>T	p.Gln641X	0	1	1
CDH1	c.1937-2A>G		0	0	1
CDH1	c.2291_2295+1del6		0	1	1
CDH1	c.2481T>G	p.Tyr827X	0	1	1
CDH1	c.283C>T	p.Gln95X	0	1	1
CDH1	c.387+1G>A		0	0	1
CDH1	c.532-1G>C		0	1	1
CDH1	c.715G>A	p.Gly239Arg	1	0	0
CDKN2A	c.131_132delAC		0	1	1
CDKN2A	c.131dupA		0	1	0
CDKN2A	c.159G>C	p.Met53Ile	0	2	1
CDKN2A	c.199G>C	p.Gly67Arg	0	1	0
CDKN2A	c.206A>G	p.Glu69Gly	0	2	1
CDKN2A	c.240_253del14		1	0	0
CDKN2A	c.286delG		1	0	0
CDKN2A	c.301G>T	p.Gly101Trp	1	0	0
CDKN2A	c.335_337dupGTC	p.Arg112dup	0	3	3
CDKN2A	c.-34G>C		0	0	2
CDKN2A	c.-34G>T		1	0	1
CDKN2A	c.379G>C	p.Ala127Pro	0	0	1
CDKN2A	c.457G>T	p.Asp153Tyr	2	0	0
CDKN2A	c.45G>A	p.Trp15X	0	1	0
CDKN2A	c.47T>G	p.Leu16Arg	3	0	0
CDKN2A	c.71G>C	p.Arg24Pro	1	4	1
CHEK2	c.1009-2A>G		0	0	1
CHEK2	c.1011C>A	p.Tyr337X	0	2	1
CHEK2	c.1095+1G>A		0	0	1
CHEK2	c.1100delC		18	288	133
CHEK2	c.1111_1127dup17		1	0	0
CHEK2	c.1139_1140delTC		0	1	0
CHEK2	c.1169A>C	p.Tyr390Ser	0	6	5
CHEK2	c.1188delT		0	1	1
CHEK2	c.1197dupT		0	1	0
CHEK2	c.1232G>A	p.Trp411X	0	2	2
CHEK2	c.1238delT		0	0	2
CHEK2	c.1260C>A	p.Cys420X	0	1	1
CHEK2	c.1263delT		0	12	6
CHEK2	c.1283C>T	p.Ser428Phe	3	9	0
CHEK2	c.1290dupT		0	0	1
CHEK2	c.1334dupA		0	1	0
CHEK2	c.1368dupA		0	5	4
CHEK2	c.1375+1_1375+2delGT		0	1	0
CHEK2	c.1375+1G>A		0	0	1
CHEK2	c.14_20del7		0	1	1
CHEK2	c.1427C>T	p.Thr476Met	5	71	36
CHEK2	c.1459C>T	p.Gln487X	0	4	0
CHEK2	c.1461+2T>G		0	0	1
CHEK2	c.1465G>T	p.Glu489X	0	1	0
CHEK2	c.1486C>T	p.Gln496X	0	1	0
CHEK2	c.1489delG		0	1	1
CHEK2	c.1510G>T	p.Glu504X	0	2	0
CHEK2	c.151C>T	p.Gln51X	0	1	1
CHEK2	c.152delA		0	1	1
CHEK2	c.1567delC		0	0	2

CHEK2	c.219_223del5		0	2	1
CHEK2	c.247delC		0	4	2
CHEK2	c.279G>A	p.Trp93X	0	6	2
CHEK2	c.283C>T	p.Arg95X	0	2	2
CHEK2	c.319+1G>A		0	0	2
CHEK2	c.319+2T>A		0	0	5
CHEK2	c.349A>G	p.Arg117Gly	2	26	13
CHEK2	c.372delC		0	1	0
CHEK2	c.409C>T	p.Arg137X	0	5	2
CHEK2	c.433C>T	p.Arg145Trp	0	8	2
CHEK2	c.444+1G>A		3	21	3
CHEK2	c.444+1G>T		0	1	0
CHEK2	c.444+2T>G		0	0	1
CHEK2	c.483_485delAGA	p.Glu161del	1	0	0
CHEK2	c.499G>A	p.Gly167Arg	0	7	3
CHEK2	c.507delT		0	1	1
CHEK2	c.524dupT		0	0	1
CHEK2	c.529A>T	p.Lys177X	0	1	0
CHEK2	c.575C>A	p.Ser192X	0	1	0
CHEK2	c.577_578delCT		0	2	0
CHEK2	c.58C>T	p.Gln20X	0	36	14
CHEK2	c.59	71del13	0	0	1
CHEK2	c.591delA		0	5	1
CHEK2	c.616_617delGT		0	1	0
CHEK2	c.622delG		0	1	2
CHEK2	c.629_632delCAGT		0	1	0
CHEK2	c.661_664dupATCA		0	4	2
CHEK2	c.668C>G	p.Ser223X	0	0	1
CHEK2	c.683+2T>C		0	0	1
CHEK2	c.703A>T	p.Lys235X	0	1	1
CHEK2	c.715G>T	p.Glu239X	0	2	0
CHEK2	c.726_727delAT		0	1	0
CHEK2	c.76dupA		0	0	1
CHEK2	c.792_792+1delAG		0	1	1
CHEK2	c.792+2T>C		0	1	1
CHEK2	c.817_818delGA		0	1	0
CHEK2	c.821dupT		0	1	0
CHEK2	c.823delG		0	0	1
CHEK2	c.85C>T	p.Gln29X	0	2	1
CHEK2	c.864dupT		0	1	0
CHEK2	c.876delT		0	0	1
CHEK2	c.902delT		0	0	1
CHEK2	c.915_924del10		0	1	0
CHEK2	c.917G>C	p.Gly306Ala	0	10	6
CHEK2	c.952delC		0	1	0
CHEK2	S428F	c.1283C>T	0	0	29
FANCC	c.*24C>T		0	0	9
FANCC	c.1069C>T	p.Gln357X	0	1	1
FANCC	c.1144delC		0	1	0
FANCC	c.1162G>T	p.Gly388X	1	2	0
FANCC	c.124C>T	p.Gln42X	0	1	1
FANCC	c.1257dupC		0	1	0
FANCC	c.1302dupT		0	1	0
FANCC	c.1387_1388delTC		0	1	1
FANCC	c.1453C>T	p.Gln485X	0	1	0
FANCC	c.1533+1G>C		0	2	2
FANCC	c.1598G>A	p.Trp533X	0	1	0
FANCC	c.1628C>A	p.Ser543X	0	2	0

FANCC	c.1642C>T	p.Arg548X	2	12	1
FANCC	c.1661T>C	p.Leu554Pro	0	3	1
FANCC	c.166-2A>G		0	0	1
FANCC	c.1663C>T	p.Arg555X	0	3	2
FANCC	c.166-4_166-1dupACAG		0	1	0
FANCC	c.265dupA		0	1	1
FANCC	c.267delT		0	1	1
FANCC	c.284_293del10		1	0	0
FANCC	c.288_301del14		0	1	0
FANCC	c.293_296delTTAA		0	1	1
FANCC	c.300dupA		0	1	0
FANCC	c.339G>A	p.Trp113X	0	1	0
FANCC	c.348dupT		0	2	1
FANCC	c.355_358delTCTC		0	3	2
FANCC	c.360delT		0	3	2
FANCC	c.37C>T	p.Gln13X	0	2	1
FANCC	c.455dupA		0	3	1
FANCC	c.456+4A>T		0	0	23
FANCC	c.487_490delGAGA		0	5	0
FANCC	c.489_490delGA		0	1	0
FANCC	c.500delA		0	2	2
FANCC	c.520C>T	p.Arg174X	0	4	2
FANCC	c.521+1G>A		0	2	0
FANCC	c.535C>T	p.Arg179X	0	2	1
FANCC	c.553C>T	p.Arg185X	0	15	3
FANCC	c.65G>A	p.Trp22X	0	1	1
FANCC	c.66G>A	p.Trp22X	0	1	0
FANCC	c.673G>T	p.Glu225X	0	3	1
FANCC	c.67delG		2	32	6
FANCC	c.808A>T	p.Arg270X	0	1	1
FANCC	c.843+1G>A		0	2	0
FANCC	c.844-1G>C		1	0	1
FANCC	c.916_917delGA		0	1	1
FANCC	c.91C>T	p.Gln31X	0	1	1
FANCC	c.946C>T	p.Gln316X	0	1	1
FANCC	c.992_995dupAGCA		1	0	0
FANCC	c.996+1G>T		0	3	2
MLH1	c.1011dupC		0	1	0
MLH1	c.1410-1G>C		0	1	1
MLH1	c.1410-2A>C		0	0	1
MLH1	c.1410-2A>G		0	0	3
MLH1	c.1489delC		0	1	1
MLH1	c.1489dupC		1	0	0
MLH1	c.1667G>C	p.Ser556Thr	0	1	0
MLH1	c.1791G>A	p.Trp597X	0	1	0
MLH1	c.1852_1854delAAG	p.Lys618del	1	1	0
MLH1	c.1912G>T	p.Gly638X	0	1	0
MLH1	c.199G>A	p.Gly67Arg	1	0	0
MLH1	c.2041G>A	p.Ala681Thr	1	0	0
MLH1	c.2059C>T	p.Arg687Trp	0	5	3
MLH1	c.2080G>T	p.Glu694X	0	4	3
MLH1	c.2137A>T	p.Lys713X	0	1	0
MLH1	c.244A>G	p.Thr82Ala	0	1	1
MLH1	c.306G>T	p.Glu102Asp	0	3	2
MLH1	c.350C>T	p.Thr117Met	0	1	0
MLH1	c.588delA		0	1	0
MLH1	c.676C>T	p.Arg226X	0	1	0
MLH1	c.677+3A>G		1	0	0

MLH1	c.793C>A	p.Arg265Ser	0	1	0
MRE11A	c.1015A>T	p.Lys339X	0	1	0
MRE11A	c.1048G>T	p.Glu350X	0	1	1
MRE11A	c.1069C>T	p.Gln357X	0	1	0
MRE11A	c.1090C>T	p.Arg364X	0	11	6
MRE11A	c.1112_1127del16		0	1	0
MRE11A	c.1135dupC		0	1	0
MRE11A	c.1222dupA		1	4	1
MRE11A	c.1225+2T>C		0	0	1
MRE11A	c.1441delA		0	1	1
MRE11A	c.1441dupA		0	1	0
MRE11A	c.1444	1445delCA	0	3	0
MRE11A	c.1447C>T	p.Arg483X	0	2	1
MRE11A	c.1458_1461delAGAA		0	1	1
MRE11A	c.1516G>T	p.Glu506X	0	7	1
MRE11A	c.1532dupA		0	0	1
MRE11A	c.1603G>T	p.Glu535X	0	2	0
MRE11A	c.163	167del5	0	0	1
MRE11A	c.1633_1640del8		0	1	0
MRE11A	c.170T>G	p.Leu57X	0	1	0
MRE11A	c.1714C>T	p.Arg572X	1	15	9
MRE11A	c.1720delA		0	1	0
MRE11A	c.1726C>T	p.Arg576X	0	7	2
MRE11A	c.1771C>T	p.Gln591X	0	1	0
MRE11A	c.1869delC		0	1	0
MRE11A	c.186delT		0	1	1
MRE11A	c.1897C>T	p.Arg633X	0	7	3
MRE11A	c.20dupT		0	3	3
MRE11A	c.21-6_26del12		0	3	2
MRE11A	c.402+1G>A		0	1	0
MRE11A	c.422dupT		0	1	0
MRE11A	c.592delG		0	1	0
MRE11A	c.659+1G>A		0	5	2
MRE11A	c.742G>T	p.Glu248X	0	1	1
MRE11A	c.820_821delCT		0	5	0
MRE11A	c.832G>T	p.Glu278X	0	1	0
MRE11A	c.916C>T	p.Gln306X	0	1	0
MRE11A	c.939delT		0	1	1
MRE11A	c.963dupT		0	1	0
MSH2	c.*38C>T		0	0	1
MSH2	c.1_8del8	p.Met1	0	1	0
MSH2	c.1046C>T	p.Pro349Leu	1	0	0
MSH2	c.1119delG		0	1	0
MSH2	c.119delG		0	0	1
MSH2	c.1661G>A	p.Ser554Asn	0	1	0
MSH2	c.1786_1788delAAT	p.Asn596del	0	1	1
MSH2	c.181C>T	p.Gln61X	0	1	0
MSH2	c.182delA		0	1	0
MSH2	c.1906G>C	p.Ala636Pro	0	0	1
MSH2	c.2038C>T	p.Arg680X	0	1	0
MSH2	c.2541delA		0	1	0
MSH2	c.2568T>A	p.Tyr856X	0	1	0
MSH2	c.2680dupA		0	3	3
MSH2	c.367-1G>T		0	1	1
MSH2	c.704_705delAA		0	1	0
MSH2	c.70C>T	p.Gln24X	0	1	0
MSH2	c.970_971delCA		0	1	0
MSH6	c.*2C>T		0	0	1

MSH6	c.1094G>A	p.Trp365X	1	0	0
MSH6	c.10C>T	p.Gln4X	0	4	0
MSH6	c.1135_1139del5		0	1	0
MSH6	c.1168delG		0	5	1
MSH6	c.1190_1191delAT		0	2	1
MSH6	c.1248dupT		0	1	1
MSH6	c.1306_1307dupTA		1	0	0
MSH6	c.1346T>C	p.Leu449Pro	0	3	2
MSH6	c.1350_1351delAT		0	1	0
MSH6	c.1421_1422dupTG		0	1	0
MSH6	c.1458_1459delTG		0	1	0
MSH6	c.1483C>T	p.Arg495X	0	1	0
MSH6	c.1670delG		0	0	1
MSH6	c.1772delC		0	1	0
MSH6	c.185	186delGC	0	0	1
MSH6	c.1871delG		0	1	1
MSH6	c.1937delA		0	1	0
MSH6	c.2150	2153delTCAG	0	2	1
MSH6	c.2194C>T	p.Arg732X	0	1	0
MSH6	c.2199_2200delGG		0	1	0
MSH6	c.2219T>A	p.Leu740X	0	0	1
MSH6	c.2219T>G	p.Leu740X	0	1	0
MSH6	c.2230dupG		0	1	1
MSH6	c.2269	2270delAC	0	1	0
MSH6	c.2314C>T	p.Arg772Trp	0	2	0
MSH6	c.2565delT		0	1	0
MSH6	c.2764C>T	p.Arg922X	0	1	1
MSH6	c.2779dupA		0	2	1
MSH6	c.2906A>G	p.Tyr969Cys	0	3	2
MSH6	c.3013C>T	p.Arg1005X	0	1	0
MSH6	c.3103C>T	p.Arg1035X	0	2	1
MSH6	c.3119	3120delTT	0	1	0
MSH6	c.3226C>T	p.Arg1076Cys	0	23	11
MSH6	c.3260_3261dupCC		0	1	1
MSH6	c.3261delC		0	0	17
MSH6	c.3261dupC		1	0	21
MSH6	c.3311	3312delTT	0	1	0
MSH6	c.3332_3335dupATGA		0	1	1
MSH6	c.3446T>A	p.Leu1149X	0	0	1
MSH6	c.3465_3466delGA		0	1	1
MSH6	c.3477C>G	p.Tyr1159X	0	1	0
MSH6	c.3513	3523del11	0	1	0
MSH6	c.3514dupA		0	2	1
MSH6	c.3539C>G	p.Ser1180X	0	2	1
MSH6	c.3557-2delA		0	0	6
MSH6	c.3577_3580dupGAAT		0	1	1
MSH6	c.3633dupT		0	1	1
MSH6	c.3647-2A>C		0	0	1
MSH6	c.3660	3663dupAACA	0	1	1
MSH6	c.3699_3702delAGAA		0	1	1
MSH6	c.3724_3726delCGT	p.Arg1242del	0	2	0
MSH6	c.3725G>A	p.Arg1242His	0	1	1
MSH6	c.3744_3773del30	p.His1248_Ser1257del	0	1	0
MSH6	c.3753_3756dupATTA		0	1	0
MSH6	c.3804dupA		1	0	0
MSH6	c.3939_3957dup19		1	1	1
MSH6	c.3957dupA		0	1	1
MSH6	c.3959_3962delCAAG		1	1	0

MSH6	c.3980_3983dupATCA		0	1	0
MSH6	c.3984_3987dupGTCA		0	0	2
MSH6	c.3991C>T	p.Arg1331X	0	1	0
MSH6	c.4008_4009dupTT		0	1	0
MSH6	c.4011	4014delCCTG	0	1	1
MSH6	c.4028C>G	p.Ser1343X	0	1	0
MSH6	c.4051	4069dup19	0	1	0
MSH6	c.4081	4082delTA	0	1	0
MSH6	c.467C>G	p.Ser156X	1	1	0
MSH6	c.625G>T	p.Glu209X	0	1	1
MSH6	c.718C>T	p.Arg240X	0	1	1
MSH6	c.742delC		0	1	0
MSH6	c.843_844insAC		0	1	0
MSH6	c.892C>T	p.Arg298X	0	1	0
NBN	c.*2T>G		0	0	1
NBN	c.1030C>T	p.Gln344X	0	2	1
NBN	c.105_135del31		0	0	1
NBN	c.1089C>A	p.Tyr363X	0	1	0
NBN	c.1124G>A	p.Trp375X	0	1	0
NBN	c.1142delC		0	5	1
NBN	c.114delT		0	1	1
NBN	c.1154	1155delAA	0	1	1
NBN	c.1155dupA		0	1	1
NBN	c.1210A>T	p.Lys404X	0	1	1
NBN	c.123delC		0	1	0
NBN	c.1255	1258delAATA	0	1	0
NBN	c.127C>T	p.Arg43X	0	4	4
NBN	c.1324delA		0	1	0
NBN	c.1397+1G>A		0	1	2
NBN	c.141_142delGT		0	0	1
NBN	c.1417C>T	p.Gln473X	0	1	0
NBN	c.1474C>T	p.Gln492X	0	1	0
NBN	c.1484delC		0	1	1
NBN	c.1515delG		0	1	1
NBN	c.156_157delTT		0	5	1
NBN	c.1640delC		0	1	1
NBN	c.1651dupA		0	2	0
NBN	c.1654dupG		0	1	1
NBN	c.1741C>T	p.Gln581X	0	1	0
NBN	c.181_182delGA		0	1	1
NBN	c.1882	1885delGAAG	0	1	0
NBN	c.1903A>T	p.Lys635X	0	0	3
NBN	c.1958dupA		0	1	1
NBN	c.2030_2037dup8		0	1	0
NBN	c.2071-2A>C		0	0	1
NBN	c.211_212insGA		0	0	1
NBN	c.2117C>G	p.Ser706X	0	1	0
NBN	c.2140C>T	p.Arg714X	0	5	1
NBN	c.215delC		0	1	0
NBN	c.2160delA		1	0	0
NBN	c.2184+1G>T		0	2	0
NBN	c.218delA		0	1	0
NBN	c.2206G>T	p.Glu736X	0	1	1
NBN	c.2234+2T>G		0	0	1
NBN	c.2235-2A>G		0	0	1
NBN	c.2238C>A	p.Tyr746X	0	1	1
NBN	c.232	235delGTTA	0	0	2
NBN	c.241delG		0	1	1

NBN	c.265C>T	p.Arg89X	0	2	0
NBN	c.306delT		0	1	0
NBN	c.317dupT		0	1	1
NBN	c.35_37+10del13		0	1	1
NBN	c.37+1G>A		0	3	1
NBN	c.38-2A>G		0	0	1
NBN	c.407dupG		0	1	1
NBN	c.480+1G>A		0	1	1
NBN	c.481-2A>T		0	0	1
NBN	c.496_512del17		0	2	0
NBN	c.4delT		0	1	0
NBN	c.585-2A>G		0	0	1
NBN	c.60delT		0	2	2
NBN	c.657_661del5		3	44	11
NBN	c.698_701delAACA		0	5	3
NBN	c.724delG		0	1	0
NBN	c.7A>T	p.Lys3X	0	3	2
NBN	c.803delC		0	1	0
NBN	c.808_809delGT		0	1	0
NBN	c.842T>G	p.Leu281X	0	1	0
NBN	c.88_89delAA		0	1	1
NBN	c.896+1G>T		0	1	1
NBN	c.896+2T>A		0	0	1
NF1	c.*41C>T		0	0	7
NF1	c.*48C>T		0	0	1
NF1	c.1040delA		0	0	1
NF1	c.1246C>T	p.Arg416X	0	1	1
NF1	c.1254delC		0	1	1
NF1	c.1318C>T	p.Arg440X	0	2	1
NF1	c.1399dupA		0	1	1
NF1	c.1466A>G	p.Tyr489Cys	0	3	1
NF1	c.1756_1759delACTA		0	1	0
NF1	c.1885G>A	p.Gly629Arg	0	0	1
NF1	c.1990_1993dupAACT		0	1	0
NF1	c.2033delC		1	0	1
NF1	c.2033dupC		0	0	6
NF1	c.2041C>T	p.Arg681X	0	1	1
NF1	c.2246C>G	p.Ser749X	0	0	1
NF1	c.2266delC		0	1	0
NF1	c.2395delA		0	1	1
NF1	c.246	247delTC	0	1	1
NF1	c.2850G>C	p.Gln950His	1	0	0
NF1	c.2873_2876delCTCA		0	1	1
NF1	c.2970_2972delAAT	p.Met992del	0	0	1
NF1	c.311T>G	p.Leu104X	1	0	0
NF1	c.3314+1G>A		0	0	1
NF1	c.3496+2T>A		0	0	1
NF1	c.3619delA		0	1	0
NF1	c.3826C>T	p.Arg1276X	0	1	1
NF1	c.3916C>T	p.Arg1306X	0	1	0
NF1	c.4430+1G>A		0	0	1
NF1	c.4600C>T	p.Arg1534X	0	2	0
NF1	c.4610dupA		0	1	0
NF1	c.4627delC		0	1	0
NF1	c.499	502delTGTT	0	1	0
NF1	c.574C>T	p.Arg192X	0	1	1
NF1	c.5771T>G	p.Leu1924X	0	1	0
NF1	c.6236delC		0	1	0

NF1	c.6428-1G>A		0	1	1
NF1	c.6565_6566dupCC		1	0	0
NF1	c.6576T>A	p.Tyr2192X	0	1	0
NF1	c.6638delT		0	0	1
NF1	c.6655_6656delGA		0	1	1
NF1	c.6855C>G	p.Tyr2285X	0	1	0
NF1	c.7188delA		0	0	1
NF1	c.7190-2A>T		0	0	2
NF1	c.910C>T	p.Arg304X	0	1	0
NF1	c.922dupG		0	0	1
PALB2	c.1010T>A	p.Leu337X	0	1	0
PALB2	c.1050	1053delAACA	0	2	0
PALB2	c.1059_1075del17		0	1	0
PALB2	c.1120_1123delATTC		0	1	0
PALB2	c.1140_1143delTCTT		0	2	0
PALB2	c.1240C>T	p.Arg414X	0	2	0
PALB2	c.12dupT		0	1	0
PALB2	c.1317delG		0	1	0
PALB2	c.1424dupC		1	0	0
PALB2	c.1455	1456delTA	0	1	0
PALB2	c.1479delC		0	2	2
PALB2	c.1592delT		0	1	3
PALB2	c.1616_1617dupTT		0	1	0
PALB2	c.162delA		0	1	1
PALB2	c.1633G>T	p.Glu545X	0	1	1
PALB2	c.1642	1643delTC	0	1	1
PALB2	c.1653T>A	p.Tyr551X	1	0	0
PALB2	c.1671_1674delTATT		0	1	0
PALB2	c.1675C>T	p.Gln559X	0	2	0
PALB2	c.1685-2A>G		0	0	1
PALB2	c.172_175delTTGT		0	8	1
PALB2	c.178C>T	p.Gln60X	0	1	0
PALB2	c.1919C>A	p.Ser640X	0	1	1
PALB2	c.1924delA		0	1	1
PALB2	c.196C>T	p.Gln66X	0	2	0
PALB2	c.2012T>G	p.Leu671X	0	2	2
PALB2	c.2052delC		0	2	0
PALB2	c.2074C>T	p.Gln692X	0	1	1
PALB2	c.2108T>G	p.Leu703X	0	2	1
PALB2	c.2120delC		0	2	1
PALB2	c.2167_2168delAT		0	13	7
PALB2	c.2257C>T	p.Arg753X	0	5	3
PALB2	c.2267_2283dup17		0	1	1
PALB2	c.226delA		0	1	0
PALB2	c.2296_2297delTC		1	0	0
PALB2	c.232	233delGT	0	1	1
PALB2	c.2323C>T	p.Gln775X	0	1	0
PALB2	c.2336C>G	p.Ser779X	0	2	1
PALB2	c.2386G>T	p.Gly796X	0	1	0
PALB2	c.2411	2412delCT	0	3	1
PALB2	c.2488delG		0	1	0
PALB2	c.2498	2505del8	0	1	0
PALB2	c.2515-1G>C		0	0	1
PALB2	c.2566C>T	p.Gln856X	0	1	0
PALB2	c.2570delT		0	1	1
PALB2	c.2585delA		0	1	1
PALB2	c.2587-1G>C		0	0	1
PALB2	c.2607delC		0	0	1

PALB2	c.2718G>A	p.Trp906X	1	0	0
PALB2	c.2727_2728delTT		0	1	0
PALB2	c.2730T>A	p.Tyr910X	0	2	0
PALB2	c.2748+1G>T		0	1	1
PALB2	c.2888delC		0	1	1
PALB2	c.2931dupA		0	1	0
PALB2	c.2968G>T	p.Glu990X	0	2	0
PALB2	c.2997-1G>A		0	0	1
PALB2	c.3048delT		0	1	0
PALB2	c.3113G>A	p.Trp1038X	1	15	2
PALB2	c.3116delA		1	3	0
PALB2	c.3247_3248insT		0	1	1
PALB2	c.3256C>T	p.Arg1086X	0	5	1
PALB2	c.3298	3305dup8	0	1	0
PALB2	c.3324C>G	p.Tyr1108X	0	1	0
PALB2	c.3350+4A>G		2	0	0
PALB2	c.3362delG		0	1	1
PALB2	c.3426_3429delACTT		0	1	1
PALB2	c.3427delC		0	2	2
PALB2	c.3456dupA		1	0	0
PALB2	c.3549C>A	p.Tyr1183X	1	1	0
PALB2	c.3549C>G	p.Tyr1183X	0	4	1
PALB2	c.395delT		0	2	2
PALB2	c.424A>T	p.Lys142X	0	2	0
PALB2	c.465delT		0	1	1
PALB2	c.472delC		0	0	1
PALB2	c.509_510delGA		1	8	2
PALB2	c.599delT		0	1	0
PALB2	c.62T>G	p.Leu21X	0	2	1
PALB2	c.654delA		0	1	0
PALB2	c.712A>T	p.Arg238X	0	1	0
PALB2	c.726_727insTTAGGTCTGTAGA		0	1	0
PALB2	c.734_735dupCG		0	0	1
PALB2	c.73A>T	p.Lys25X	0	1	0
PALB2	c.757_758delCT		0	2	0
PALB2	c.758dupT		0	6	2
PALB2	c.79G>T	p.Glu27X	0	2	0
PALB2	c.886delA		0	1	0
PALB2	c.945	954del10	0	0	1
PALB2	c.948delC		1	0	0
PALB2	c.979	980insATGA	0	1	0
PMS2	c.*3G>T		0	0	3
PMS2	c.1009dupA		0	1	0
PMS2	c.1017_1018dupTA		0	1	0
PMS2	c.1067delA		0	1	1
PMS2	c.1119_1122delTCAG		0	2	2
PMS2	c.121G>T	p.Glu41X	0	1	0
PMS2	c.137G>T	p.Ser46Ile	0	38	11
PMS2	c.164-1G>A		0	0	1
PMS2	c.1939A>T	p.Lys647X	0	0	1
PMS2	c.1A>G	p.Met1?	0	6	5
PMS2	c.1A>T	p.Met1?	0	1	0
PMS2	c.2192	2196del5	0	0	1
PMS2	c.23+1G>T		0	3	1
PMS2	c.241G>T	p.Glu81X	0	1	0
PMS2	c.2444C>T	p.Ser815Leu	0	0	1
PMS2	c.250+2T>G		0	0	1
PMS2	c.251-1G>C		0	0	1

PMS2	c.2T>A	p.Met1?	0	2	1
PMS2	c.2T>C	p.Met1?	0	1	0
PMS2	c.2T>G	p.Met1?	0	1	0
PMS2	c.325dupG		0	4	3
PMS2	c.353+2T>C		0	0	1
PMS2	c.400C>T	p.Arg134X	0	1	1
PMS2	c.445delT		0	1	1
PMS2	c.452delG		0	1	1
PMS2	c.520C>T	p.Gln174X	0	1	0
PMS2	c.537+1G>A		0	1	0
PMS2	c.538-2A>G		0	0	1
PMS2	c.543delT		0	1	0
PMS2	c.564dupA		0	2	0
PMS2	c.613C>T	p.Gln205X	0	1	1
PMS2	c.631C>T	p.Arg211X	0	0	1
PMS2	c.65C>A	p.Ser22X	0	1	1
PMS2	c.686_687delCT		0	1	1
PMS2	c.697C>T	p.Gln233X	0	2	1
PMS2	c.706-2A>G		0	0	1
PMS2	c.802dupT		0	0	2
PMS2	c.823C>T	p.Gln275X	0	2	1
PMS2	c.825A>G	p.=	0	2	0
PMS2	c.84delT		0	1	0
PMS2	c.861_864delACAG		0	1	2
PMS2	c.873delT		0	1	0
PMS2	c.901A>T	p.Lys301X	0	0	1
PMS2	c.903G>A	p.=	0	2	2
PMS2	c.903G>T	p.Lys301Asn	2	1	0
PMS2	c.904-1G>A		0	0	1
RAD51C	c.1005C>A	p.Cys335X	0	2	1
RAD51C	c.1026+5_1026+7delGTA		0	0	1
RAD51C	c.145+1G>A		0	0	1
RAD51C	c.181_182delCT		0	3	2
RAD51C	c.186	187delAA	0	1	1
RAD51C	c.189delT		0	1	0
RAD51C	c.216_220del5		0	1	0
RAD51C	c.224dupA		2	3	0
RAD51C	c.238G>T	p.Glu80X	0	1	0
RAD51C	c.394dupA		0	9	2
RAD51C	c.397C>T	p.Gln133X	1	1	0
RAD51C	c.404+1G>A		0	1	1
RAD51C	c.404+2T>C		0	1	0
RAD51C	c.404G>A	p.Cys135Tyr	0	1	1
RAD51C	c.414G>C	p.Leu138Phe	0	1	0
RAD51C	c.458dupG		0	1	0
RAD51C	c.498delT		0	1	1
RAD51C	c.502A>T	p.Arg168X	0	1	0
RAD51C	c.525dupC		0	2	0
RAD51C	c.571+1delG		0	1	0
RAD51C	c.571+1G>A		0	0	1
RAD51C	c.572-2A>G		0	0	1
RAD51C	c.577C>T	p.Arg193X	0	9	3
RAD51C	c.622	623delAT	0	1	1
RAD51C	c.650	651insCAAGAAACCTAAGAGGGTGGCAGGAA	0	0	1
RAD51C	c.701C>G	p.Ser234X	0	16	9
RAD51C	c.706-2A>G		0	0	2
RAD51C	c.709C>T	p.Arg237X	0	4	0

RAD51C	c.72_73insAT		0	0	1
RAD51C	c.773G>A	p.Arg258His	0	3	2
RAD51C	c.774delT		0	11	11
RAD51C	c.80_99del20		0	1	1
RAD51C	c.821delA		0	1	0
RAD51C	c.837+1G>A		0	0	1
RAD51C	c.838-2A>T		0	0	1
RAD51C	c.904+5G>T		0	0	1
RAD51C	c.905-2_905-1delAG		0	3	2
RAD51C	c.905-2A>C		0	0	3
RAD51C	c.93delG		0	8	9
RAD51C	c.942_948del7		0	1	0
RAD51C	c.955C>T	p.Arg319X	0	2	1
RAD51C	c.97C>T	p.Gln33X	0	2	1
TP53	c.1009C>T	p.Arg337Cys	1	0	1
TP53	c.1010G>A	p.Arg337His	0	2	0
TP53	c.1167delG		0	0	1
TP53	c.1175C>A	p.Ser392X	0	1	1
TP53	c.329G>T	p.Arg110Leu	0	0	1
TP53	c.374C>T	p.Thr125Met	1	0	0
TP53	c.394A>G	p.Lys132Glu	0	0	1
TP53	c.401T>G	p.Phe134Cys	0	1	1
TP53	c.455C>T	p.Pro152Leu	0	0	2
TP53	c.472C>T	p.Arg158Cys	0	2	1
TP53	c.524G>A	p.Arg175His	0	1	1
TP53	c.584T>C	p.Ile195Thr	1	0	0
TP53	c.586C>T	p.Arg196X	0	1	0
TP53	c.659A>G	p.Tyr220Cys	0	2	2
TP53	c.661G>T	p.Glu221X	0	1	0
TP53	c.686_687delGT		1	0	0
TP53	c.711G>A	p.Met237Ile	0	1	1
TP53	c.713G>A	p.Cys238Tyr	0	0	1
TP53	c.722C>T	p.Ser241Phe	0	1	0
TP53	c.742C>T	p.Arg248Trp	0	1	0
TP53	c.743G>A	p.Arg248Gln	0	5	5
TP53	c.743G>T	p.Arg248Leu	1	0	0
TP53	c.752T>G	p.Ile251Ser	0	0	1
TP53	c.772G>A	p.Glu258Lys	0	1	0
TP53	c.817C>T	p.Arg273Cys	0	2	0
TP53	c.818G>A	p.Arg273His	0	0	3
TP53	c.818G>T	p.Arg273Leu	0	1	0
TP53	c.830G>A	p.Cys277Tyr	0	0	1
TP53	c.844C>T	p.Arg282Trp	0	1	1
TP53	c.850_860del11		1	0	0
TP53	c.869_870insGCGGAAACCG		0	1	1

Abbreviations: AC, allele count; gnomAD, Genome Aggregation Database; ExAC (ExAC_non_TCGA)Exome Aggregation Consortium_nonTCGA.

eTable 4. Frequency of Mutation Carriers by Gene Among Pancreatic Cancer Cases With Personal and Family History of Cancers

Gene	Overall (n=3030)	Personal History		Family History							
		Other cancers (n=513)		Pancreatic cancer (n=343)		Breast cancer (n=675)		Colorectal cancer (n=513)		Common cancers[a] (n=1233)	
	N	N	% by gene	N	% by gene	N	% by gene	N	% by gene	N	% by gene
ATM	69	14	20.3	11	15.9	18	26.1	10	14.5	30	43.5
BARD1	4	0	0.0	1	25.0	0	0.0	0	0.0	1	25.0
BRCA1	18	6	33.3	2	11.1	4	22.2	2	11.1	6	33.3
BRCA2	59	14	23.7	7	11.9	22	37.3	10	16.9	32	54.2
BRIP1	5	0	0.0	1	20.0	0	0.0	0	0.0	2	40.0
CDH1	1	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0
CDKN2A	10	2	20.0	5	50.0	2	20.0	2	20.0	6	60.0
CHEK2	33	9	27.3	8	24.2	11	33.3	5	15.2	18	54.5
FANCC	8	2	25.0	1	12.5	1	12.5	1	12.5	4	50.0
MLH1	5	3	60.0	0	0.0	1	20.0	3	60.0	3	60.0
MRE11A	2	0	0.0	0	0.0	1	50.0	0	0.0	1	50.0
MSH2	1	0	0.0	1	100.0	1	100.0	1	100.0	1	100.0
MSH6	7	3	42.9	1	14.3	3	42.9	4	57.1	4	57.1
NBN	4	1	25.0	1	25.0	1	25.0	1	25.0	2	50.0
NF1	4	3	75.0	0	0.0	1	25.0	1	25.0	2	50.0
PALB2	12	3	25.0	2	16.7	5	41.7	3	25.0	6	50.0
PMS2	2	2	100.0	2	100.0	1	50.0	2	100.0	2	100.0
RAD51C	3	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0
TP53	6	3	50.0	2	33.3	4	66.7	0	0.0	4	66.7
All genes	**253**	**65**	**25.7**	**45**	**17.8**	**76**	**30.0**	**43**	**17.0**	**124**	**49.0**

[a] Family history of any pancreatic, breast, colorectal, non-ovarian gynecologic, or ovary cancer.

eTable 5. Frequency of Mutations in Individual Genes Among Cases With Pancreatic Cancer and ExAC_non_TCGA Reference Controls From Different Racial and Ethnic Populations

	Cases								ExAC_non_TCGA Controls							
Gene	AFR	AMR	SAS/ EAS	NFE	Total AFR	Total AMR	Total SAS/ EAS	Total NFE	AFR	AMR	SAS/ EAS	NFE	Total AFR	Total AMR	Total SAS/ EAS	Total NFE
ATM	3	0	1	65	50	42	11	2896	13	22	39	102	4429	5505	11973	26644
BARD1	0	0	0	4	50	42	11	2896	3	3	12	27	4259	5413	11695	26078
BRCA1	0	0	0	18	50	42	11	2896	1	6	30	85	4478	5556	12059	26911
BRCA2	0	2	0	55	50	42	11	2896	12	27	33	111	4373	5557	11987	26791
BRIP1	1	0	0	4	50	42	11	2896	14	7	21	51	4470	5543	12044	26840
CDH1	0	0	0	1	50	42	11	2896	1	3	13	3	4323	5343	11741	25961
CDKN2A	0	0	0	9	50	42	11	2896	3	0	0	8	3502	5183	11382	24312
CHEK2	0	0	0	33	50	42	11	2896	11	14	25	257	4147	5249	11604	25215
FANCC	0	0	0	8	50	42	11	2896	7	3	15	50	4398	5403	11830	26434
MLH1	0	0	0	4	50	42	11	2896	3	1	4	7	4405	5492	11945	26639
MRE11A	0	0	0	2	50	42	11	2896	0	1	13	25	4463	5514	12002	26767
MSH2	0	0	0	1	50	42	11	2896	0	1	0	7	4128	5058	11395	25105
MSH6	0	0	0	6	50	42	11	2896	18	11	26	36	4280	5409	11665	26151
NBN	0	0	0	4	50	42	11	2896	5	7	11	41	4368	5467	11900	26264
NF1	0	0	0	4	50	42	11	2896	5	2	6	27	4360	5402	11810	26130
PALB2	0	1	0	11	50	42	11	2896	7	11	11	30	4463	5540	12040	26869
PMS2	0	0	0	2	50	42	11	2896	3	8	13	29	4028	5110	11390	24617
RAD51C	0	0	0	3	50	42	11	2896	4	2	20	37	4430	5533	12002	26647
TP53	0	0	0	6	50	42	11	2896	3	0	5	18	4413	5477	11963	26789

Abbreviations: AFR, African American; AMR, Hispanic; SAS, South Asian; EAS, East Asian; NFE, non-Finnish European; ExAC_non_TCGA, Exome Aggregation Consortium controls excluding cancer cases from the TCGA study; TCGA, The Cancer Genome Atlas.

eTable 6. Associations Between Panel Gene Mutations and Pancreatic Cancer Using ExAC_non_TCGA Reference Controls										
Gene	Cases[a]			ExAC_ non_TCGA Controls			Cancer risk[b]			
	Cases with Mutations	Individuals Tested	Carrier Frequency (%)	Controls with Mutations	Individuals Tested	Carrier Frequency (%)	OR	95% CI lower	95% CI upper	Adjusted p-value[c]
CDKN2A	9	2999	0.30	11	44379	0.02	9.06	3.8	20.45	<.001
ATM	69	2999	2.30	176	48551	0.36	6.15	4.63	8.09	<.001
MLH1	4	2999	0.13	15	48481	0.03	4.97	1.40	14.03	.096
BRCA2	57	2999	1.90	183	48708	0.38	4.68	3.45	6.25	<.001
PALB2	12	2999	0.40	59	48912	0.12	3.53	1.80	6.37	.002
TP53	6	2999	0.20	26	48642	0.05	3.03	1.14	6.74	.19
CDH1	1	2999	0.03	20	47368	0.04	2.62	0.14	15.31	>.99
BRCA1	18	2999	0.60	122	49004	0.25	1.96	1.16	3.10	.14
MSH6	6	2999	0.20	91	47505	0.19	1.39	0.54	2.96	>.99
NF1	4	2999	0.13	45	47445	0.09	1.30	0.39	3.20	>.99
BARD1	4	2999	0.13	40	47702	0.08	1.30	0.39	3.20	>.99
MSH2	1	2999	0.03	8	45686	0.02	1.23	0.07	6.19	>.99
CHEK2	33	2999	1.10	307	46215	0.66	1.11	0.76	1.56	>.99
NBN	5	2999	0.17	93	48897	0.19	0.87	0.31	1.93	>.99
RAD51C	4	2999	0.13	64	47999	0.13	0.86	0.26	2.08	>.99
MRE11A	2	2999	0.07	39	48746	0.08	0.73	0.12	2.38	>.99
BRIP1	3	2999	0.10	63	48612	0.13	0.73	0.18	1.97	>.99
FANCC	8	2999	0.27	75	48066	0.16	0.64	2.77	0.33	>.99
PMS2	2	2999	0.07	53	45145	0.12	0.57	0.09	1.82	>.99

Abbreviations: ExAC, Exome Aggregation Consortium; ExAC_non_TCGA, Exome Aggregation Consortium controls excluding cancer cases from the TCGA study; TCGA, The Cancer Genome Atlas; OR, odds ratio.

[a] Analyses do not include cases with missing (n=12) and other (n=19) race/ethnicity, leaving 2999 of 3030.

[b] Logistic regression weighted by race and ethnicity

[c] Adjusted by Bonferroni correction for 19 genes.

eTable 7. Mutation Frequencies for Individual Genes Among Non-Hispanic White Cases With Pancreatic Cancer by Personal and Family History of Cancers

Gene	Overall (n=2896)		Personal History		Family History					
			Other cancers (n=499)		Pancreatic cancer (n=334)		Breast cancer (n=662)		Colorectal cancer (n=505)	
	N	%	N	%	N	%	N	%	N	%
ATM	65	2.24	14	2.81	11	3.21	18	2.67	10	1.95
BARD1	4	0.14	0	0.00	1	0.29	0	0.00	0	0.00
BRCA1	18	0.62	6	1.20	2	0.58	4	0.59	2	0.39
BRCA2	55	1.90	14	2.81	7	2.04	22	3.26	10	1.95
BRIP1	4	0.14	0	0.00	1	0.29	0	0.00	0	0.00
CDH1	1	0.03	0	0.00	0	0.00	0	0.00	0	0.00
CDKN2A	9	0.31	2	0.40	5	1.46	2	0.30	2	0.39
CHEK2	33	1.14	9	1.80	8	2.33	11	1.63	5	0.97
FANCC	8	0.28	2	0.40	1	0.29	1	0.15	1	0.19
MLH1	4	0.14	3	0.60	0	0.00	1	0.15	3	0.58
MRE11A	2	0.07	0	0.00	0	0.00	1	0.15	0	0.00
MSH2	1	0.03	0	0.00	1	0.29	1	0.15	1	0.19
MSH6	6	0.21	3	0.60	1	0.29	3	0.44	4	0.78
NBN	4	0.14	1	0.20	1	0.29	1	0.15	1	0.19
NF1	4	0.14	3	0.60	0	0.00	1	0.15	1	0.19
PALB2	11	0.38	3	0.60	2	0.58	5	0.74	3	0.58
PMS2	2	0.07	0	0.00	0	0.00	1	0.15	2	0.39
RAD51C	3	0.10	0	0.00	0	0.00	0	0.00	0	0.00
TP53	6	0.21	3	0.60	2	0.58	4	0.59	0	0.00
All genes	**240**	**8.29**	**63**	**12.62**	**43**	**12.52**	**76**	**11.27**	**45**	**8.74**

eTable 8. Comparisons of Mutation Frequencies Between Non-Hispanic White Cases With Pancreatic Cancer and gnomAD_NFE Controls

	Cases				gnomAD_NFE Controls				Cancer risk[a]			
Gene	Cases with Mutations	Individuals Tested	Carrier Frequency (%)		Controls with Mutations	Individuals Tested	Carrier Frequency (%)		OR	95% CI lower	95% CI upper	Adjusted p-value[b]
CDKN2A	9	2896	0.32		13	52145	0.02		12.48	5.15	29.11	<.001
MLH1	4	2896	0.14		11	55479	0.02		6.97	2.05	22.00	.10
TP53	6	2896	0.20		17	55753	0.04		6.80	2.63	17.65	.013
BRCA2	55	2896	1.90		172	55214	0.32		6.15	4.47	8.34	<.001
ATM	65	2896	2.24		231	55657	0.42		5.46	4.11	7.19	<.001
NF1	4	2896	0.14		20	55550	0.04		3.84	1.20	11.40	.57
BRCA1	18	2896	0.62		132	55710	0.24		2.63	1.58	4.31	.009
PALB2	11	2896	0.38		96	55754	0.18		2.21	1.11	4.13	.38
CDH1	1	2896	0.04		8	54597	0.02		2.36	0.11	15.03	>.99
MSH6	6	2896	0.20		56	55060	0.10		2.04	0.85	4.75	>.99
FANCC	8	2896	0.28		89	55694	0.16		1.73	0.77	3.50	>.99
MSH2	1	2896	0.04		12	55350	0.02		1.59	0.08	9.92	>.99
BARD1	4	2896	0.14		56	54802	0.10		1.35	0.45	3.72	>.99
CHEK2	33	2896	1.14		475	54887	0.86		1.32	0.91	1.89	>.99
RAD51C	3	2896	0.10		51	55740	0.10		1.13	0.30	3.52	>.99
NBN	4	2896	0.14		87	55583	0.16		0.88	0.30	2.33	>.99
MRE11A	2	2896	0.06		53	55663	0.10		0.73	0.13	2.74	>.99
PMS2	2	2896	0.06		52	54420	0.10		0.72	0.13	2.73	>.99
BRIP1	4	2896	0.14		119	55698	0.22		0.65	0.22	1.68	>.99

Abbreviations: gnomAD, Genome Aggregation Database; NFE, non-Finnish European; OR, odds ratio.
[a] Fishers exact test.
[b] Adjusted by Bonferroni correction of 19 genes.

eTable 9. Associations of Panel Gene Mutations With Pancreatic Cancer Based on Non-Hispanic white Patients With Pancreatic Cancer and ExAC_NFE_non-TCGA Reference Controls

Gene	Cases			ExAC_NFE_non_TCGA Controls			Cancer risk[a]			
	Cases with Mutations	Individuals Tested	Carrier Frequency (%)	Controls with Mutations	Individuals Tested	Carrier Frequency (%)	OR	95% CI lower	95% CI upper	Adjusted p-value[b]
CDKN2A	9	2896	0.32	8	24312	0.04	9.46	3.39	24.82	<.001
ATM	65	2896	2.24	102	26644	0.38	5.92	4.30	8.16	<.001
MLH1	4	2896	0.14	7	26639	0.02	5.26	1.44	18.44	.38
BRCA2	55	2896	1.90	111	26791	0.42	4.62	3.29	6.40	<.001
PALB2	11	2896	0.38	30	26869	0.12	3.41	1.68	6.91	.03
TP53	6	2896	0.20	18	26789	0.06	3.09	1.20	7.82	.38
CDH1	1	2896	0.04	3	25961	0.02	2.99	0.12	27.10	>.99
BRCA1	18	2896	0.62	85	26911	0.32	1.97	1.15	3.30	.19
MSH6	6	2896	0.20	36	26151	0.14	1.51	0.62	3.58	>.99
FANCC	8	2896	0.28	50	26434	0.18	1.46	0.63	3.03	>.99
NF1	4	2896	0.14	27	26130	0.10	1.34	0.43	3.66	>.99
BARD1	4	2896	0.14	27	26078	0.10	1.33	0.43	3.65	>.99
MSH2	1	2896	0.04	7	25105	0.02	1.24	0.06	8.67	>.99
CHEK2	33	2896	1.14	257	25215	1.02	1.12	0.76	1.62	>.99
NBN	4	2896	0.14	41	26264	0.16	0.88	0.29	2.40	>.99
RAD51C	3	2896	0.10	37	26647	0.14	0.75	0.19	2.27	>.99
MRE11A	2	2896	0.06	25	26767	0.10	0.74	0.12	2.87	>.99
BRIP1	4	2896	0.14	51	26840	0.20	0.73	0.24	2.02	>.99
PMS2	2	2896	0.06	29	24617	0.12	0.59	0.10	2.21	>.99

Abbreviations: ExAC, Exome Aggregation Consortium; NFE, non-Finnish European; TCGA, The Cancer Genome Atlas; OR, odds ratio.

[a] Fishers exact test.

[b] Adjusted by Bonferroni correction of 19 genes.

eTable 10. Comparisons of Mutation Frequencies Between Patients With Pancreatic Cancer as the Initial Cancer and gnomAD Controls

Gene	Cases			gnomAD Controls			Cancer risk[a]			
	Cases with Mutations	Individuals Tested	Carrier Frequency (%)	Controls with Mutations	Individuals Tested	Carrier Frequency (%)	OR	95% CI lower	95% CI upper	Adjusted p-value[c]
ATM	56	2509	2.23	386	104016	0.37	5.54	4.14	7.28	<.001
BARD1	4	2509	0.16	86	102189	0.08	1.58	0.48	3.76	>.99
BRCA1	13	2509	0.52	208	104122	0.20	2.23	1.21	3.74	.10
BRCA2	41	2509	1.63	313	102739	0.31	5.58	3.99	7.60	<.001
BRIP1	4	2509	0.16	194	104071	0.19	0.94	0.33	2.04	>.99
CDH1	1	2509	0.04	15	102110	0.02	2.76	0.15	13.64	>.99
CDKN2A	7	2509	0.28	15	99493	0.02	11.49	4.57	25.30	<.001
CHEK2	24	2509	0.96	572	102856	0.56	1.14	0.74	1.67	>.99
FANCC	6	2509	0.24	129	104042	0.12	1.51	0.59	3.13	>.99
MLH1	1	2509	0.04	25	103526	0.02	1.99	0.11	9.50	>.99
MRE11A	2	2509	0.08	96	104071	0.10	0.85	0.14	2.67	>.99
MSH2	1	2509	0.04	16	103327	0.02	1.90	0.11	9.05	>.99
MSH6	3	2509	0.12	101	102802	0.10	1.18	0.29	3.14	>.99
NBN	3	2509	0.12	125	103912	0.12	0.77	0.19	2.04	>.99
NF1	2	2509	0.08	31	103812	0.03	2.21	0.36	7.21	>.99
PALB2	8	2509	0.32	153	104169	0.15	2.09	0.99	3.85	.62
PMS2	2	2509	0.08	86	101976	0.08	0.84	0.14	2.66	>.99
RAD51C	3	2509	0.12	94	104128	0.09	1.34	0.33	3.56	>.99
TP53	3	2509	0.12	25	104163	0.02	4.01	0.96	11.23	.43

Abbreviations: gnomAD, Genome Aggregation Database; OR, odds ratio.

[a] Logistic regression weighted by race and ethnicity.

[b] Analyses do not include cases with missing and other race/ethnicity, leaving 2,509 of 2,535.

[c] Adjusted by Bonferroni correction of 19 genes.

eTable 11. Comparisons of Mutation Frequencies Between Cases With Pancreatic Cancer With Family History of Pancreatic, Breast, Ovarian, Colorectal, or Gynecologic Cancer and gnomAD Controls

Gene	Cases			gnomAD Controls			Cancer risk[a]			
	Cases with Mutations	Individuals Tested	Carrier Frequency (%)	Controls with Mutations	Individuals Tested	Carrier Frequency (%)	OR	95% CI lower	95% CI upper	Adjusted p-value[b]
ATM	30	1227	2.44	386	104016	0.37	6.04	4.07	8.63	<.001
BARD1	1	1227	0.08	86	102189	0.08	0.80	0.05	3.60	>.99
BRCA1	6	1227	0.49	208	104122	0.20	2.09	0.82	4.29	>.99
BRCA2	32	1227	2.61	313	102739	0.31	8.56	5.81	12.16	<.001
BRIP1	2	1227	0.16	194	104071	0.19	0.77	0.13	2.39	>.99
CDH1	0	1227	0.00	15	102110	0.02	0.00	NA	2.93	>.99
CDKN2A	6	1227	0.49	15	99493	0.02	19.95	7.39	45.72	<.001
CHEK2	18	1227	1.47	572	102856	0.56	1.73	1.04	2.68	.40
FANCC	4	1227	0.33	129	104042	0.12	2.06	0.63	4.87	>.99
MLH1	3	1227	0.24	25	103526	0.02	12.30	2.90	35.79	<.001
MRE11A	1	1227	0.08	96	104071	0.10	0.86	0.05	3.86	>.99
MSH2	1	1227	0.08	16	103327	0.02	3.82	0.21	18.19	>.99
MSH6	4	1227	0.33	101	102802	0.10	3.23	0.99	7.70	.38
NBN	2	1227	0.16	125	103912	0.12	1.05	0.17	3.29	>.99
NF1	2	1227	0.16	31	103812	0.03	4.53	0.74	14.81	.76
PALB2	6	1227	0.49	153	104169	0.15	2.85	1.12	5.90	.19
PMS2	2	1227	0.16	86	101976	0.08	1.71	0.28	5.40	>.99
RAD51C	0	1227	0.00	94	104128	0.09	0.00	0.00	0.00	>.99
TP53	4	1227	0.33	25	104163	0.02	10.84	3.22	27.41	<.001

Abbreviations: gnomAD, Genome Aggregation Database; OR, odds ratio.
[a] Logistic regression weighted by race and ethnicity.
[b] Adjusted by Bonferroni correction of 19 genes.

eTable 12. Comparisons of Mutation Frequencies Between Cases With Pancreatic Cancer Without Family History of Pancreatic, Breast, Ovarian, Colorectal, or Gynecologic Cancer and gnomAD Controls

Gene	Cases			gnomAD Controls			Cancer risk[a]			
	Cases with Mutations	Individuals Tested	Carrier Frequency (%)	Controls with Mutations	Individuals Tested	Carrier Frequency (%)	OR	95% CI lower	95% CI upper	Adjusted p-value[b]
ATM	39	1772	2.20	386	104016	0.37	5.48	3.87	7.53	<.001
BARD1	3	1772	0.17	86	102189	0.08	1.68	0.41	4.46	>.99
BRCA1	12	1772	0.68	208	104122	0.20	2.93	1.55	5.01	.006
BRCA2	25	1772	1.41	313	102739	0.31	4.58	2.97	6.76	<.001
BRIP1	3	1772	0.17	194	104071	0.19	0.80	0.20	2.09	>.99
CDH1	1	1772	0.06	15	102110	0.02	3.90	0.22	19.30	>.99
CDKN2A	3	1772	0.17	15	99493	0.02	6.99	1.66	20.02	.03
CHEK2	15	1772	0.85	572	102856	0.56	1.01	0.58	1.63	>.99
FANCC	4	1772	0.23	129	104042	0.12	1.43	0.44	3.38	>.99
MLH1	1	1772	0.06	25	103526	0.02	2.80	0.16	13.41	>.99
MRE11A	1	1772	0.06	96	104071	0.10	0.60	0.03	2.69	>.99
MSH2	0	1772	0.00	16	103327	0.02	0.00	NA	0.10	>.99
MSH6	2	1772	0.11	101	102802	0.10	1.12	0.18	3.53	>.99
NBN	2	1772	0.11	125	103912	0.12	0.73	0.12	2.29	>.99
NF1	2	1772	0.11	31	103812	0.03	3.12	0.51	10.21	>.99
PALB2	6	1772	0.34	153	104169	0.15	1.97	0.78	4.08	>.99
PMS2	0	1772	0.00	86	101976	0.08	0.00	0.00	0.00	>.99
RAD51C	3	1772	0.17	94	104128	0.09	1.90	0.47	5.05	>.99
TP53	2	1772	0.11	25	104163	0.02	3.80	0.61	12.55	>.99

Abbreviations: gnomAD, Genome Aggregation Database; OR, odds ratio.

[a] Logistic regression weighted by race and ethnicity.

[b] Adjusted by Bonferroni correction of 19 genes.

eTable 13. Comparisons of mutation Frequencies Between Cases With Pancreatic Cancer, Excluding Those With Family History of Pancreatic Cancer, and gnomAD Controls

Gene	Cases			gnomAD Controls			Cancer risk[a]			
	Cases with Mutations	Individuals Tested	Carrier Frequency (%)	Controls with Mutations	Individuals Tested	Carrier Frequency (%)	OR	95% CI lower	95% CI upper	Adjusted p-value[b]
ATM	58	2659	2.18	386	104016	0.37	5.41	4.06	7.07	<.001
BARD1	3	2659	0.11	86	102189	0.08	1.12	0.27	2.96	>.99
BRCA1	16	2659	0.60	208	104122	0.20	2.59	1.50	4.16	.004
BRCA2	50	2659	1.88	313	102739	0.31	6.13	4.49	8.20	<.001
BRIP1	4	2659	0.15	194	104071	0.19	0.71	0.22	1.67	>.99
CDH1	1	2659	0.04	15	102110	0.02	2.60	0.14	12.85	>.99
CDKN2A	4	2659	0.15	15	99493	0.02	6.18	1.82	15.99	.01
CHEK2	25	2659	0.94	572	102856	0.56	1.12	0.73	1.63	>.99
FANCC	7	2659	0.26	129	104042	0.12	1.67	0.71	3.29	>.99
MLH1	4	2659	0.15	25	103526	0.02	7.51	2.19	19.76	.004
MRE11A	2	2659	0.08	96	104071	0.10	0.80	0.13	2.52	>.99
MSH2	0	2659	0.00	16	103327	0.02	0.00	NA	0.02	>.99
MSH6	5	2659	0.19	101	102802	0.10	1.86	0.66	4.12	>.99
NBN	3	2659	0.11	125	103912	0.12	0.73	0.18	1.92	>.99
NF1	4	2659	0.15	31	103812	0.03	4.17	1.25	10.40	.13
PALB2	10	2659	0.38	153	104169	0.15	2.19	1.08	3.93	>.99
PMS2	2	2659	0.08	86	101976	0.08	0.79	0.13	2.50	>.99
RAD51C	3	2659	0.11	94	104128	0.09	1.26	0.31	3.35	>.99
TP53	4	2659	0.15	25	104163	0.02	5.04	1.50	12.74	.04

Abbreviations: gnomAD, Genome Aggregation Database; OR, odds ratio.

[a] Logistic regression weighted by race and ethnicity.

[b] Adjusted by Bonferroni correction of 19 genes.

eTable 14. Comparisons of Mutation Frequencies Between Cases With Pancreatic Cancer, Excluding Those With Family History of Breast Cancer, and gnomAD Controls

Gene	Cases			gnomAD Controls			Cancer risk[a]			
	Cases with Mutations	Individuals Tested	Carrier Frequency (%)	Controls with Mutations	Individuals Tested	Carrier Frequency (%)	OR	95% CI lower	95% CI upper	Adjusted p-value[b]
ATM	51	2326	2.19	386	104016	0.37	5.45	4.01	7.23	<.001
BARD1	4	2326	0.17	86	102189	0.08	1.70	0.52	4.06	>.99
BRCA1	14	2326	0.60	208	104122	0.20	2.60	1.44	4.28	.02
BRCA2	35	2326	1.50	313	102739	0.31	4.89	3.38	6.85	<.001
BRIP1	5	2326	0.22	194	104071	0.19	1.01	0.36	2.21	>.99
CDH1	1	2326	0.04	15	102110	0.02	2.97	0.16	14.70	>.99
CDKN2A	7	2326	0.30	15	99493	0.02	12.41	4.94	27.34	<.001
CHEK2	22	2326	0.95	572	102856	0.56	1.13	0.72	1.68	>.99
FANCC	7	2326	0.30	129	104042	0.12	1.91	0.81	3.77	>.99
MLH1	3	2326	0.13	25	103526	0.02	6.43	1.52	18.68	.05
MRE11A	1	2326	0.04	96	104071	0.10	0.46	0.03	2.05	>.99
MSH2	0	2326	0.00	16	103327	0.02	0.00	NA	0.02	>.99
MSH6	3	2326	0.13	101	102802	0.10	1.28	0.31	3.39	>.99
NBN	3	2326	0.13	125	103912	0.12	0.84	0.21	2.20	>.99
NF1	3	2326	0.13	31	103812	0.03	3.57	0.86	9.88	.60
PALB2	7	2326	0.30	153	104169	0.15	1.75	0.74	3.46	>.99
PMS2	1	2326	0.04	86	101976	0.08	0.45	0.03	2.03	>.99
RAD51C	3	2326	0.13	94	104128	0.09	1.44	0.35	3.84	>.99
TP53	2	2326	0.09	25	104163	0.02	2.88	0.47	9.53	>.99

Abbreviations: gnomAD, Genome Aggregation Database; OR, odds ratio.
[a] Logistic regression weighted by race and ethnicity.
[b] Adjusted by Bonferroni correction of 19 genes.

eTable 15. Comparisons of Mutation Frequencies Between Cases With Pancreatic Cancer, Excluding Those With Family History of Ovarian Cancer, and gnomAD Controls

Gene	Cases			gnomAD Controls			Cancer risk[a]			
	Cases with Mutations	Individuals Tested	Carrier Frequency (%)	Controls with Mutations	Individuals Tested	Carrier Frequency (%)	OR	95% CI lower	95% CI upper	Adjusted p-value[b]
ATM	63	2849	2.21	386	104016	0.37	5.49	4.16	7.11	<.001
BARD1	3	2849	0.11	86	102189	0.08	1.04	0.26	2.76	>.99
BRCA1	16	2849	0.56	208	104122	0.20	2.42	1.40	3.88	.01
BRCA2	53	2849	1.86	313	102739	0.31	6.07	4.48	8.06	<.001
BRIP1	4	2849	0.14	194	104071	0.19	0.66	0.20	1.55	>.99
CDH1	1	2849	0.04	15	102110	0.02	2.43	0.13	11.99	>.99
CDKN2A	8	2849	0.28	15	99493	0.02	11.55	4.86	24.62	<.001
CHEK2	29	2849	1.02	572	102856	0.56	1.21	0.82	1.72	>.99
FANCC	8	2849	0.28	129	104042	0.12	1.78	0.80	3.38	>.99
MLH1	4	2849	0.14	25	103526	0.02	7.01	2.04	18.45	.007
MRE11A	2	2849	0.07	96	104071	0.10	0.75	0.12	2.35	>.99
MSH2	1	2849	0.04	16	103327	0.02	1.67	0.09	7.95	>.99
MSH6	6	2849	0.21	101	102802	0.10	2.09	0.81	4.36	>.99
NBN	4	2849	0.14	125	103912	0.12	0.91	0.28	2.15	>.99
NF1	3	2849	0.11	31	103812	0.03	2.92	0.70	8.07	>.99
PALB2	11	2849	0.39	153	104169	0.15	2.25	1.15	3.95	.17
PMS2	1	2849	0.04	86	101976	0.08	0.37	0.02	1.66	>.99
RAD51C	3	2849	0.11	94	104128	0.09	1.17	0.29	3.12	>.99
TP53	6	2849	0.21	25	104163	0.02	7.06	2.65	15.76	<.001

Abbreviations: gnomAD, Genome Aggregation Database; OR, odds ratio.
[a] Logistic regression weighted by race and ethnicity.
[b] Adjusted by Bonferroni correction of 19 tested genes.

eTable 16. Comparisons of Mutation Frequencies Between Cases With Pancreatic Cancer, Excluding Those With Family History of Colorectal Cancer, and gnomAD Controls

Gene	Cases			gnomAD Controls			Cancer risk[a]			
	Cases with Mutations	Individuals Tested	Carrier Frequency (%)	Controls with Mutations	Individuals Tested	Carrier Frequency (%)	OR	95% CI lower	95% CI upper	Adjusted p-value[b]
ATM	59	2487	2.37	386	104016	0.37	5.90	4.44	7.70	<.001
BARD1	4	2487	0.16	86	102189	0.08	1.59	0.49	3.80	>.99
BRCA1	16	2487	0.64	208	104122	0.20	2.77	1.60	4.45	.002
BRCA2	47	2487	1.89	313	102739	0.31	6.17	4.47	8.31	<.001
BRIP1	5	2487	0.20	194	104071	0.19	0.95	0.34	2.06	>.99
CDH1	1	2487	0.04	15	102110	0.02	2.78	0.15	13.75	>.99
CDKN2A	7	2487	0.28	15	99493	0.02	11.60	4.61	25.54	<.001
CHEK2	28	2487	1.13	572	102856	0.56	1.35	0.90	1.92	>.99
FANCC	7	2487	0.28	129	104042	0.12	1.78	0.76	3.52	>.99
MLH1	1	2487	0.04	25	103526	0.02	2.00	0.11	9.58	>.99
MRE11A	2	2487	0.08	96	104071	0.09	0.86	0.14	2.70	>.99
MSH2	0	2487	0.00	16	103327	0.02	0.00	NA	0.02	>.99
MSH6	2	2487	0.08	101	102802	0.10	0.80	0.13	2.51	>.99
NBN	3	2487	0.12	125	103912	0.12	0.78	0.19	2.06	>.99
NF1	3	2487	0.12	31	103812	0.03	3.34	0.81	9.24	.76
PALB2	9	2487	0.36	153	104169	0.15	2.11	1.00	3.89	.57
PMS2	0	2487	0.00	86	101976	0.08	0.00	0.00	0.00	>.99
RAD51C	3	2487	0.12	94	104128	0.09	1.35	0.33	3.59	>.99
TP53	6	2487	0.24	25	104163	0.02	8.10	3.05	18.10	<.001

Abbreviations: gnomAD, Genome Aggregation Database; OR, odds ratio.
[a] Logistic regression weighted by race and ethnicity.
[b] Adjusted by Bonferroni correction of 19 tested genes.

eTable 17. Comparisons of Mutation Frequencies Between Cases With Pancreatic Cancer, Excluding Those With Family History of Gynecologic Cancer, and gnomAD Controls

Gene	Cases			gnomAD Controls			Cancer risk[a]			
	Cases with Mutations	Individuals Tested	Carrier Frequency (%)	Controls with Mutations	Individuals Tested	Carrier Frequency (%)	OR	95% CI lower	95% CI upper	Adjusted p-value[b]
ATM	67	2837	2.36	386	104016	0.37	5.87	4.49	7.56	<.001
BARD1	3	2837	0.11	86	102189	0.08	1.05	0.26	2.78	>.99
BRCA1	17	2837	0.60	208	104122	0.20	2.58	1.52	4.09	.003
BRCA2	52	2837	1.83	313	102739	0.31	5.98	4.40	7.95	<.001
BRIP1	5	2837	0.18	194	104071	0.19	0.83	0.29	1.81	>.99
CDH1	1	2837	0.04	15	102110	0.02	2.44	0.13	12.05	>.99
CDKN2A	8	2837	0.28	15	99493	0.02	11.6	4.88	24.72	<.001
CHEK2	30	2837	1.06	572	102856	0.56	1.26	0.85	1.78	>.99
FANCC	7	2837	0.25	129	104042	0.12	1.56	0.66	3.08	>.99
MLH1	3	2837	0.11	25	103526	0.02	5.28	1.25	15.34	.13
MRE11A	2	2837	0.07	96	104071	0.09	0.75	0.12	2.36	>.99
MSH2	0	2837	0.00	16	103327	0.02	0.00	NA	0.01	>.99
MSH6	5	2837	0.18	101	102802	0.10	1.75	0.62	3.86	>.99
NBN	4	2837	0.14	125	103912	0.12	0.91	0.28	2.16	>.99
NF1	4	2837	0.14	31	103812	0.03	3.91	1.17	9.74	.18
PALB2	11	2837	0.39	153	104169	0.15	2.26	1.16	3.96	.17
PMS2	2	2837	0.07	86	101976	0.08	0.74	0.12	2.35	>.99
RAD51C	3	2837	0.11	94	104128	0.09	1.18	0.29	3.14	>.99
TP53	6	2837	0.21	25	104163	0.02	7.09	2.67	15.83	<.001

Abbreviations: gnomAD, Genome Aggregation Database; OR, odds ratio.
[a] Logistic regression weighted by race and ethnicity.
[b] Adjusted by Bonferroni correction of 19 tested genes.

eTable 18. Associations of Mutations in Six Pancreatic Cancer Genes With Personal and Family History of Cancers[a]

Gene	Personal History of Other cancers (n=513)[b]		No Personal History of Other cancers (n=2517)		Association		
	N	%	N	%	OR	95% CI	Adjusted p-value[c]
ATM	14	2.73	55	2.19	1.39	0.73-2.48	>.99
BRCA1	6	1.17	12	0.48	2.57	0.88-6.8	.35
BRCA2	14	2.73	45	1.79	1.88	0.98-3.39	.23
CDKN2A	2	0.39	8	0.32	1.29	0.19-5.29	>.99
MLH1	3	0.58	2	0.08	NT	NT	NT
TP53	3	0.58	3	0.12	4.76	0.85-26.64	.30
Total	42	8.18	125	4.97			
Gene	**Family History of Pancreatic cancer (n=343)**		**No Family History of Pancreatic cancer (n=2687)**		**Association**		
	N	**(%)**	**N**	**(%)**	**OR**	**95% CI**	**Adjusted p-value[b]**
ATM	11	3.21	58	2.30	1.49	0.73-2.76	>.99
BRCA1	2	0.58	16	0.64	0.98	0.15-3.46	>.99
BRCA2	7	2.04	52	2.07	1.04	0.43-2.17	>.99
CDKN2A	5	1.46	5	0.20	**7.91**	2.19-28.57	**.005**
MLH1	0	0	5	0.20	NT	NT	NT
TP53	2	0.58	4	0.16	3.98	0.55-20.5	.55
Total	27	7.87	140	5.56			
Gene	**Family History of Breast cancer (n=675)**		**No Family History of Breast cancer (n=2355)**		**Association**		
	N	**(%)**	**N**	**(%)**	**OR**	**95% CI**	**Adjusted p-value[b]**
ATM	18	2.67	51	2.03	1.22	0.69-2.07	>.99
BRCA1	4	0.59	14	0.56	1.00	0.28-2.79	>.99
BRCA2	22	3.26	37	1.47	**2.07**	1.19-3.5	**.04**
CDKN2A	2	0.3	8	0.32	0.87	0.13-3.47	>.99
MLH1	1	0.15	4	0.16	NT	NT	NT
TP53	4	0.59	2	0.08	7.12	1.38-51.47	.10
Total	51	7.56	116	4.61			
Gene	**Family History of Colorectal cancer (n=513)**		**No Family History of Colorectal cancer (n=2517)**		**Association**		
	N	**(%)**	**N**	**(%)**	**OR**	**95% CI**	**Adjusted p-value[b]**
ATM	10	1.95	59	2.34	0.82	0.39-1.54	>.99
BRCA1	2	0.39	16	0.64	0.61	0.1-2.15	>.99
BRCA2	10	1.95	49	1.95	0.99	0.47-1.89	>.99
CDKN2A	2	0.39	8	0.32	1.22	0.18-4.89	>.99
MLH1	3	0.58	2	0.08	NT	NT	NT
TP53	0	0	6	0.24	NT	NT	NT
Total	27	5.26	140	5.56			

Abbreviation: OR, odds ratio; NT, not testing because insufficient number of cases.

[a] Logistic regression adjusted for age at pancreatic cancer diagnosis. *MLH1* was not included due to limited number of patients with mutations. [b] Personal history of cancer before or after pancreatic cancer.

[c] Adjusted by Bonferroni correction of 5 tested genes.

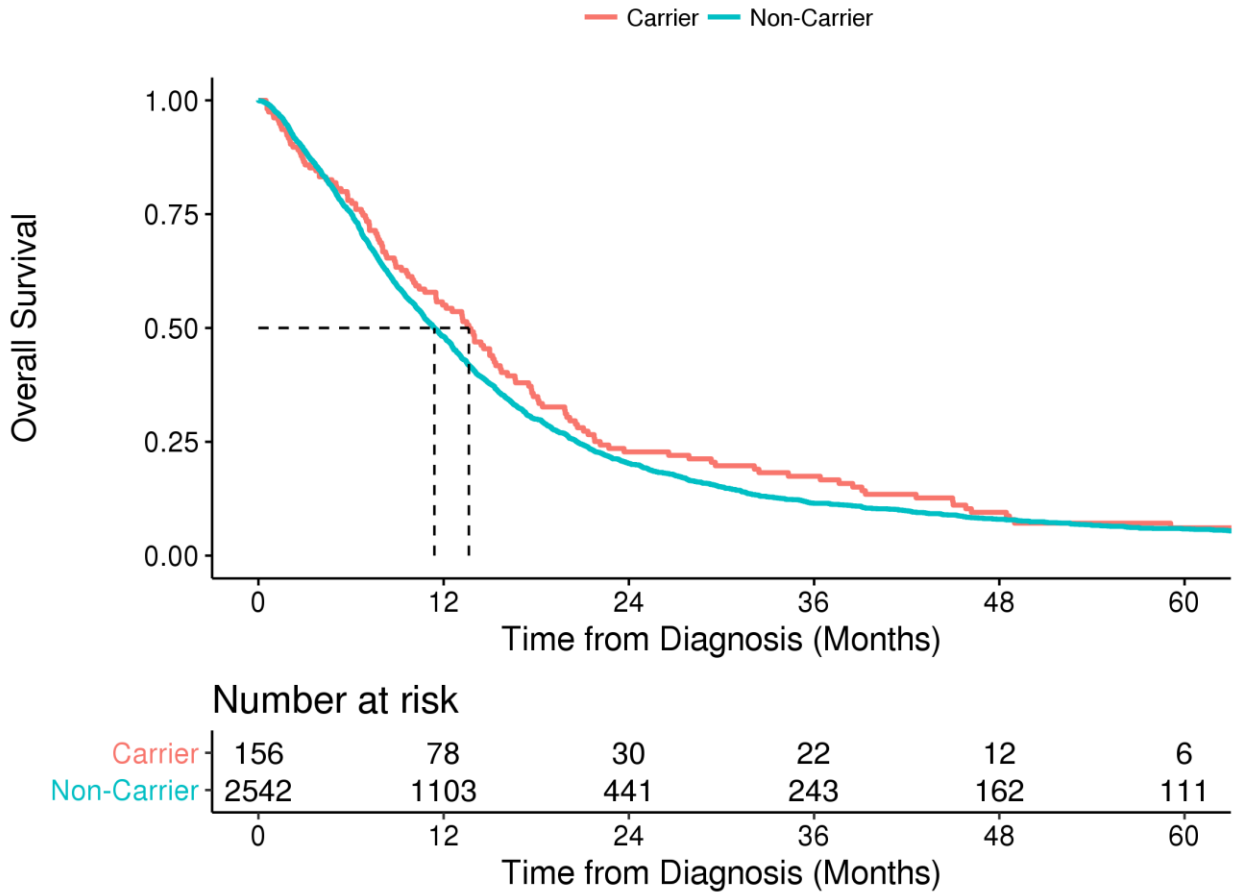
eTable 19. Associations Between Mutations in Each Panel Gene and Age of Diagnosis of Pancreatic Cancer Cases (N=3030)

Gene	KS[a] p-value	Mutation carriers (N)	Age at diagnosis of pancreatic cancer, years			
			Mean	Minimum	Maximum	Median
ATM	0.10	69	62.9	43	84	62
BARD1	0.83	4	64.0	52	71	66.5
BRCA1	0.58	18	65.1	50	90	62
BRCA2	0.01	59	60.5	39	83	60
BRIP1	0.13	5	60.2	51	65	63
CDH1	0.88	1	63.0	63	63	63
CDKN2A	0.38	10	64.3	56	73	65
CHEK2	0.70	33	65.5	34	84	65
FANCC	0.23	8	59.9	37	77	59.5
MLH1	0.73	5	62.0	44	74	70
MRE11A	0.73	2	62.0	58	66	62
MSH2	0.47	1	54.0	54	54	54
MSH6	0.27	7	65.3	59	69	66
NBN	0.50	4	71.5	61	82	71.5
NF1	0.13	4	72.3	69	78	71
PALB2	0.08	12	61.1	42	85	60
PMS2	0.91	2	61.5	51	72	61.5
RAD51C	0.93	3	67.0	60	71	70
TP53	0.57	6	67.7	52	82	71
No mutations[b]			63.3	37	90	63

Abbreviation: KS, Kolmogorov-Smirnov.
[a] KS test for mutated compared to non-mutated.
[b] Age data of cases without mutations in tested 21 genes.

eTable 20. Demographic and Clinical Characteristics of Patients With Pancreatic Cancer Included in the Survival Analysis	
Patient characteristics	(N=2698)
Age at Diagnosis	65.7 (10.8)
Gender	
Male	1552 (57.5%)
Female	1146 (42.5%)
Race/Ethnicity	
Non-Hispanic White	2589 (96.0%)
Black/African-American	37 (1.4%)
Hispanic	36 (1.3%)
Asian/Asian-American	10 (0.4%)
Other[a]	16 (0.6%)
Missing	10
Disease Stage	
Missing	4
Resectable	726 (26.9%)
Locally Advanced	997 (37.0%)
Metastatic	971 (36.0%)
Adult Body Mass Index (BMI)	28.57 (5.64)
Patient Reported Diabetes	691 (25.6%)
Ever Smokers	1502 (57.3%)
Pack Years in Ever Smokers	28.10 (24.33)

[a] Including Multiracial, American Indian/Alaskan Native, Native Hawaiian/Other Pacific Islander.



eFigure. Kaplan-Meier Analysis of Overall Survival for pancreatic Cancer Cases With Mutations (Carrier) in the 6 Genes Associated With Pancreatic Cancer and Cases Without Mutations (Non-Carrier)

The 6 genes are: *ATM, BRCA1, BRCA2, CDKN2A, MLH1,* and *TP53*. The hazard ratio for mutation status adjusting for age at diagnosis, sex, and stage was not statistically significant (overall survival, median OS=13.6 v 11.4 months: HR=0.86, 95% CI 0.72-1.02; p=.087). The median duration of follow-up for the carrier and non-carrier groups was 101.0 (range: 0.5-154.1) months and 103.0 (range: 0.0-220.2) months, respectively. The dashed lines represent the unadjusted median survival for carriers (13.6 months) and non-carriers (11.4 months).

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