

Genetic variance in cancer

Genetic variance refers to the phenotypic variation caused by differences in genotype and is one of the main causes of resistance to cancer drugs. Advanced understanding of the relationship between genetic variance and the resulting phenotype is crucial for the design of better treatment strategies. This resource gives an overview of the genetic variations most often associated with specific cancers and provides you with information about QIAGEN's tools for focused target enrichment for downstream NGS applications.

Overview

Genetic variance is the genetic change in a gene of interest compared to the wild-type gene. This variance may be caused by endogenous or exogenous factors. Some of the most frequent genetic variations arise from single nucleotide polymorphisms (SNPs), structural variations such as copy-number variation (CNV), and tandem repeats.

With its ability to deliver focused and accurate results, next-generation sequencing (NGS) is the method of choice for variant detection. Three different approaches to NGS — whole genome sequencing, whole transcriptome sequencing, and targeted DNA sequencing — enable identification of specific DNA changes according to project needs. Targeted DNA sequencing facilitates detection of rare mutations (known or unknown) from very small amounts of sample. Furthermore, by focusing on the genes that are most relevant, accurate data are obtained rapidly and with low costs.

GeneRead™ DNaseq Targeted Panels V2 are the simplest tools for analyzing the genetic variants of a focused panel of genes via NGS. Each panel consists of multiplex PCR primer sets to amplify exonic regions of a thoroughly researched panel of biologically and clinically relevant and disease-focused genes. GeneRead DNaseq Targeted Panels V2 can also be customized to include genes or other genomic regions tailored to your specific NGS project needs. Our high-quality primer design and targeted enrichment chemistry provide high design coverage, specificity, and uniformity, which are essential for detecting low-frequency variants in your precious samples.

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Genes frequently mutated in a variety of cancers

Some genes may play a role in just a few types of cancer whereas other genes are involved in a variety of different cancer types (Table 1).

Table 1. List of genes associated with multiple cancer types

Gene name	Associated cancer	Gene name	Associated cancer
ACVR1B	Breast, colon	KIT	Lung, myeloid, ovarian
AKT1	Breast, colon, lung, ovarian	KMT2D	Lung, prostate
APC	Colon, gastric, liver, lung, prostate	KRAS	Colon, liver, lung, myeloid, ovarian
ARID1A	Liver, ovarian	MAP2K4	Breast, colon
ATM	Breast, colon, liver, lung	MDM2	Breast, lung
BAI3	Gastric, lung	MET	Liver, lung
BAP1	Breast, lung	MLH1	Colon, lung, ovarian
BRAF	Colon, liver, lung, ovarian	MSH2	Colon, ovarian
BRCA1	Breast, ovarian	MUC16	Breast, lung
BRCA2	Breast, gastric, ovarian	MYC	Breast, gastric, lung, prostate
CDH1	Breast, gastric	NF1	Lung, myeloid, ovarian
CDK12	Ovarian, prostate	NOTCH1	Gastric, lung
CDKN2A	Breast, liver, lung, ovarian, prostate	NRAS	Colon, myeloid, ovarian
CSMD3	Liver, ovarian	PDGFRA	Lung, ovarian
CTNNB1	Colon, gastric, liver, ovarian	PIK3CA	Breast, colon, gastric, liver, lung, ovarian, prostate
DCC	Colon, gastric	PIK3R1	Breast, colon, ovarian
EGFR	Breast, lung, ovarian	PTEN	Breast, lung, ovarian, prostate
EP300	Breast, colon	RB1	Breast, lung, myeloid, ovarian, prostate
ERBB2	Breast, colon, gastric, liver, lung, ovarian	RET	Breast, gastric, lung
FBXW7	Colon, gastric, lung	RUNX1	Lung, myeloid
FGFR1	Breast, lung	SMAD4	Colon, lung
FGFR2	Breast, gastric, lung	STK11	Gastric, lung
KEAP1	Liver, lung	TP53	Breast, colon, gastric, liver, lung, myeloid, ovarian, prostate

Predesigned cancer-specific target enrichment panels for NGS

The Cancer Genome Atlas and the International Cancer Genome Consortium have created a comprehensive catalog of significantly mutated genes across all major cancer types. This has the major advantage that when looking for new or known rare genetic variance in these proven genes, a targeted approach can be followed instead of whole genome sequencing. This targeted approach has multiple benefits including use of only small sample amounts (10 ng) and less time and resources needed for sample processing, data storage, and analysis.

GeneRead DNAseq Targeted Panels V2 are wet-bench verified, ready-to-use panels for analyzing the genetic variants of a focused panel of genes via NGS. The panels employ overlapping primer sets across the exonic portions of a gene or group of genes to maximize target coverage. The following information provides lists of genes that are frequently mutated in specific cancers and which are included in GeneRead DNAseq Targeted Panels V2.

Breast cancer

Table 2 shows the 44 genes most commonly mutated in human breast cancer samples. The Human Breast Cancer GeneRead DNAseq Targeted Panel V2 enables targeted enrichment of the exonic regions of these 44 genes.

Table 2. Genes most commonly mutated in human breast cancer samples

Gene name						
ACVR1B	CDH1	EXOC2	ITCH	NCOR1	PTEN	WEE1
AKT1	CDKN2A	EXT2	KMT2C	NEK2	PTGFR	ZBED4
ATM	EGFR	FBXO32	MAP2K4	PBRM1	RB1	
BAP1	EP300	FGFR1	MAP3K1	PCGF2	RET	
BRCA1	ERBB2	FGFR2	MDM2	PIK3CA	SEPT9	
BRCA2	ERBB3	GATA3	MUC16	PIK3R1	TP53	
CBFB	ESR1	IRAK4	MYC	PPM1L	TRAF5	

Colorectal cancer

Table 3 shows the 38 genes most commonly mutated in human colorectal cancer samples. The Human Colorectal Cancer GeneRead DNAseq Targeted Panel V2 enables targeted enrichment of the exonic regions of these 38 genes.

Table 3. Genes most commonly mutated in human colorectal cancer samples

Gene name						
ACVR1B	BRAF	EP300	MAP2K4	MSH6	SLC9A9	TP53
AKT1	CASP8	ERBB2	MAP7	MYO1B	SMAD2	WBSCR17
APC	CDC27	FBXW7	MIER3	NRAS	SMAD4	
ATM	CTNNB1	FZD3	MLH1	PIK3CA	TCERG1	
ATP6V0D2	DCC	GPC6	MSH2	PIK3R1	TCF7L2	
BAX	DMD	KRAS	MSH3	PTPN12	TGFBR2	

Myeloid neoplasms

Table 4 shows the 50 genes most commonly mutated in human myeloid leukemia samples. The Human Myeloid Neoplasms GeneRead DNAseq Targeted Panel V2 enables targeted enrichment of the exonic regions of these 50 genes.

Table 4. Genes most commonly mutated in human myeloid leukemia samples

Gene name						
ABL1	DNMT3A	IDH2	KRAS	RAD21	SMC1A	WT1
ASXL1	EED	IKZF1	MPL	RB1	SMC3	ZRSR2
ATRX	ETV6	JAK1	NF1	RUNX1	STAG2	
BCOR	EZH2	JAK2	NPM1	SETBP1	SUZ12	
BCORL1	FLT3	JAK3	NRAS	SF1	TET2	
CBL	GATA1	KAT6A	PHF6	SF3A1	TP53	
CBLB	GNAS	KIT	PRPF40B	SF3B1	U2AF1	
DAXX	IDH1	KMT2A	PTPN11	SH2B3	U2AF2	

Liver cancer

Table 5 shows the 33 genes most commonly mutated in human liver cancer samples. The Human Liver Cancer GeneRead DNAseq Targeted Panel V2 enables targeted enrichment of the exonic regions of these 33 genes.

Table 5. Genes most commonly mutated in human liver cancer samples

Gene name						
ALB	ATM	CDKN2A	ERBB2	IGSF10	PIK3CA	WWP1
AMPH	AXIN1	CSMD3	ERRFI1	KEAP1	SAMD9L	ZIC3
APC	BAZ2B	CTNNB1	GXYLT1	KRAS	TP53	ZNF226
ARID1A	BRAF	DSE	HNF1A	MET	UBR3	
ARID2	CCDC178	ELMO1	IGF2R	OTOP1	USP25	

Lung cancer

Table 6 shows the 45 genes most commonly mutated in human lung cancer samples. The Human Lung Cancer GeneRead DNAseq Targeted Panel V2 enables targeted enrichment of the exonic regions of these 45 genes.

Table 6. Genes most commonly mutated in human lung cancer samples

Gene name						
AKT1	CDKN2A	FGFR2	LRP1B	NFE2L2	RARB	SOX2
ALK	EGFR	GRM8	MDM2	NOTCH1	RB1	STK11
APC	EPHA5	KDR	MET	PDGFRA	RET	TP53
ATM	ERBB2	KEAP1	MLH1	PIK3CA	ROS1	
BAI3	ERBB4	KIT	MUC16	PIK3CG	RUNX1T1	
BAP1	FBXW7	KMT2D	MYC	PKHD1	SMAD4	
BRAF	FGFR1	KRAS	NF1	PTEN	SMARCA4	

Ovarian cancer

Table 7 shows the 32 genes most commonly mutated in human ovarian cancer samples. The Human Ovarian Cancer GeneRead DNAseq Targeted Panel V2 enables targeted enrichment of the exonic regions of these 32 genes.

Table 7. Genes most commonly mutated in human ovarian cancer samples

Gene name						
AKT1	CBLC	CTNNB1	GABRA6	MLH1	PIK3CA	TP53
ARID1A	CCNE1	CUBN	KIT	MSH2	PIK3R1	USP16
BRAF	CDK12	EGFR	KRAS	NF1	PPP2R1A	
BRCA1	CDKN2A	ERBB2	KREMEN1	NRAS	PTEN	
BRCA2	CSMD3	FAT3	MAS1L	PDGFRA	RB1	

Prostate cancer

Table 8 shows the 32 genes most commonly mutated in human prostate cancer samples. The Human Prostate Cancer GeneRead DNAseq Targeted Panel V2 enables targeted enrichment of the exonic regions of these 32 genes.

Table 8. Genes most commonly mutated in human prostate cancer samples

Gene name						
AKAP9	CDKN2A	KMT2D	NKX3-1	PTEN	TBX20	ZNF473
APC	GLI1	MED12	NRCAM	RB1	TFG	ZNF595
AR	IKZF4	MYC	OR5L1	SCN11A	THSD7B	
CDK12	KDM4B	NCOA2	PDZRN3	SPOP	TP53	
CDKN1B	KLF6	NIPA2	PIK3CA	SYNE3	ZFH3	

Gastric cancer

Table 9 shows the 29 genes most commonly mutated in human gastric cancer samples. The Human Gastric Cancer GeneRead DNAseq Targeted Panel V2 enables targeted enrichment of the exonic regions of these 29 genes.

Table 9. Genes most commonly mutated in human gastric cancer samples

Gene name					
APC	CDH1	FGFR2	MET	RET	TP53
ATP4A	CTNNB1	GPR78	MYC	S1PR2	TRIO
BAI3	DCC	LPAR2	NOTCH1	SPEG	TRRAP
BRCA2	ERBB2	LRP1B	PIK3CA	SSTR1	WNK2
CCNE1	FBXW7	LRRK2	PRKDC	STK11	



Wet-bench verified primer sets for creating custom target enrichment panels

GeneRead DNAseq Custom Panels V2 enable analysis of the genetic variants of a customized panel of genes or genomic regions via NGS. Each panel consists of multiplex PCR primer sets, sufficient for 480 samples, to amplify genomic regions of interest tailored to your specific NGS needs. QIAGEN currently provides the largest number of wet-bench verified genes for target enrichment for custom panels (Table 10).

Table 10. List of all wet-bench verified genes for design of custom panels

Gene name						
ABCB11	ANTXR2	BCOR	CDKN1B	CYP27A1	EMD	FANCF
ABCC8	APC	BCORL1	CDKN2A	CYP27B1	EP300	FANCG
ABCC9	AR	BLM	CEP290	DAXX	EPCAM	FANCI
ABCD1	ARID1A	BMPR1A	CFTR	DBT	EPHA5	FANCL
ABL1	ARID2	BRAF	CHEK1	DCC	EPHB2	FANCM
ACADM	ARSA	BRCA1	CHEK2	DCX	ERBB2	FAS
ACADS	ASAH1	BRCA2	CHM	DDB2	ERBB3	FAT3
ACADVL	ASCC1	BRIP1	CIC	DDR2	ERBB4	FBXO11
ACTC1	ASL	BTD	CLN3	DES	ERCC2	FBXO32
ACTN2	ASPA	BTK	CLN5	DHCR7	ERCC3	FBXW7
ACVR1B	ASS1	BUB1B	CLN6	DICER1	ERCC4	FGD4
ADA	ASXL1	CALR3	CLN8	DIS3L2	ERCC5	FGFR1
ADAMTS13	ATM	CARD11	COL1A2	DKC1	ERCC6	FGFR2
ADAMTS2	ATP4A	CASP8	COL4A3	DLD	ERRFI1	FGFR3
AGA	ATP6V0D2	CAV3	COL4A4	DMD	ESCO2	FH
AGL	ATP7A	CBFB	COL7A1	DNAJB2	ESR1	FKTN
AGPS	ATP7B	CBL	COX15	DNMT3A	ETV6	FLCN
AHI1	ATP8B1	CBLB	CREBBP	DSC2	EXOC2	FLT3
AIP	ATR	CBLC	CRLF2	DSE	EXT1	FMR1
AKAP9	ATRX	CBS	CRTAP	DSG2	EXT2	FUBP1
AKT1	AXIN1	CCDC178	CRYAB	DSP	EYA4	FZD3
AKT2	AXIN2	CCNE1	CSF1R	DTNA	EZH2	G6PC
ALB	BAG3	CD79A	CSMD3	ECT2L	F11	GAA
ALDH3A2	BAI3	CD79B	CSRP3	EDA	F5	GABRA6
ALDOB	BAP1	CD96	CTNNB1	EDN3	FAH	GALNT12
ALK	BARD1	CDC27	CTNS	EDNRB	FAM46C	GALT
ALS2	BAX	CDC73	CTSK	EED	FANCA	GATA1
AMER1	BAZ2B	CDH1	CUBN	EGFR	FANCB	GATA2
AMPD1	BCKDHA	CDH23	CYLD	EGR2	FANCC	GATA3
AMPH	BCKDHB	CDK12	CYP11A1	EHBP1	FANCD2	GATAD1
ANTXR1	BCL6	CDK4	CYP21A2	ELMO1	FANCE	GBA

Gene name						
GCDH	JAK1	MDM2	NEK2	PLOD1	ROS1	SMPD1
GJB2	JAK2	MECP2	NEXN	PLP1	RPGRIP1L	SOX10
GLA	JAK3	MED12	NF1	PMP22	RS1	SOX2
GLB1	JUP	MEFV	NF2	PMS2	RSPO1	SPEG
GLI1	KAT6A	MEN1	NFE2L2	POLD1	RTKL	SPOP
GLI3	KCNQ1	MET	NFKBIA	POLE	RUNX1	SRC
GLMN	KDM4B	MFSD8	NIPA2	POLH	RUNX1T1	SSTR1
GNA11	KDM6A	MIER3	NKX3-1	POMGNT1	RYR2	STAG2
GNAQ	KDR	MITF	NOTCH1	POMT1	S1PR2	STAR
GNAS	KEAP1	MKS1	NOTCH2	POU1F1	SAMD9L	STK11
GNPTAB	KIF1B	MLH1	NPC1	POU6F2	SBDS	SUFU
GPC3	KIT	MLH3	NPC2	PPM1L	SCN11A	SUZ12
GPC6	KLF6	MMAB	NPHP1	PPP2R1A	SCN5A	SYNE3
GPR78	KLHDC8B	MPL	NPHP4	PPT1	SCNN1A	TAZ
GRIN2A	KMT2A	MPZ	NPM1	PRDM1	SCNN1B	TBX20
GRM8	KMT2C	MRE11A	NRAS	PRKAG2	SCNN1G	TCAP
GXYLT1	KMT2D	MSH2	NRCAM	PRKAR1A	SCO2	TCERG1
H3F3A	KRAS	MSH3	NTRK1	PRKDC	SDHA	TCF7L2
HADHA	KREMEN1	MSH6	NUP62	PROC	SDHAF2	TERT
HADHB	L1CAM	MSMB	OR5L1	PROP1	SDHB	TET2
HBB	LAMA2	MSR1	OTC	PRPF40B	SDHC	TFG
HESX1	LAMA4	MTAP	OTOP1	PRX	SDHD	TGFB3
HEXA	LAMP2	MTHFR	PAH	PSAP	SEPT9	TGFBR1
HEXB	LDB3	MTM1	PALB2	PSEN1	SETBP1	TGFBR2
HFE	LEPRE1	MTOR	PALLD	PSEN2	SETD2	THSD7B
HGSNAT	LIG4	MUC16	PAX5	PTCH1	SF1	TINF2
HIST1H3B	LMNA	MUT	PAX6	PTCH2	SF3A1	TMC6
HNF1A	LPAR2	MUTYH	PBRM1	PTEN	SF3B1	TMC8
HRAS	LRP1B	MYBPC3	PCDH15	PTGFR	SGCD	TMEM127
HSPH1	LRPPRC	MYC	PCGF2	PTPN11	SGSH	TMEM43
IDH1	LRRK2	MYD88	PDE11A	PTPN12	SH2B3	TMEM67
IDH2	LYST	MYH6	PDGFRA	RAC1	SLC25A4	TMPO
IGF2R	MAP2K1	MYH7	PDHA1	RAD21	SLC26A2	TNFAIP3
IGHMBP2	MAP2K2	MYL2	PDZRN3	RAD50	SLC37A4	TNFRSF14
IGSF10	MAP2K4	MYL3	PEX1	RAD51B	SLC7A8	TNNC1
IKBKAP	MAP3K1	MYLK2	PEX7	RAD51C	SLC9A9	TNNI3
IKZF1	MAP4K3	MYO1B	PHF6	RAD51D	SLX4	TNNT1
IKZF4	MAP7	MYO7A	PIK3CA	RARB	SMAD2	TNNT2
IL2RG	MAPK10	MYOZ2	PIK3CG	RB1	SMAD4	TP53
IL6ST	MAS1L	MYPN	PIK3R1	RBM20	SMARCA4	TPM1
IL7R	MAX	NBN	PKHD1	RECQL4	SMARCB1	TPP1
INVS	MC1R	NCOA2	PKP2	RET	SMC1A	TRAF5
IRAK4	MCCC2	NCOR1	PLEKHG5	RHBDF2	SMC3	TRIO
ITCH	MCOLN1	NDUFA13	PLN	RNASEL	SMO	TRPV4



Gene name					
TRRAP	U2AF1	USH1C	WAS	WWP1	ZIC3
TSC1	U2AF2	USH1G	WBSCR17	XPA	ZNF2
TSC2	UBA1	USP16	WEE1	XPC	ZNF226
TSHB	UBR3	USP25	WNK2	XRCC3	ZNF473
TSHR	UROD	VCL	WRN	ZBED4	ZNF595
TTN	UROS	VHL	WT1	ZFHX3	ZRSR2

Ordering Information

Product	Contents	Cat. no.
GeneRead DNaseq Targeted Panel V2	14 predesigned panels focused on specific diseases	181900
GeneRead DNaseq Mix-n-Match Panel V2	Design your own panel from 570 wet-bench verified assays	181905
GeneRead DNaseq Custom Panel V2	Build your own unique panel for any gene or genomic region	181902

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