

# G-Mendelioma Disease-Specific Panel

## KEY FEATURES

### 1. Comprehensive analysis of a broad range of diseases

Identifying diseases associated with:  
Acute lymphatic leukemia, Acute Myeloid Leukemia, Cardiac disease, Coagulation, Epilepsy, Hearing loss, Inborn errors of metabolism, Lymphoma, Lysosomal storage disease, Common hereditary cancer for a medical checkup, Neuromuscular disease, Parkinson's disease, Alzheimer's disease, Dementia, Dystonia, RASopathies, Retinitis pigmentosa, Short stature, Skin disease, and Somatic cancer

### 2. Collaboration with the leading CRO in the country

Developed 17 different panels for assessing genes of related diseases

## SPECIFICATION

Gene count*	Ranges from 14 to 320 genes
Covered regions	Whole CDS, hotspots
Target size	37.5-1,160 Kb
Mutation type	SNV, Indel, CNV
Sample type	Differs by somatic or germline panel
Platform	All sequencers from Illumina, Thermo Fisher, MGI, PacBio, and Oxford Nanopore
Bioinformatics pipeline	Primary, Secondary and Tertiary analysis result (FASTQ to VCF, VCF to Clinical report)

\* Gene Add-On Service: Genes can be added by customer's request

## PACKAGE COMPOSITION

Package name	Compositions		Package option	Options	
Target Enrichment	Target capture Probe	-	Pooling method	Single Reaction	Pre-capture Pooling
Standard	Target Enrichment reagents	Library prep Kit	Library Preparation kits	Standard Kit	EP-kit
All-In-One		Beads / Polymerase	Hybridization Enhancer	Included	Not included

## LIST OF PANELS FOR VARIOUS DISEASES

Panel Name	Gene List											
<b>Alzheimer-Dementia Panel</b> (101 genes, 244.8 Kb)  <b>Related Diseases:</b> Alzheimer's disease, Parkinson's disease, Dystonia	AARS	ABCA13	ABCA7	ABCB11	ADCY5	ALS2	ANG	ANO3	APP	ATP13A2	ATPIA3	ATP7B
	C19orf12	CACNA1B	CHCHD10	CHMP2B	CHRNA4	CIZ1	COG1	COL4A4	COL6A3	DAO	DCTN1	DNMT1
	EVC	FERMT1	FIG4	FREM2	FUS	GBA	GCH1	GNAL	GNAO1	GRM1	GRN	HNRNPA1
	HNRNPA2B1	HPCA	HPSE2	IL12RB2	KCTD17	KMT2B	L2HGDH	LAMA3	LRRK2	MAPT	MATR3	MECR
	NDUFV3	NEK1	NPHS2	OPTN	PANK2	PARK7	PDPI	PINK1	PLA2G6	PNKD	PRKN	PRKRA
	PRNP	PRRT2	PSEN1	PSEN2	RELN	SERPIND1	SETX	SGCE	SIGMAR1	SLC12A6	SLC19A3	SLC2A1
	SLC30A10	SLC6A3	SNCA	SOD1	SORL1	SOX6	SPG11	SQSTM1	SRY	SUMF1	TAF1	TAF15
	TARDBP	TBK1	TDRD7	TH	THAP1	TIMM8A	TOR1A	TREM2	TUBA4A	TUBB4A	UBQLN2	VAC14
	VAPB	VCAN	VCP	VPSI3A	WNK1							
	<b>Bleeding Disorder-Coagulopathy Panel</b> (147 genes, 326.3 Kb)  <b>Related Diseases:</b> Bleeding Disorder, Coagulation	AARS	ABCA1	ABCA13	ABCB11	ACTN1	ANKRD26	ANO6	AP3B1	ARHGAP35	BLOC1S3	BLOC1S6
BRCA2		BRIP1	CD36	CDAN1	COG1	COL4A4	CPNE1	CYCS	DDX41	DKC1	DNMT1	DTNBPI
ELANE		ERCC4	ETV6	EVC	F10	F11	F13A1	F13B	F2	F5	F7	F8
F9		FANCA	FANCB	FANCC	FANCD2	FANCE	FANCF	FANCG	FANCI	FANCL	FANCM	FERMT1
FERMT2		FGA	FGB	FGG	FIL1	FREM2	FYB1	GATA1	GATA2	GF11	GF1B	GPIBA
GPIBB		GP6	GP9	GRM1	HAX1	HOXA11	HPS1	HPS3	HPS4	HPS5	HPS6	HPSE2
IFNG		IL12RB2	ITGA2B	ITGB3	KIAA1244	L2HGDH	LAMA3	LMAN1	LYST	MASTL	MCFD2	MLPH
MPL		MYH9	MYO5A	NBEAL2	NBN	NDUFV3	NHP2	NOPI1	NPHS2	P2RY12	PALB2	PCNXL2
PDP1		PLA2G4A	PLAU	PRF1	PRKACG	PRUNE2	RAB27A	RAD51C	RASGRP2	RBM8A	RIPK3	RPL11
RPL35A		RPL5	RPS10	RPS19	RPS24	RPS26	RPS7	RUNX1	SBD5	SEC23B	SERPIND1	SERPINE1
SERPINF2		SLC12A6	SLFN14	SLX4	SOX6	SP4	SRC	SRP72	SRY	STIM1	SUMF1	TBXA2R
TBXAS1		TDRD7	TERC	TERT	TINF2	UBE2T	USP6NL	VCAN	VIPAS39	VPS33B	VWF	WAS
WIPF1	WNK1	XRCC2										
<b>Cardiovascular Panel</b> (206 genes, 681.3 Kb)  <b>Related Diseases:</b> Cardiac diseases	AARS	ABCA13	ABCB11	ABCC9	ABCG5	ABCG8	ACTA1	ACTA2	ACTC1	ACTN2	AKAP9	ALMS1
	ANK2	ANKRD1	APOA4	APOA5	APOB	APOC2	APOE	ARHGAP35	BAG3	BRAF	CACNA1C	CACNA2D1
	CACNB2	CALM1	CALR3	CASQ2	CAV3	CBL	CBS	CETP	COG1	COL3A1	COL4A4	COL5A1
	COL5A2	COX15	CPNE1	CREB3L3	CRELD1	CRYAB	CSRP3	CTF1	DES	DMD	DNAJC19	DNMT1
	DOLK	DPP6	DSC2	DSG2	DSP	DTNA	EFEMP2	ELN	EMD	EVC	EYA4	FBN1
	FBN2	FERMT1	FHL1	FHL2	FKRP	FKTN	FREM2	FXN	GAA	GATAD1	GCKR	GJA5
	GLA	GPDIL	GPIHBP1	GRM1	HADHA	HCN4	HFE	HPSE2	HRAS	HSPB8	IL12RB2	ILK
	JAG1	JPH2	JUP	KCNA5	KCND3	KCNE1	KCNE2	KCNE3	KCNH2	KCNJ2	KCNJ5	KCNJ8
	KCNQ1	KIAA1244	KLF10	KRAS	L2HGDH	LAMA2	LAMA3	LAMA4	LAMP2	LDB3	LDLR	LDLRAP1
	LMF1	LMNA	LPL	LTPB2	MAP2K1	MAP2K2	MI61	MURC	MYBPC3	MYH11	MYH6	MYH7
	MYL2	MYL3	MYLK	MYLK2	MYO6	MYO22	MYPN	NDUFV3	NEXN	NKX2-5	NODAL	NOTCH1
	NPHS2	NPPA	NRAS	PCNXL2	PCSK9	PDLIM3	PDP1	PKP2	PLN	PRDM16	PRKAG2	PRKARIA
PRUNE2	PTPN11	RAF1	RANGRF	RBM20	RIPK3	RYR1	RYR2	SALL4	SCN1B	SCN2B	SCN3B	
SCN4B	SCN5A	SCO2	SDHA	SEPN1	SERPIND1	SGCB	SGCD	SGCG	SHOC2	SLC12A6	SLC25A4	
SLC2A10	SMAD3	SMAD4	SNTA1	SOS1	SOX6	SP4	SREBF2	SUMF1	TAZ	TBX20	TBX3	
TBX5	TCAP	TDRD7	TGFB2	TGFB3	TGFBRI	TGFBRI2	TMEM43	TMPO	TNNC1	TNNI3	TNNT2	
TPM1	TRDN	TRIM63	TRPM4	TTN	TTR	TXNRD2	USP6NL	VCAN	VCL	WNK1	ZBTB17	
ZHX3	ZIC3											
<b>Common Hereditary Cancer Panel</b> (93 genes, 218 Kb)  <b>Related Diseases:</b> Medical checkup	AARS	ABCA13	ABCB11	APC	ARHGAP35	ATM	ATRX	BARD1	BMPRIA	BRAF	BRCA1	BRCA2
	BRIP1	CDHI	CDKN2A	CHEK2	COG1	COL4A4	CPNE1	DNMT1	EGLN1	EGLN2	EPAS1	EPCAM
	EVC	FERMT1	FGFR1	FH	FREM2	GRM1	H3F3A	HPSE2	HRAS	IDH2	IL12RB2	KIAA1244
	KIF1B	KMT2D	L2HGDH	LAMA3	MAX	MDH2	MEN1	MERTK	MET	MLH1	MRE11	MSH2
	MSH6	MUTYH	NBN	NDUFV3	NF1	NF2	NPHS2	PALB2	PCNXL2	PDP1	PMS2	POLD1
	POLE	PRSS1	PRUNE2	PTEN	RAD50	RAD51C	RAD51D	RBI	RET	RIPK3	SDHA	SDHAF2
	SDHB	SDHC	SDHD	SERPIND1	SLC12A6	SMAD4	SOX6	SP4	SPINK1	STK11	SUMF1	TDRD7
	TMEM127	TP53	TSC1	TSC2	USP6NL	VCAN	VHL	WNK1	WT1			
	<b>Epilepsy Panel</b> (150 genes, 401.6 Kb)  <b>Related Diseases:</b> Epilepsy	AARS	ABCA13	ABCB11	ADGRV1	ADSL	ALDH7A1	ALG13	ARHGAP35	ARHGEF15	ARHGEF9	ARX
ATPIA2		ATP6AP2	CACNA1A	CASK	CDKL5	CHD2	CHRNA2	CHRNA4	CHRNA7	CHRN2	CLCN4	CLN3
CLN5		CLN6	CLN8	CNTNAP2	COG1	COL4A4	CPNE1	CSTB	CTSD	DCX	DEPDC5	DLG3
DNAJC5		DNMI	DNMT1	DOCK7	DYRK1A	EEF1A2	EPM2A	EVC	FERMT1	FOLR1	FOXG1	FREM2
GABRA1		GABRA2	GABRB3	GABRG2	GAMT	GATM	GNAO1	GOSR2	GRIN1	GRIN2A	GRIN2B	GRM1
HCN1		HDAC4	HNRNPU	HPSE2	IL12RB2	IQSEC2	KANSL1	KCNA2	KCNB1	KCNH5	KCNJ10	KCNMA1
KCNQ2		KCNQ3	KCNT1	KCTD7	KIAA1244	L2HGDH	LAMA3	LGII	MAGI2	MBD5	MECP2	MEF2C
MFSDB		NDUFV3	NECAP1	NHLRC1	NPHS2	NR2F1	NRXN1	PCDH19	PCNXL2	PDP1	PIGA	PIGO
PIGQ		PIGV	PLCB1	PNKP	PNPO	POLG	PPT1	PRICKLE1	PRICKLE2	PRRT2	PRUNE2	QARS
RELN		RIPK3	SCARB2	SCN1A	SCN1B	SCN2A	SCN8A	SCN9A	SERPIND1	SLC12A6	SLC13A5	SLC25A22
SLC2A1		SLC35A2	SLC6A8	SLC9A6	SMS	SOX6	SP4	SPTANI	SRPX2	SRY	ST3GAL3	STXBPI
SUMF1		SYN1	SYNGAP1	SYNJ1	SZT2	TBC1D24	TCF4	TDRD7	TPPI	TSCI	TSC2	UBE3A
USP6NL	VCAN	WDR45	WNK1	WWOX	ZEB2							

# LIST OF PANELS FOR VARIOUS DISEASES

Panel Name	Gene List											
<b>Hearing Loss-Deafness Panel</b> (62 genes, 130.3 Kb)	AARS	ABCA13	ABCB11	ARHGAP35	CDH23	CLRN1	COCH	COG1	COL11A1	COL2A1	COL4A4	CPNE1
	DIAPH1	DNMT1	EDNRB	EVC	EYA1	FERMT1	FREM2	GJB2	GJB6	GRM1	HPSE2	IL12RB2
	KCNE1	KCNQ1	KCNQ4	KIAA1244	L2HGDH	LAMA3	MTF	MYO15A	MYO7A	NDUFV3	NPHS2	OTOF
	PAX3	PCNXL2	PDP1	POU3F4	PRUNE2	RIPK3	SERPIND1	SIX5	SLC12A6	SLC26A4	SNAI2	SOX10
	SOX6	SP4	SUMF1	TDRD7	TECTA	TMC1	TMIE	TMPRSS3	USH1C	USH2A	USP6NL	VCAN
<b>Related Diseases:</b> Hearing loss, Deafness	WFS1	WNK1										
<b>Lymphoid Leukemia Panel</b> (85 genes, 109.2 Kb)	AARS	ABCA13	ABCB11	ABL1	AMELX	AMELY	ARHGAP35	BRAF	BTG1	CDKN2A	COG1	COL4A4
	CPNE1	CREBBP	CRLF2	DNM2	DNMT1	DNMT3A	EP300	ETV6	EVC	EZH2	FBXW7	FERMT1
	FLT3	FREM2	GATA3	GRM1	HPSE2	IDH1	IDH2	IKZF1	IL12RB2	IL7R	JAK1	JAK2
	JAK3	KDM6A	KIAA1244	KMT2A	KMT2D	KRAS	L2HGDH	LAMA3	LEF1	LMO1	MAPK1	NDUFV3
	NF1	NOTCH1	NPHS2	NRAS	NSD2	NT5C2	NUDT15	PAX5	PCNXL2	PDP1	PHF6	PRUNE2
<b>Related Diseases:</b> Acute lymphatic leukemia	PTEN	PTPN11	RBI	RIPK3	RUNX1	SERPIND1	SETD2	SH2B3	SLC12A6	SOX6	SP4	SRY
	STAG2	STAT3	STAT5B	SUMF1	TBL1XR1	TCF3	TDRD7	TP53	TPMT	USP6NL	VCAN	WNK1
	WT1											
<b>Lymphoma Panel</b> (83 genes, 118.3 Kb)	AARS	ABCA13	ABCB11	ALK	ARHGAP35	ATM	B2M	BCL6	BIRC3	BRAF	BTM	CARD11
	CD79A	CD79B	COG1	COL4A4	CPNE1	CREBBP	CXCR4	DNMT1	EGR2	EP300	EVC	EZH2
	FAS	FAT4	FBXO11	FERMT1	FREM2	GRM1	HPSE2	ID3	IDH2	IKBKB	IKZF1	IL12RB2
	JAK3	KIAA1244	KLF2	L2HGDH	LAMA3	MYC	MYD88	NDUFV3	NFKBIE	NOTCH1	NOTCH2	NPHS2
	PCNXL2	PDP1	PLCG1	PLCG2	POT1	PRDM1	PRUNE2	RHOA	RIPK3	RPS15	RRAGC	SERPIND1
<b>Related Diseases:</b> Lymphoma	SF3B1	SLC12A6	SOC3	SOX6	SP4	SRY	STAT3	STAT5B	SUMF1	TBL1XR1	TCF3	TDRD7
	TE12	TNFAIP3	TNFRSF14	TP53	TP63	TRAF3	UBR5	USP6NL	VCAN	WNK1	XPO1	
<b>Lysosomal Storage Diseases Panel</b> (117 genes, 195.9 Kb)	AARS	ABCA13	ABCB11	ABCD1	ACOX1	AGA	AGL	ALDOA	ALDOB	ARHGAP35	ARSA	ARSB
	ATP13A2	ATP7A	ATP7B	CLN3	CLN5	CLN6	CLN8	COG1	COL4A4	CPNE1	CTNS	CTSA
	CTSD	CTSF	DNAJC5	DNMT1	EVC	FERMT1	FREM2	FUCA1	G6PC	GAA	GALC	GALE
	GALK1	GALK2	GALNS	GALT	GBA	GBE1	GJB2	GLA	GLB1	GNPTAB	GNPTG	GNS
	GRM1	GRN	GUSB	GYS1	GYS2	HEXA	HEXB	HGSNAT	HPRT1	HPSE2	HYAL1	IDS
<b>Related Diseases:</b> Lysosomal storage disease	IDUA	IL12RB2	KCTD7	KIAA1244	L2HGDH	LAMA3	LDHA	LIPA	MAN2B1	MANBA	MCOLN1	MFSDB8
	NAGA	NAGLU	NDUFV3	NEU1	NPC1	NPC2	NPHS2	PCNXL2	PDP1	PEX1	PEX10	PEX12
	PEX13	PEX14	PEX16	PEX19	PEX2	PEX26	PEX3	PEX5	PEX6	PFKM	PHKA2	PHKB
	PHKG2	PPT1	PRUNE2	PYGL	PYGM	RIPK3	SERPIND1	SGSH	SLC12A6	SLC17A5	SLC2A2	SLC37A4
	SMPD1	SOX6	SP4	SUMF1	TDRD7	TPPI	USP6NL	VCAN	WNK1			
<b>Metabolic Disorders Panel</b> (103 genes, 138.3 Kb)	AARS	ABCA13	ABCB11	ABCD1	ACAD8	ACADM	ACADS	ACADSB	ACADVL	ACAT1	AHCY	ARG1
	ARHGAP35	ASL	ASS1	AUH	BCKDHA	BCKDHB	BTD	CBS	COG1	COL4A4	CPNE1	CPS1
	CPT1A	CPT2	DBT	DECRI	DHCR7	DLD	DNMT1	ETFA	ETFB	ETFDH	EVC	FAH
	FERMT1	FREM2	GALE	GALK1	GALT	GAMT	GATM	GCDH	GCHI	GNMT	GRM1	HADH
	HADHA	HADHB	HLC3	HMGCL	HPD	HPSE2	HSD17B10	IL12RB2	IVD	KIAA1244	L2HGDH	LAMA3
<b>Related Diseases:</b> Inborn errors of metabolism	LMBRD1	MATIA	MCCC1	MCCC2	MLYCD	MMAA	MMAB	MMACHC	MMADHC	MMUT	MTHFR	MTR
	MTRR	NDUFV3	NPHS2	OPA3	OTC	PAH	PCBD1	PCCA	PCCB	PCNXL2	PDP1	PRUNE2
	PTS	QDPR	RIPK3	SERPIND1	SLC12A6	SLC22A5	SLC25A13	SLC25A20	SLC6A8	SOX6	SP4	SUMF1
	TAT	TAZ	TCN2	TDRD7	USP6NL	VCAN	WNK1					
<b>Myeloid Leukemia Panel</b> (83 genes, 94.8 Kb)	AARS	ABCA13	ABCB11	AMELX	AMELY	ANKRD26	ARHGAP35	ASXL1	ATRX	BCOR	BCORL1	BRAF
	CALR	CBL	CBLB	CEBPA	COG1	COL4A4	CPNE1	CSF3R	DDX41	DNMT1	DNMT3A	ETV6
	EVC	EZH2	FERMT1	FLT3	FREM2	GATA1	GATA2	GRM1	HPSE2	HRAS	IDH1	IDH2
	IL12RB2	JAK2	JAK3	KDM6A	KIAA1244	KIT	KRAS	L2HGDH	LAMA3	MPL	NDUFV3	NOTCH1
	NPHS2	NPM1	NRAS	PCNXL2	PDGFRA	PDP1	PHF6	PPM1D	PRUNE2	PTPN11	RAD21	RIPK3
<b>Related Diseases:</b> Acute myeloid leukemia	RUNX1	SERPIND1	SETBP1	SF3B1	SLC12A6	SMC1A	SMC3	SOX6	SP4	SRSF2	STAG1	STAG2
	STAT3	SUMF1	TDRD7	TET2	TP53	U2AF1	USP6NL	VCAN	WNK1	WT1	ZRSR2	
<b>Neuromuscular Panel</b> (320 genes, 1,160 Kb)	AARS	ABCA13	ABCB11	ABCB7	ABCD1	ABHD12	ACAD9	ACADL	ACADM	ACO2	ACTA1	ADCK3
	AFG3L2	AGL	AIFM1	ALDH3A2	AMPD1	ANO10	ANO5	AP4B1	AP4E1	AP4M1	AP4S1	AP5Z1
	APTIX	ARHGAP35	ARSA	ATCAY	ATL1	ATM	ATP2A1	ATP7A	ATP7B	ATP8A2	BAG3	BEAN1
	BINI	BSCL2	C10orf2	C12orf65	C19orf12	CACNA1A	CACNA1S	CACNB4	CAPN3	CASK	CAV3	CCDC78
	CCDC88C	CFL2	CHAT	CHRNA1	CHRN1	CHRNA1	CHRNA1	CHRNA1	CHRNA1	CHRNA1	CHRNA1	CHRNA1
<b>Related Diseases:</b> Neuromuscular disease	COG1	COL4A4	COL6A1	COL6A2	COL6A3	COLQ	CPNE1	CPT1B	CPT2	CRYAB	CTDPI	CWF19L1
	CYP27A1	CYP2U1	CYP7B1	DAG1	DCTN1	DDHD1	DDHD2	DES	DMD	DNAJB2	DNAJB6	DNM2
	DNMT1	DOX7	DYNCH1H	DYSF	EEF2	EGR2	ELOVL4	ELOVL5	EMD	ERLIN2	ETFA	ETFB
	EVC	FA2H	FAM134B	FERMT1	FGD4	FIG14	FHL1	FIG4	FKRP	FKTN	FLNC	FLVCR1
	FREM2	FRMD7	FUS	FXN	GAA	GAD1	GALC	GAN	GARS	GBA2	GDAP1	GJB1
	GJC2	GLA	GLE1	GNB4	GNE	GOSR2	GPRI43	GRID2	GRM1	GYSI	HADHA	HADHB

Panel Name	Gene List														
<b>Neuromuscular Panel</b> (320 genes, 1,2 Mb)	HINT1	HOXD10	HPSE2	HSPB1	HSPB8	HSPD1	HSPG2	IGHMBP2	IKBKAP	IL12RB2	ISPD	ITGA7			
	ITPR1	JPH3	KBTBD13	KCNA1	KCNC3	KCND3	KCNE3	KCNJ10	KCNJ18	KIAA0196	KIAA1244	KIF1A			
	KIF1B	KIF1C	KIF5A	KLHL40	KLHL41	LICAM	L2HGDH	LAMA1	LAMA2	LAMA3	LARGE	LDB3			
	LITAF	LMNA	LPIN1	LRSAM1	MARS	MARS2	MATR3	MED25	MFN2	MPZ	MRE11A	MTM1			
	MTMR14	MTMR2	MTPAP	MTPP	MUSK	MYF6	MYH2	MYH7	MYOT	NDRG1	NDUFV3	NEB			
	NEFL	NGF	NIPAI	NOP56	NPHS2	NTNR1	OPA1	OPA3	OPHN1	PABPN1	PANK2	PCNXL2			
	PDK3	PDP1	PDYN	PEX7	PFKM	PGAM2	PHKA1	PHYH	PLEC	PLEKHG5	PLP1	PMM2			
	PMP22	PNKP	PNPLA6	POLG	POLG2	POMGNT1	POMT1	POMT2	PRKCG	PRPS1	PRUNE2	PRX			
	PTFIA	PTRF	PYGM	RAB7A	RAPSN	REEPI	RIPK3	RNF216	RRM2B	RTN2	RUBCN	RYR1			
	RYR2	SACS	SBF2	SCN4A	SCN9A	SEPN1	SERPIND1	SETX	SGCA	SGCB	SGCD	SGCE			
	SGCG	SH3TC2	SIL1	SLC12A6	SLC16A2	SLCIA3	SLC33A1	SLC39A4	SLC52A2	SLC9A1	SLC9A6	SMN1			
	SNX14	SOD1	SOX6	SP4	SPAST	SPG11	SPG20	SPG21	SPG7	SPTBN2	SPTLC1	SPTLC2			
	STAC3	STUB1	SUCLA2	SUMF1	SYNE1	SYNE2	SYT14	TBP	TCAP	TDP1	TDRD7	TECP2			
	TGM6	TK2	TMEM240	TNNI2	TNNT1	TPM2	TPM3	TPPI	TRIM32	TRPV4	TTBK2	TTN			
	TPA	TTR	TUBB4A	TYP	USP6NL	VAMP1	VCAN	VCP	VLDLR	VPS13A	VPS37A	VRK1			
	WFS1	WNK1	WWOX	XK	YARS	ZFYVE26	ZFYVE27	ZNF592							
	<b>Related Diseases:</b> Neuromuscular disease														
	<b>RASopathy Panel</b> (14 genes, 37.5 Kb)	BRAF	CBL	HRAS	KRAS	MAP2K1	MAP2K2	NF1	NRAS	PTPN11	RAF1	RIT1	SHOC2		
		SOS1	SPRED1												
	<b>Related Diseases:</b> RASopathies														
<b>Retinitis Pigmentosa Panel</b> (111 genes, 325.3 Kb)	ABCA4	ABHD12	ADAM9	ADGRA3	AGBL5	AIPL1	ARHGEF18	ARL2BP	ARL3	ARL6	BBS1	BBS2			
	BEST1	C2orf71	C8orf37	CA4	CABP4	CACNA1F	CACNA2D4	CDHR1	CERKL	CLRN1	CNGA1	CNGB1			
	CNGB3	CNNM4	CRB1	CRX	CWC27	CYP4V2	DHDDS	DHX38	ELOVL4	EMC1	EYS	FAM161A			
	FLVCR1	FSCN2	GNAT2	GUCA1A	GUCA1B	GUCY2D	HGSNAT	HK1	IDH3B	IFT140	IFT172	IMPDH1			
	IMPG2	KCNV2	KIAA1549	KIZ	KLHL7	LRAT	MAK	MERTK	MVK	NEK2	NEUROD1	NR2E3			
	NRL	OFD1	PDE6A	PDE6B	PDE6C	PDE6G	PDE6H	PITPNM3	POMGNT1	PRCD	PRKCG	PROM1			
	PRPF3	PRPF31	PRPF4	PRPF6	PRPF8	PRPH2	RAB28	RAX2	RBP3	RDH12	RDH5	REEP6			
	RGR	RGS9	RGS9BP	RHO	RIMS1	RLBP1	ROM1	RPI	RP2	RP9	RPE65	RPGR			
	RPGRIP1	SAG	SEMA4A	SLC7A14	SNRNP200	SPATA7	SPP2	TOPORS	TRNT1	TTC8	TULP1	UNC119			
	USH2A	ZNF408	ZNF513												
	<b>Related Diseases:</b> Retinitis pigmentosa														
	<b>Short Stature Panel</b> (192 genes, 616.1 Kb)	AARS	ABCA13	ABCB11	ACTA2	ADAMTS10	ADAMTS2	ADAMTS4	AGPS	ALPL	ARHGAP35	ARSE	ATP6V0A2		
		ATP7A	ATRX	B3GALT6	B4GALT7	BGN	BLM	BRAF	CBL	CBS	CDC6	CDT1	CHST14		
COG1		COL10A1	COL11A1	COL1A1	COL1A2	COL2A1	COL3A1	COL4A4	COL5A1	COL5A2	COL9A1	COL9A2			
COL9A3		COMP	CPNE1	CREBBP	CRTAP	CTSK	CUL7	DHCR7	DLL3	DNMT1	DYNC2H1	DYRK1A			
EBP		EFEMP2	ELN	EP300	ERCC6	ERCC8	EVC	EVC2	EXT1	EXT2	FBLN5	FBN1			
FBN2		FERMT1	FGD1	FGF23	FGFR1	FGFR2	FGFR3	FKBP10	FLNA	FLNB	FOXO3	FREM2			
GHI		GHR	GHRHR	GLI2	GLI3	GNAS	GNPT	GRM1	HESX1	HPSE2	HRAS	HSPG2			
IFITM5		IFT80	IGF1	IGF1R	IL12RB2	INPPL1	KCNJ2	KCNJ8	KDM6A	KIAA1244	KMT2D	KRAS			
L2HGDH		LAMA3	LBR	LHX3	LIFR	LOX	LTBP2	LZTR1	LZTR1	MAP2K2	MAT2A	MATN3			
MED12		MFAP5	MYH11	MYLK	NBAS	NBN	NDUFV3	NEK1	NF1	NIPBL	NPHS2	NRAS			
NSDHL		OBSL1	ORC1	ORC4	ORC6	P3H1	PCNT	PCNXL2	PDP1	PEX7	PHEX	PLOD1			
POR		POU1F1	PP1B	PPP1CB	PRKG1	PROPI	PRUNE2	PTPN11	PYCR1	RAF1	RIN2	RIPK3			
RIT1		RMRP	ROR2	RPS6KA3	RUNX2	SBD5	SERPIND1	SERPINH1	SHOC2	SKI	SLC12A6	SLC26A2			
SLC2A10		SLC34A3	SLC35D1	SLC39A13	SMAD3	SMARCAL1	SMCIA	SMC3	SMS	SOS1	SOS2	SOX3			
SOX6		SOX9	SP4	SPRED1	SRCAP	SUMF1	TDRD7	TGFB2	TGFB2	TGFB2	TGFB2	TGFB2			
THR1		TRIM37	TRIP11	TRPS1	TRPV4	TTC21B	USP6NL	VCAN	WDR19	WDR35	WNK1	WRN			
<b>Related Diseases:</b> Short stature															
<b>Skin Disorder Panel</b> (152 genes, 545.7 Kb)	ABCA12	ABC8	ABCC6	ABHD5	ADAMTS2	ADAR	ALAD	ALAS2	ALDH3A2	ALOX12B	ALOXE3	APIS1			
	ATM	ATP2A2	ATP2C1	ATP6V0A2	BLM	CARD14	CDH3	CDSN	CLDN1	COL17A1	COL1A1	COL1A2			
	COL3A1	COL5A1	COL5A2	COL7A1	CPOX	CTCI	CTSC	CYP4F22	DDB2	DKC1	DOCK8	DSG1			
	DSG4	DSP	DST	EBP	ECM1	EDA	EDAR	EDARADD	EFEMP2	ELN	ERCC2	ERCC3			
	ERCC4	ERCC5	EXPH5	FANCA	FANCC	FANCG	FECH	FERMT1	FLCN	FLG	GJB2	GJB3			
	GJB4	GJB6	GNAS	GORAB	GPRI43	GSN	GTF2H5	HFE	HMB5	HR	IL36RN	ITGA3			
	ITGA6	ITGB4	JUP	KIT	KRT1	KRT10	KRT14	KRT16	KRT17	KRT2	KRT5	KRT6A			
	KRT6B	KRT6C	KRT81	KRT83	KRT86	KRT9	LAMA3	LAMB3	LAMC2	LIPH	LIPN	LOR			
	LPAR6	LYST	MBTPS2	NF1	NF2	NHP2	NIPAL4	NOPI0	NSDHL	OCA2	PKP1	PLEC			
	PLOD1	PNPLA1	POFUT1	POGLUT1	POLH	POMP	PPOX	PRKAR1A	PTCH1	PTCH2					