

Invitae is a College of American Pathologists (CAP)-accredited and Clinical Laboratory Improvement Amendments (CLIA)-certified clinical diagnostic laboratory performing full-gene sequencing and deletion/duplication analysis using next-generation sequencing technology (NGS). Invitae's panel tests include:

- Full-gene sequencing: Covers clinically important regions of each gene, including coding exons, +/- 10 base pairs of adjacent intronic sequence (+/-20 bases for BRCA1 and BRCA2), and select noncoding variants.
- Deletion/duplication analysis: Detects most intragenic deletions and duplications at single exon resolution.
- Coverage: Our assay provides a Q30 quality-adjusted mean coverage depth of 350x (50x minimum, or supplemented with additional analysis).
- Confirmation: Variants classified as pathogenic or likely pathogenic are confirmed with orthogonal methods, except individual variants that have high quality scores and previously validated in at least ten unrelated samples.

For some genes, analysis may extend to the promoter region, include additional intronic variants, or be limited to targeted variants or exons. Details can be found at www.invitae.com.

A2ML1	ADAMTS2	ALG11	AP4S1	ATP6AP2	BCKDHA	C21orf59
AARS	ADAR	ALG12	AP5Z1	ATP6V0A2	BCKDHB	C5orf42
ABAT	ADSL	ALG13	APC	ATP7A	BCKDK	CA5A
ABCC9	AFF4	ALG14	APOB	ATP7B	BCL10	CACNA1A
ABCD1	AGA	ALG2	APP	ATRX	BCOR	CACNA1C
ABCD4	AGK	ALG3	AR	AUH	BFSP1	CACNA1H
ABCG5	AGL	ALG6	ARG1	AXIN2	BFSP2	CACNA1S
ABCG8	AGPS	ALG8	ARHGEF15	B2M	BICD2	CACNA2D1
ACAD8	AGRN	ALG9	ARHGEF9	B3GALNT2	BIN1	CACNA2D2
ACADM	AHCY	ALK	ARL13B	B3GALT6	BLM	CACNB2
ACADS	AHI1	ALMS1	ARL6	B3GAT3	BLNK	CACNB4
ACADSB	AICDA	ALS2	ARL6IP1	B3GLCT	BLOC1S6	CALM1
ACADVL	AIFM1	AMACR	ARMC4	B4GALNT1	BMP4	CALM2
ACAT1	AIPL1	AMN	ARSA	B4GALT1	BMPR1A	CALM3
ACD	AIRE	AMPD1	ARSB	B4GALT7	BMPR1B	CALR3
ACOX1	AK2	AMPD2	ARSE	B4GAT1	BMPR2	CAPN3
ACP5	AKAP9	AMT	ARSI	B9D1	BRAF	CARD11
ACSF3	AKT1	ANK2	ARX	B9D2	BRAT1	CARD14
ACTA1	AKT2	ANKRD1	ASAH1	BAG3	BRCA1	CARD9
ACTA2	AKT3	ANKRD11	ASL	BAP1	BRCA2	CARS2
ACTB	ALAD	ANKS6	ASPA	BARD1	BRIP1	CASK
ACTC1	ALDH18A1	ANO3	ASS1	BBS1	BSCL2	CASP10
ACTG1	ALDH1A3	ANO5	ATL1	BBS10	BTD	CASP8
ACTN2	ALDH4A1	ANOS1	ATL3	BBS12	BTK	CASQ2
ACVR2B	ALDH5A1	AP1S1	ATM	BBS2	BUB1B	CASR
ACVRL1	ALDH7A1	AP3B1	ATP13A2	BBS4	C12orf57	CAV1
ADA	ALDOA	AP4B1	ATP1A2	BBS5	C12orf65	CAV3
ADA2	ADLOB	AP4E1	ATP1A3	BBS7	C19orf12	CBL
ADAM17	ALG1	AP4M1	ATP2A1	BBS9	C1GALT1C1	CBS

Gene List



INVITAE

CC2D2A	CHKB	CP	CYP2U1	DOCK2	ETFA	FKRP
CCDC103	CHM	CPA6	CYP7B1	DOCK7	ETFB	FKTN
CCDC114	CHMP2B	CPOX	D2HGDH	DOCK8	ETFDH	FLCN
CCDC151	CHMP4B	CPS1	DAG1	DOK7	ETHE1	FLNA
CCDC39	CHRM2	CPT1A	DBH	DOLK	EVC	FLNC
CCDC40	CHRNA1	CPT1C	DBT	DPAGT1	EVC2	FLRT1
CCDC65	CHRNA2	CPT2	DCAF17	DPM1	EXOSC3	FOLR1
CCDC78	CHRNA4	CR2	DCDC2	DPM2	EXT1	FOXC1
CCM2	CHRN1	CRB1	DCLRE1B	DPM3	EXT2	FOXO3
CCNO	CHRN2	CREBBP	DCLRE1C	DRC1	EYA1	FOXP1
CCT5	CHRND	CRELD1	DCTN1	DRD2	EYA4	FOXH1
CD247	CHRNE	CRTAP	DDC	DSC2	EZH2	FOXN1
CD27	CHST14	CRX	DDHD1	DSE	F2	FOXP3
CD320	CHST3	CRYAA	DDHD2	DSG2	F5	FPR1
CD3D	CHST6	CRYAB	DDOST	DSP	F9	FRRS1L
CD3E	CHSY1	CRYBA1	DECR1	DST	FA2H	FTCD
CD3G	CIITA	CRYBA4	DEPDC5	DTNA	FADD	FTL
CD40LG	CIZ1	CRYBB1	DES	DYNC1H1	FAH	FUCA1
CD79A	CLCN1	CRYBB2	DHCR7	DYNC2H1	FAM126A	FUS
CD79B	CLCN4	CRYBB3	DHDDS	DYRK1A	FAM134B	FYCO1
CD8A	CLN2 (TPP1)	CRYGB	DHH	DYSF	FAM175A	G6PC
CDC73	CLN3	CRYGC	DHTKD1	DYX1C1	FANCA	G6PC3
CDH1	CLN5	CRYGD	DIAPH1	EDA	FANCB	G6PD
CDK4	CLN6	CRYGS	DICER1	EDAR	FANCC	GAA
CDKL5	CLN8	CSF2RA	DIS3L2	EDARADD	FANCD2	GABBR2
CDKN1B	CLPB	CSF3R	DKC1	EED	FANCE	GABRA1
CDKN1C	CNTN1	CSPP1	DLAT	EEF1A2	FANCF	GABRA6
CDKN2A	CNTN2	CSRP3	DLD	EFEMP2	FANCG	GABRB2
CDON	CNTNAP2	CSTB	DMD	EFHC1	FANCI	GABRB3
CEBPA	COASY	CTC1	DNAAF1	EGFR	FANCL	GABRD
CEBPE	COG1	CTDP1	DNAAF2	EGLN1	FANCM	GABRG2
CEP104	COG2	CTF1	DNAAF3	EGR2	FARS2	GAD1
CEP120	COG4	CTLA4	DNAAF5	EHMT1	FAS	GAL
CEP164	COG5	CTNNA1	DNAH1	ELAC2	FASLG	GALC
CEP290	COG6	CTNNA3	DNAH11	ELANE	FASN	GALE
CEP41	COG7	CTNS	DNAH5	ELN	FBN1	GALK1
CEP57	COG8	CTPS1	DNAH8	EMD	FBN2	GALNS
CEP83	COL12A1	CTRC	DNAI1	ENG	FBP1	GALNT12
CERS1	COL1A1	CTSA	DNAI2	ENO3	FBXO38	GALNT3
CFAP52	COL1A2	CTSC	DNAJB2	ENTPD1	FBXO7	GALT
CFAP53	COL3A1	CTSD	DNAJB6	EOGT	FERMT3	GAMT
CFL2	COL5A1	CTSF	DNAJC19	EP300	FGD4	GAN
CFTR	COL5A2	CTSK	DNAJC5	EPCAM	FGFR1	GARS
CHAT	COL6A1	CUBN	DNAJC6	EPG5	FGFR2	GAS8
CHCHD10	COL6A2	CUL4B	DNAL1	EPHA2	FGFR3	GATA1
CHCHD2	COL6A3	CXCR4	DNM1	EPM2A	FH	GATA2
CHD2	COLQ	CYBA	DNM2	ERCC4	FHL1	GATA4
CHD7	COPA	CYBB	DNMT1	ERF	FHL2	GATA6
CHEK2	COQ4	CYP1B1	DNMT3A	ERLIN1	FIG4	GATAD1
CHIT1	CORO1A	CYP27A1	DNMT3B	ERLIN2	FKBP14	GATM

Gene List



INVITAE

GBA2	GRIN2A	ICOS	ITPA	KPNA7	MAOA	MPC1
GBE1	GRIN2B	IDH2	IVD	KPTN	MAP2K1	MPDU1
GCDH	GRN	IDS	JAG1	KRAS	MAP2K2	MPI
GCH1	GSS	IDUA	JAGN1	KRIT1	MAP3K1	MPL
GCNT2	GUCY2D	IER3IP1	JAK3	L1CAM	MAP3K14	MPZ
GCSH	GUSB	IFIH1	JMJD1C	L2HGDH	MAPT	MRE11
GDAP1	GYG1	IFNGR1	JPH2	LAMA2	MARS	MSH2
GDF1	GYS1	IFNGR2	JUP	LAMA4	MASP1	MSH6
GDF2	GYS2	IFT122	KANSL1	LAMB2	MAT1A	MSX1
GDF3	HADH	IFT140	KAT6B	LAMP2	MAT2A	MSX2
GDF6	HADHA	IFT172	KBTBD13	LAMTOR2	MATR3	MTHFR
GFI1	HADHB	IFT80	KCNA1	LARGE1	MAX	MTM1
GFPT1	HAMP	IGHMBP2	KCNA2	LAS1L	MBD5	MTMR2
GIF	HAND1	IGLL1	KCNA5	LCA5	MC1R	MTO1
GJA1	HARS	IKBKAP	KCNB1	LCK	MCCC1	MTOR
GJA3	HAX1	IKBKB	KCNC1	LDB3	MCCC2	MTR
GJA5	HCFC1	IL10	KCND2	LDHA	MCEE	MTRR
GJA8	HCN1	IL10RA	KCND3	LDLR	MCIDAS	MUSK
GJB1	HCN4	IL10RB	KCNE1	LDLRAP1	MCOLN1	MUT
GJB6	HDAC8	IL12B	KCNE2	LEFTY2	MDM2	MUTYH
GJC2	HESX1	IL12RB1	KCNE3	LFNG	MECP2	MVK
GLA	HEXA	IL17F	KCNE5	LGI1	MED12	MYBPC3
GLB1	HEXB	IL17RA	KCNH2	LIAS	MED13L	MYD88
GLDC	HFE	IL17RC	KCNH5	LIG4	MED25	MYF6
GLI2	HFE2	IL1RN	KCNJ10	LIM2	MEF2C	MYH11
GLI3	HGD	IL21	KCNJ13	LIMS2	MEFV	MYH2
GLIS2	HGSNAT	IL21R	KCNJ2	LIPA	MEGF10	MYH6
GLRA1	HIBCH	IL2RA	KCNJ5	LITAF	MEGF8	MYH7
GLRB	HINT1	IL2RG	KCNJ8	LMBRD1	MEIS2	MYL2
GLUD1	HLCS	IL36RN	KCNK3	LMNA	MEN1	MYL3
GLUL	HMBS	IL7R	KCNMA1	LMNB2	MET	MYL4
GM2A	HMGCL	ILK	KCNQ1	LMOD3	MFN2	MYLK
GMPPA	HMGCS2	IMPDH1	KCNQ2	LPIN2	MFRP	MYLK2
GMPPB	HNRNPA2B1	INF2	KCNQ3	LRAT	MFSD8	MYOM1
GNAL	HNRNPDL	INPP5E	KCNT1	LRBA	MGAT2	MYOT
GNAO1	HNRNPU	INVS	KCTD17	LRP4	MIP	MYOZ2
GNB4	HOXB13	IQCB1	KCTD7	LRRC10	MITF	MYPN
GNE	HPCA	IQSEC2	KDM5C	LRRC6	MKKS	NADK2
GNMT	HPD	IRAK4	KDM6A	LRRK2	MKS1	NAGA
GNPAT	HPRT1	IRF6	KIAA0586	LRSAM1	MLH1	NAGLU
GNPTAB	HRAS	IRF7	KIAA2022	LTBP3	MLH3	NAGS
GNPTG	HSD17B10	IRF8	KIF1A	LYST	MLYCD	NBN
GNS	HSD17B4	ISG15	KIF1B	MAB21L2	MMAA	NCF2
GOSR2	HSF4	ISPD	KIF1C	MAF	MMAB	NCF4
GPC3	HSPB1	ITCH	KIF5A	MAG	MMACHC	NDRG1
GPD1L	HSPB3	ITGA2B	KIF7	MAGT1	MMADHC	NEB
GPHN	HSPB8	ITGA7	KIT	MALT1	MOCOS	NEBL
GREM1	HSPD1	ITGB2	KLHL40	MAN1B1	MOCS1	NECAP1
GRHL3	HYAL1	ITGB3	KLHL41	MAN1B2	MOGS	NEDD4L
GRIN1	IBA57	ITK	KMT2D	MANBA	MORC2	NEFL



NEK1	OCRL	PEX6	PNKP	PSPH	RINT1	SCN4A
NEK8	OFD1	PEX7	PNP	PSTPIP1	RIT1	SCN4B
NEU1	OGDH	PFKM	PNPLA2	PTCH1	RMRP	SCN5A
NEXN	OPA3	PFN1	PNPLA6	PTCH2	RNASEH2A	SCN8A
NF1	OPLAH	PGAM2	PNPO	PTEN	RNASEH2B	SCN9A
NF2	OPTN	PGAP1	POFUT1	PTPN11	RNASEH2C	SDCCAG8
NFAT5	ORAI1	PGM1	POGLUT1	PTPRC	RNF125	SDHA
NFIX	OTC	PGM3	POLD1	PTS	ROGDI	SDHAF2
NFKB2	OTX2	PHF6	POLE	PURA	RORC	SDHB
NFKBIA	OXCT1	PHGDH	POLG	PXDN	RPE65	SDHC
NFU1	P3H1	PHKA1	POMGNT1	PYGL	RPGR	SDHD
NGF	PACS1	PHKA2	POMGNT2	PYGM	RPGRIP1	SEC23A
NGLY1	PAH	PHKB	POMK	QARS	RPGRIP1L	SEC23B
NHEJ1	PALB2	PHKG2	POMT1	QDPR	RPL11	SEMA3E
NHLRC1	PALLD	PHOX2B	POMT2	RAB23	RPL26	SELENON
NHP2	PANK2	PHYH	POR	RAB27A	RPL35A	SERAC1
NHS	PAPSS2	PIGA	POT1	RAB3GAP2	RPL5	SERPINC1
NIPA1	PARK2	PIGG	PPM1K	RAB7A	RPN2	SERPINI1
NIPBL	PARK7	PIGL	PPOX	RAC2	RPS10	SETD2
NKX2-5	PARN	PIGM	PPT1	RAD21	RPS19	SETX
NKX2-6	PAX2	PIGN	PQBP1	RAD50	RPS24	SGCA
NLRC4	PAX6	PIGO	PRDM12	RAD51C	RPS26	SGCB
NLRP12	PAX9	PIGQ	PRDM16	RAD51D	RPS6KA3	SGCD
NLRP3	PC	PIGT	PRDM8	RAF1	RPS7	SGCE
NME8	PCBD1	PIGV	PREPL	RAG1	RRAS	SGCG
NMNAT1	PCCA	PIGW	PRF1	RAG2	RSPH1	SGSH
NOD2	PCCB	PIK3AP1	PRICKLE1	RANBP2	RSPH3	SH2D1A
NODAL	PCDH19	PIK3CA	PRICKLE2	RANGRF	RSPH4A	SH3BP2
NOP10	PCSK9	PIK3CD	PRIMA1	RAPSN	RSPH9	SH3TC2
NOTCH1	PDCD10	PIK3R1	PRKAG2	RARB	RTEL1	SHH
NOTCH2	PDE6D	PIK3R2	PRKAR1A	RASA1	RTN2	SHOC2
NPC1	PDGFRA	PINK1	PRKCD	RAX	RUNX1	SIGMAR1
NPC2	PDGFRB	PITX2	PRKDC	RB1	RYR1	SIK1
NPHP1	PDHA1	PITX3	PRKG1	RBCK1	RYR2	SIL1
NPHP3	PDHB	PKD2	PRKRA	RBFOX1	RYR3	SIX1
NPHP4	PDHX	PKHD1	PRNP	RBFOX3	SACS	SIX3
NPPA	PDK3	PKP2	PROC	RBM20	SALL1	SKI
NPR2	PDLIM3	PLA2G6	PRODH	RBM8A	SALL4	SLC12A5
NPRL3	PDP1	PLCB1	PROS1	RD3	SAMHD1	SLC13A5
NR0B1	PEX1	PLCG2	PRPH2	RDH12	SATB2	SLC16A2
NR2F2	PEX10	PLEC	PRPS1	RECQL4	SBF2	SLC17A5
NR5A1	PEX12	PLEKHG5	PRRT2	REEP1	SCARB2	SLC19A3
NRAS	PEX13	PLEKHM2	PRSS1	REEP2	SCN10A	SLC22A5
NRXN1	PEX14	PLN	PRSS56	RELN	SCN11A	SLC25A1
NSD1	PEX16	PLOD1	PRX	RET	SCN1A	SLC25A12
NSDHL	PEX19	PLP1	PSAP	RFT1	SCN1B	SLC25A13
NT5C2	PEX2	PMM2	PSAT1	RFX5	SCN2A	SLC25A15
NTRK1	PEX26	PMP22	PSEN1	RFXANK	SCN2B	SLC25A19
NUS1	PEX3	PMS2	PSEN2	RFXAP	SCN3A	SLC25A20
OAT	PEX5	PNKD	PSMB8	RHOH	SCN3B	SLC25A22

SLC25A46	SOS2	SURF1	TMEM216	TXNRD2	ZFR
SLC26A2	SOX2	SYN1	TMEM231	TYK2	ZFYVE26
SLC29A3	SOX9	SYNE1	TMEM237	UBA1	ZFYVE27
SLC2A1	SP110	SYNE2	TMEM43	UBE3A	ZIC2
SLC2A10	SPAG1	SYNGAP1	TMEM5	UBQLN2	ZIC3
SLC2A2	SPAST	SYNJ1	TMEM67	UMPS	ZMYND10
SLC33A1	SPATA5	SZT2	TMEM70	UNC13D	ZNF423
SLC35A1	SPATA7	TAP1	TMPO	UNC93B1	
SLC35A2	SPG11	TAP2	TNFRSF13B	UNG	
SLC35A3	SPG20	TAPBP	TNFRSF13C	UPF3B	
SLC35C1	SPG21	TARDBP	TNFRSF1A	USP8	
SLC35D1	SPG7	TAT	TNFRSF4	VAMP1	
SLC37A4	SPINK1	TAZ	TNFSF12	VAPB	
SLC39A13	SPINK5	TBC1D24	TNNC1	VAX1	
SLC3A1	SPR	TBK1	TNNI3	VCL	
SLC40A1	SPRED1	TBL1XR1	TNNT1	VCP	
SLC52A1	SPTAN1	TBX1	TNNT2	VHL	
SLC52A2	SPTLC1	TBX3	TNPO3	VIM	
SLC52A3	SPTLC2	TBX5	TOR1A	VMA21	
SLC5A1	SQSTM1	TCAP	TOR1AIP1	VPS13B	
SLC5A7	SRD5A2	TCF4	TP53	VPS35	
SLC6A1	SRD5A3	TCN1	TP63	VPS37A	
SLC6A3	SRPX2	TCN2	TPK1	VPS45	
SLC6A5	SRY	TCOF1	TPM1	VRK1	
SLC6A8	SSR4	TCTN1	TPM2	VSX2	
SLC6A9	ST3GAL3	TCTN2	TPM3	WAS	
SLC7A7	ST3GAL5	TCTN3	TPP2	WASHC5	
SLC7A9	STAC3	TDRD7	TRAF3	WDPCP	
SLC9A6	STAT1	TECPR2	TRAF3IP2	WDR19	
SLMAP	STAT2	TERC	TRAPPC11	WDR34	
SLX4	STAT3	TERT	TRDN	WDR35	
SMAD3	STAT5B	TFG	TREX1	WDR45	
SMAD4	STIM1	TFR2	TRIM2	WDR60	
SMAD6	STK11	TGFB2	TRIM32	WIPF1	
SMAD9	STK4	TGFB3	TRIP11	WNK1	
SMARCA4	STRA6	TGFBR1	TRNT1	WNT10A	
SMARCAL1	STRADA	TGFBR2	TRPM4	WRN	
SMARCB1	STT3A	TGIF1	TRPS1	WT1	
SMARCE1	STT3B	TH	TRPV4	WWOX	
SMC1A	STX1B	THAP1	TSC1	XIAP	
SMC3	STX11	TIA1	TSC2	XDH	
SMCHD1	STXBP1	TICAM1	TTC21B	XPNPEP3	
SMN1, SMN2	STXBP2	TINF2	TTC7A	XRCC2	
SMPD1	SUCLA2	TLR3	TTC8	XYLT1	
SNAP25	SUCLG1	TMC6	TTN	YARS	
SNCA	SUFU	TMC8	TTR	ZAP70	
SNTA1	SUMF1	TMEM127	TUBB4A	ZBTB24	
SNX27	SUN1	TMEM138	TULP1	ZDHHC9	
SOD1	SUN2	TMEM165	TUSC3	ZEB2	
SOS1	SUOX	TMEM173	TWIST1	ZFPM2	