

ÖRNEK BİLGİSİ

Hasta Adı Soyadı:	<input type="checkbox"/> Kan	<input type="checkbox"/> CVS
D.Tarihi/Cinsiyet:	<input type="checkbox"/> Amniyon Sıvısı	<input type="checkbox"/> Düşük
T.C. Kimlik No:	<input type="checkbox"/> Kemik İliği	<input type="checkbox"/> Diğer.....
İletişim Bilgisi :	NOT:	
Göndren Merkez:		
Gönderen Doktor:		
Endikasyon:		

MOLEKÜLER GENETİK

PANEL	GEN
<input type="checkbox"/> BRCA 1, 2 Panel	SNV, InDel, CNV : BRCA1 & BRCA2.
<input type="checkbox"/> Somatic Mutation Cancer Panel	Rearrangement : ALK, RET, ROS1 CNV : ERBB2, MET SNV-InDel : APC, BRAF, EGFR, ERBB2, KRAS, MET, NRAS, PIK3CA, SMAD4, TP53.
<input type="checkbox"/> Hereditary Cancer Panel	SNV, InDel, CNV : APC, ATM, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, EPCAM, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, PRSS1, PTEN, RAD50, RAD51C, RAD51D, SLX4, SMAD4, STK11, TP53, VHL (SNV, InDel, CNV).
<input type="checkbox"/> Mitochondrial DNA Depletion Syndrome Panel	DGUOK, MPV17, POLG, TRMU , SURF1, NDUFV1, TMEM70, BCS1L, NDUFS1, NDUFS3, SCO2, NF1, NF2.
<input type="checkbox"/> Tuberous Sclerosis Syndrome Panel	TSC1 , TSC2.
<input type="checkbox"/> Polycystic Kidney Diseases Panel	PKD1 , PKD2 , PKHD1.
<input type="checkbox"/> FGFR-Related Craniosynostosis Panel	FGFR1 , FGFR2 , FGFR3.
<input type="checkbox"/> Glycogen Storage Disease Panel	AGL, ALDOA, ENO3, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, SLC2A2, SLC37A4.
<input type="checkbox"/> Retinitis Pigmentosa Disease Panel	ABCA4, ARL6, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C2orf71, C8ORF37, CERKL, CNGA1, CNGB1, CRB1, CRX, DHDDS, EYS, FAM161A, FLVCR1, GNPTG, FSCN2, IDH3B, IMPG2, LRAT, MAK, MERTK, NR2E3, NRL, OAT, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PRCD, PROM1, RBP3, RBP4, RDH12, RGR, RHO, RLBP1, RP1, RP2, RP9, RPE65, RPGR, SAG, SEMA4A, SPATA7, TTC8, TULP1, USH2A, ZNF513, ZNF469, ZNF644.

PANEL	GEN
<input type="checkbox"/> Alport Syndromes Panel	COL4A3 , COL4A4 , COL4A5.
<input type="checkbox"/> Ehlers-Danlos Syndromes Panel	COL5A1 , COL5A2 , COL3A1 , PLOD1, COL1A1, COL1A2.
<input type="checkbox"/> Osteogenesis Imperfecta Panel	COL1A1 , COL1A2.
<input type="checkbox"/> Fatty Acid Oxidation Disorders Panel	ACAT1, ACAD9, ACADL, ACADM, ACADS, ACADVL, CPT1A, CPT1B, CPT2, ETFA , ETFB, ETFDH, GLUD1, HADH, HADHA, HADHB, HMGCL, HMGCS2, HSD17B10, LPIN1, SLC22A5, SLC25A20, SCOT, TAZ
<input type="checkbox"/> 46,XY Disorders of Sex Development/Complete Gonadal Dysgenesis Disorder Panel	AMH, AMHR2, ANKR1C, AR, ARX, ATRX, B3GALTL, CYB5A, CYN2H1, CYP11A1, CYP17A1, DHCR7, DHH, GATA4, HCCS, HSD17B3, LHCGR, MAMLD1, MAP3K1, NR5A1, OPHN1, SOX9, SRD5A2, SRY, WT1, ZFPM2.
<input type="checkbox"/> 46,XX Disorders of Sex Development/Complete Gonadal Dysgenesis Panel	B3GLCT, CYP11B1, CYP19A1, CYP21A2, HCCS, PSMC3IP, RSPO1, WNT4, WT1
<input type="checkbox"/> Cardiomyopathy Panel	AKAP9, ANK2, CACNA1C, CACNB2, CAV3, GPD1L, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, PRKAG2.
<input type="checkbox"/> Congenital Dyserythropoietic Anemia Panel	C15orf41, CDAN1, GATA1, KLF1, SEC23B, F8, F9, GP1BA, VWF.
<input type="checkbox"/> X-Linked Mental Retardation Panel	AP1S2, ARX, ATRX, BRWD3, CASK, CUL4B, DCX, DLG3, FGD1, FMR1, IL1RAPL1, IQSEC2, KDM5C, MED12, MID1, OPHN1, PHF8, PQBP1, RPS6KA3(RSK2), SLC9A6, UBE2A, UPF3B, ZDHHC9.
<input type="checkbox"/> Early Infantile Epileptic Encephalopathy Panel	AARS, ALG13, ARHGEF9, ARV1, ARX, CACNA1A, CDKL5, DNM1, DOCK7, EEF1A2, FRRS1L, GABRA1, GABRB3, GNAO1, GRIN2B, GUF1, HCN1, ITPA, KCNA2, KCNB1, KCNQ2, KCNQ3, KCNT1, NECAP1, PCDH19, PIGA, PLCB1, PNKP, SCN1A, SCN2A, SCN8A, SCN9A, SLC2A1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC35A2, SPTAN1, ST3GAL3, STXBP1, SZT2, TBC1D24, WWOX, ALDH7A1, PNPO, MAOA, DBH, TH, FOLR1, GCH1, DCX.
<input type="checkbox"/> Thyroid Panel	GLIS3, TSHB, THRA, PAX8, NKX2-5, NKX2-1, FOXE1, TSHR, SLC5A5, SLC26A4, TG, TPO, DUOX2, DUOXA2, IYD, SLC26A7, DUOX1, ZNF607, SLC6A4, DIO1, DIO2, DIO3, TTR, GNAS, TRH, ALB, POR, TRHR, PHEX, SLC16A2, SERPINA7, IGSF1.
<input type="checkbox"/> Congenital Hearing Loss Panel	COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL9A1, COL9A2, DFNA5, DFNB31, DFNB59, GJB2, GJB3, GJB6, KCNQ1, KCNQ4, MYH14, MYH9, MYO15A, MYO1A, MYO3A, MYO6, MYO7A, SERPINB6, SLC17A8, SLC26A4, SLC26A5, SLC4A11.
<input type="checkbox"/> Congenital Myopathy Panel	ACTA1, CFL2, KBTBD13, NEB, CCDC78, CNTN1, DNM2, LMOD3, MTM1, MYF6, MYH7, PTPLA, RYR1, SEPN1, TTN.

PANEL	GEN
<input type="checkbox"/> Congenital Muscular Dystrophy Panel	CHKB, COL6A1, COL6A2, COL6A3, DAG1, DPM2, DPM3, FKRP, FKTN, GAA, POMGNT2(GTDC3), POMK, ISPD, ITGA7, LAMA2, LARGE, LMNA, POMGNT1, POMT1, POMT2, RYR1, SEPN1, SYNE1, TMEM5.
<input type="checkbox"/> Limb Girdle Muscular Dystrophy Panel	MYOT, LMNA, CAV3, DES, DNAJB6, FLNC, CAPN3, DYSF, SGCG, SGCA, SGCB, SGCD, TCAP, TRIM32, FRKP, TTN, POMT1, ANO5, FKTN, POMT2, POMGNT1, DAG1, PLEC, GAA, GMPPB, HNRNPDL, TNPO3, LIMS2, POMK, TRAPPC11, ISPD.
<input type="checkbox"/> Hereditary Cancer PLUS Panel	APC, ATM, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, EPCAM, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, PRSS1, PTEN, RAD50, RAD51C, RAD51D, SLX4, SMAD4, STK11, TP53, VHL, NF1, MEN1, CTNNB1, RBL.
<input type="checkbox"/> Joubert Syndrome Panel	AHI1, C5orf42, CC2D2A, CEP290, CSPP1, INPP5E, KIAA0586, MKS1, NPHP1, RRGRI1L, TCTN2, TMEM67, TMEM216, ARL13B, B9D1, B9D2, C2CD3, CEP41, CEP104, CEP120, IFT172, KIAA0556, KIF7, OFD1, PDE6D, POC1B, TCTN1, TCTN3, TMEM107, TMEM138, TMEM231, TMEM237, TTC21B, ZNF423.
<input type="checkbox"/> Hypertrophic cardiomyopathy (HCM) Panel	ACTC1, ACTN2, CSRP3, CAV3, DTNA, GLA, JUP, LAMP2, MAP2K2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, NEXN, PLN, PRKAG2, PTPN11, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR.
<input type="checkbox"/> Dilated cardiomyopathy (DCM) NGS Panel Panel	ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CRYAB, CSRP3, DES, DMD, DSC2, DSG2, DSP, EYA4, FLNC, ILK, LAMA4, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYPN, NEBL, NEXN, PDLIM3, PKP2, PLN, PSEN1, PSEN2, RBM20, SCN5A, SGCD, TAZ, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, VCL.
<input type="checkbox"/> Arrhythmia Panel	NOTCH1, AKAP9, ANK2, CACNA1C, CACNB2, CAV3, CTNNA3, DES, DSC2, DSG2, DSP, GPD1L, JUP, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNQ1, LMNA, PKP2, PLN, PRKAG2, RYR2, SCN1B, SCN3B, SCN4B, SCN5A, SNTA1, TGFB3, TMEM43, TTN.
<input type="checkbox"/> Familial Hypercholesterolemia Panel	LDLR, APOB, PCSK9, CYP27A1, APOE, ABCG5, ABCG8, LDLRAP1, USF1, LPL.
<input type="checkbox"/> Aortopathy Panel	ACTA2, CBS, COL3A1, FBN1, FBN2, MYH11, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB3, TGFB3, TGFB3.
<input type="checkbox"/> Pulmonary hypertension Panel	BMPR2, ACVRL1, EIF2AK4, KCNK3, CAV1, SMAD9, BMPR1B, ENG.
<input type="checkbox"/> Phenylketonuria Panel	PAH, GCH1, PCBD1, PTS, QDPR, DNAJC12, FAH.
<input type="checkbox"/> Charge, Kabuki Meckel Syndrome Panel	CHD7, SEMA3E, EYA1, FLNB, IRF6, KDM6A, MLL2(KMT2D), SIX5.
<input type="checkbox"/> Methemoglobinemia Panel	CYB5R3, CYB5A.
<input type="checkbox"/> Combined Pituitary Hormone Deficiency Panel	PROP1, POU1F1, HESX1, LHX3, LHX4, GH1, GHRHR, GLI2, OTX2, SOX2, SOX3.
<input type="checkbox"/> Otosomal Dominant Tubulointerstitial Panel	UMOD, HNF1B, REN, MUC1.

PANEL

GEN

Tubuler Disorder Panel

SLC12A1, KCNJ1, BSND, CLCNKA, CLCNKB, SLC12A3, MAGED2, CTNS, CLCN5, CLCN1, OCRL1, ATP6V0A4, ATP6V1B1, SLC4A1, CA2, CLDN19, CLDN16, CNNM2, EGF, FXD2, RET, WNT11, GDNF, WT1, EYA1, PAX2, CD2AP, ANLN, CRB2, PAX2, TRPC6, INF2, MYO1E, ACTN4, APOL1, COL4A3, COL4A4, COL4A5, MYH9, SLC12A3, SCNN1B, SCNN1G, CaSR, GNA11, AP2S1.

Demantia Syndrome Panel

APOE, APP, C9ORF72, CHMP2B, CSF1R, DNMT1, FUS, GRN, MAPT, PRNP, PSEN1, PSEN2, SNCA, SNCB, SORL1, SQSTM1, TARDBP, TREM2, UBQLN2, A2M, AAAS, ACE, ATP13A2, ATP1A3, DCTN1, EIF4G1, FBXO7, GBA, GCH1, HTRA2, LRRK2, MPO, NOTCH3, PARK7, PINK1, PLA2G6, POLG, PRKN, PRKRA, SLC6A3, TAF1, TH, TYROBP, UCHL1, VPS35.

Thyroid Plus Panel

GLIS3, TSHB, THRA, THRB, PAX8, NKX2-5, NKX2-1, FOXE1, TSHR, SLC5A5, SLC26A4, TG, TPO, DUOX2, DUOX2, IYD, SLC26A7, DUOX1, ZNF607, SLC6A4, DIO1, DIO2, DIO3, TTR, GNAS, TRH, ALB, POR, TRHR, PHEX, SLC16A2, SERPINA7, IGSF1.

Obesity-Mental Reterdation Panel

ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, CUL4B, DYRK1B, GNAS, IFT27, LEP, LEPR, LZTFL1, MAGEL2, MC4R, MKKS, MKS1, NR0B2, NTRK2, PCSK1, PHF6, POMC, SDCCAG8, SIM1, TRIM32, TTC8, UCP3, VPS13B, WDPCP, SNRPN.

Rasopathy Panel

A2ML1, ACTB, ACTG1, BRAF, CBL, CDC42, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1.

Hyperlipidemia Panel

ABCA1, ABCG5, ABCG8, APOB, APOC3, APOE, LDLR, LDLRAP1, LPL, PCSK9, ALMS1, APOA1, APOA5, APOC2, CREB3L3, GPIHBP1, LIPA, LMF1, PCSK9.

Rubinstein-Taybi Panel

CREBBP, EP300.

Osteopetrosis Panel

TCIRG1, CLCN7, TNFS11, TNFS11A, CA2, OSTM1, LRP5, PLEKHM1, SNX10.

Mucopolysaccharidoses Syndrome Panel

ARSB, GALNS, GUSB, IDS, IDUA, GNS, HGSNAT, LDB3, MYOT, NAGLU, SGSH.

Autoinflammatory Diseases Panel

NLRP3, MVK, TNFRSF1A, NLRP12, PSTPIP1, NOD2, ELANE, IL1RN, LPIN2, ADA2, TNFRSF11A, MEFV.

Nephronophthisis Panel

CEP164, CEP290, CEP83, DCDC2, FAN1, GLIS2, IFT172, INVS, IQCB1, MRE11, NEK8, NPHP1, NPHP3, NPHP4, RFGRIPL1, SDCCAG8, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423, CFH, CFI.

Nephrotic Syndrome Panel

NPHS1, NPHS2, LMX1B, COL4A3, COL4A4, COL4A5, TRPC6.

PANEL	GEN
<input type="checkbox"/> Atypical Hemolytic Uremic Syndrome and Membranoproliferative Glomerulonephritis Panel	CFH ,CFI.
<input type="checkbox"/> Peroxisomal Panel	ABCD1, ABCD3, ACOX1, AGPS, DNM1L, HSD17B4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH.
<input type="checkbox"/> Congenital Disorders of Glycosylation Panel	ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ATP6V0A2, B3GLCT, B4GALT1, COG1, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM3, GNE, MAGT1, MGAT2, MOGS, MPDU1, MPI, PMM2, RFT1, RPN2, SEC23B, SLC35A1, SLC35C1, SRD5A3, TUSC3.
<input type="checkbox"/> Congenital Myasthenic Syndrome Panel	AGRN, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COLQ, DOK7, GFPT1, LAMB2, MUSK, PLEC, RAPSN, SCN4A.
<input type="checkbox"/> Primary Autosomal Recessive Microcephalies and Seckel Syndrome Panel	MCPH1, WDR62(MCPH2), CDK5RAP2(MCPH3), KNL1(MCPH4), ASPM(MCPH5), CENPJ(MCPH6/SCKL4), STIL(MCPH7), CEP135(MCPH8), CEP152(MCPH9).
<input type="checkbox"/> Primary Immunodeficiency Panel	RAG1, RAG2, ATM, NBN, TBX1, STAT3, PGM3, BTK, TACI (TNFRSF13B), FOXP3, CTLA4, LRBA, F12, SERPING1 (C1NH), GF11, JAGN1, USB1, CSF3R, WAS, G6PC3, VPS45A (VPS45), ELANE, HAX1, CYBA, CYBB, NCF1, NCF2, XK, NOD2, NCF4, FAS, FASLG, CASP10, PRKCD, IRAK4, MYD88, IRAK1, DOCK8, IL10, IL10Ra, IL10Rb, STXBP2, STX11, PRF1, UNC13D, IL12B1 (IL12B), ISG15, IKBKG, IFNGR2, IFNGR1, STAT1, IL6, RECQL3 (BLM).
<input type="checkbox"/> Chronic Malabsorption Panel	MTP (MTTP),ApoB, PCSK9, SAR1B, UBR1, SBDS, PNLIP, MYO5B, EPCAM, PCSK1, NEUROG3
<input type="checkbox"/> Hypomotility Panel	MNGIE (TYMP), ACTG2, FLNA,NEUROG3, EDNRB, EDN3, SOX10
<input type="checkbox"/> Chronic Diarrhea Panel	SLC26A3, SPINT2, SLC5A1, TTC37, SKIV2L, FOXP3, SLC39A4, SLC10A2, STAT3, CD25 (IL2RA), STAT5B, STAT1, LCT, GLUT5 (SLC2A5), SLC7A7, PRSS7 (TMPRSS15), NEUROG3, PCSK1, SI
<input type="checkbox"/> Inflammatory Intestinal Panel	IL10RA, IL10, IL10RB, CRFB4 (IL10RB), EPCAM, FOXP3, SKIV2L, TTC37, TTC7A, MEFV
<input type="checkbox"/> Chronic Pancreatitis Panel	SPINK1, PRSS1, CTRC, CFTR, SBDS, PNLIP, UBR1
<input type="checkbox"/> Cholestase Panel	JAG1, NOTCH2, SMPD1, NPC1, NPC2, ATP8B1, ABCB11, ABCB4, AMACR, SLCO1B3, SLCO1B1, ABCC2
<input type="checkbox"/> SPG Panel	ATL1, SPAST, REEP1, CYP7B1, SPG11, ZFYVE26, SPG21, L1CAM, PLP1, NIPA1, SPG7, KIAA0196 (WASHC5), ALDH18A1, KIF5A, BSCL2, SPG20 (SPART), DDHD1, KIF1A, FA2H, GJC2, AP5Z1, DDHD2
<input type="checkbox"/> MODY Neonatal Diabetes Panel	ABCC8, AKT2, BLK, CEL, CISD2, CP, EIF2AK3, FOXP3, GATA6, GCK, GLIS3, GLUD1, HADH, HNF1A, HNF1B, HNF4A, IER3IP1, INS, INSR, KCNJ11, KLF11, NEUROD1, NEUROG3, PAX4, PDX1, PTF1A, RFX6, SLC2A2, WFS1, ZFP57, G6PC2, NKX2-2, SLC16A1

PANEL

GEN

Common Hereditary Cancer Panel

APC, ATM, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MEN1, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, NF1, NF2, PALB2, PMS2, POLD1, POLE, PRSS1, PTEN, RAD50, RAD51C, RAD51D, RB1, RET, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SPINK1, STK11, TP53, TSC1, TSC2, VHL, WT1

Metabolic Disorders Panel

ABCD1, ACAD8, ACADM, ACADS, ACADSB, ACADVL, ACAT1, AHCY, ARG1, ASL, ASS1, AUH, BCKDHA, BCKDHB, BTB, CBS, CPS1, CPT1A, CPT2, DBT, DECR1, DHCR7, DLD, ETFA, ETFB, ETFDH, FAH, GALE, GALK1, GALT, GAMT, GATM, GCDH, GCH1, GNMT, HADH, HADHA, HADHB, HLCS, HMGCL, HPD, HSD17B10, IVD, LMBRD1, MAT1A, MCCC1, MCCC2, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MTHFR, MTR, MTRR, OPA3, OTC, PAH, PCBD1, PCCA, PCCB, PTS, QDPR, SLC22A5, SLC25A13, SLC25A20, SLC6A8, TAT, TAZ, TCN2

Lysosomal Storage Diseases Panel

ABCD1, ACOX1, AGA, AGL, ALDOA, ALDOB, ARSA, ARSB, ATP13A2, ATP7A, ATP7B, CLN3, CLN5, CLN6, CLN8, CTNS, CTSA, CTSD, CTSF, DNAJC5, FUCA1, G6PC, GAA, GALT, GALE, GALK1, GALK2, GALNS, GALT, GBA, GBE1, GJB2, GLA, GLB1, GNPTAB, GNPTG, GNS, GRN, GUSB, GYS1, GYS2, HEXA, HEXB, HGSNAT, HPRT1, HYAL1, IDS, IDUA, KCTD7, LDHA, LIPA, MAN2B1, MANBA, MCOLN1, MFSD8, NAGA, NAGLU, NEU1, NPC1, NPC2, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PFKM, PHKA2, PHKB, PHKG2, PPT1, PYGL, PYGM, SGSH, SLC17A5, SLC2A2, SLC37A4, SMPD1, SUMF1, TPP1

Retinitis Pigmentosa Panel

ABCA4, ABHD12, ADGRA3, AGL5, AIPL1, ARHGEF18, ARL2BP, ARL3, ARL6, BBS1, BBS2, BEST1, C2orf71, C8orf37, CA4, CDHR1, CERKL, CLRN1, CNGA1, CNGB1, CRB1, CRX, CWC27, CYP4V2, DHDDS, DHX38, EMC1, EYS, FAM161A, FLVCR1, FSCN2, GUCA1B, HGSNAT, HK1, IDH3B, IFT140, IFT172, IMPDH1, IMPG2, KIAA1549, KIZ, KLHL7, LRAT, MAK, MERTK, MVK, NEK2, NEUROD1, NR2E3, NRL, OFD1, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RBP3, RDH12, REEP6, RGR, RHO, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RPGR, SAG, SEMA4A, SLC7A14, SNRNP200, SPATA7, SPP2, TOPORS, TRNT1, TTC8, TULP1, USH2A, ZNF408, ZNF513

PANEL

GEN

Cardiovascular Panel

ABCC9, ABCG5, ABCG8, ACTA1, ACTA2, ACTC1, ACTN2, AKAP9, ALMS1, ANK2, ANKRD1, APOA4, APOA5, APOB, APOC2, APOE, BAG3, BRAF, CACNA1C, CACNA2D1, CACNB2, CALM1, CALR3, CASQ2, CAV3, CBL, CBS, CETP, COL3A1, COL5A1, COL5A2, COX15, CREB3L3, CRELD1, CRYAB, CSRP3, CTF1, DES, DMD, DNAJC19, DOLK, DPP6, DSC2, DSG2, DSP, DTNA, EFEMP2, ELN, EMD, EYA4, FBN1, FBN2, FHL1, FHL2, FKRP, FKTN, FXN, GAA, GATAD1, GCKR, GJA5, GLA, GPD1L, GPIHBP1, HADHA, HCN4, HFE, HRAS, HSPB8, ILK, JAG1, JPH2, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KLF10, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LDLR, LDLRAP1, LMF1, LMNA, LPL, LTBP2, MAP2K1, MAP2K2, MIB1, MURC, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYO6, MYOZ2, MYPN, NEXN, NKX2-5, NODAL, NOTCH1, NPPA, NRAS, PCSK9, PDLIM3, PKP2, PLN, PRDM16, PRKAG2, PRKAR1A, PTPN11, RAF1, RANGRF, RBM20, RYR1, RYR2, SALL4, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCO2, SDHA, SEPN1, SGCB, SGCD, SGCG, SHOC2, SLC25A4, SLC2A10, SMAD3, SMAD4, SNTA1, SOS1, SREBF2, TAZ, TBX20, TBX3, TBX5, TCAP, TGFB2, TGFB3, TGFB1, TGFB2, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TTN, TTR, TXNRD2, VCL, ZBTB17, ZHX3, ZIC3

Myeloid Leukemia Panel

ANKRD26, ASXL1, ATRX, BCOR, BCORL1, BRAF, CALR, CBL, CBLB, CEBPA, CSF3R, DDX41, DNMT3A, ETV6, EZH2, FLT3, GATA1, GATA2, HRAS, IDH1, IDH2, JAK2, JAK3, KDM6A, Panel, KRAS, MPL, NOTCH1, NPM1, NRAS, PDGFRA, PHF6, PPM1D, PTPN11, RAD21, RUNX1, SETBP1, SF3B1, SMC1A, SMC3, SRSF2, STAG1, STAG2, STAT3, TET2, TP53, U2AF1, WT1, ZRSR2

Hearing Loss and Deafness Panel

CDH23, CLRN1, COCH, COL11A1, COL2A1, DIAPH1, EDNRB, EYA1, GJB2, GJB6, KCNE1, KCNQ1, KCNQ4, MITF, MYO15A, MYO7A, OTOF, PAX3, POU3F4, SIX5, SLC26A4, SNAI2, SOX10, TECTA, TMC1, TMIE, TMPRSS3, USH1C, USH2A, WFS1

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Neuromuscular Panel

AARS, ABCB7, ABCD1, ABHD12, ACAD9, ACADL, ACADM, ACO2, ACTA1, ADCK3, AFG3L2, AGL, AIFM1, ALDH3A2, AMPD1, ANO10, ANO5, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APTX, ARSA, ATCAY, a, ATM, ATP2A1, ATP7A, ATP7B, ATP8A2, BAG3, BEAN1, BIN1, BSCL2, C10orf2, C12orf65, C19orf12, CACNA1A, CACNA1S, CACNB4, CAPN3, CASK, CAV3, CCDC78, CCDC88C, CFL2, CHAT, CHRNA1, CHRN1, CHRND, CHRNE, CHRNG, CLCN1, CLCN2, CLN5, CNTN1, COL6A1, COL6A2, COL6A3, COLQ, CPT1B, CPT2, CRYAB, CTD1P1, CWF19L1, CYP27A1, CYP2U1, CYP7B1, DAG1, DCTN1, DDHD1, DDHD2, DES, DMD, DNAJB2, DNAJB6, DNMT2, DNMT1, DOK7, DYNC1H1, DYSL, EEF2, EGR2, ELOVL4, ELOVL5, EMD, ERLIN2, ETF, ETFB, FA2H, FAM134B, FGD4, FGF14, FHL1, FIG4, FKBP, FKTN, FLNC, FLVCR1, FRMD7, FUS, FXN, GAA, GAD1, GALC, GAN, GARS, GBA2, GDAP1, GJB1, GJC2, GLA, GLE1, GNB4, GNE, GOSR2, GPR143, GRID2, GRM1, GYS1, HADHA, HADHB, HINT1, HOXD10, HSPB1, HSPB8, HSPD1, HSPG2, IGHMBP2, IKBKAP, ISPD, ITGA7, ITPR1, JPH3, KBTBD13, KCNA1, KCNC3, KCND3, KCNE3, KCNJ10, KCNJ18, KIAA0196, KIF1A, KIF1B, KIF1C, KIF5A, KLHL40, KLHL41, L1CAM, LAMA1, LAMA2, LARGE, LDB3, LITAF, LMNA, LPIN1, LRSAM1, MARS, MARS2, MATR3, MED25, MFN2, MPZ, MRE11A, MTM1, MTMR14, MTMR2, MTPAP, MTPP, MUSK, MYF6, MYH2, MYH7, MYOT, NDRG1, NEB, NEFL, NGF, NIPA1, NOP56, NTRK1, OPA1, OPA3, OPHN1, PABPN1, PANK2, PDK3, PDYN, PEX7, PFKM, PGAM2, PHKA1, PHYH, PLEC, PLEKHG5, PLP1, PMM2, PMP22, PNKP, PNPLA6, POLG, POLG2, POMGNT1, POMT1, POMT2, PRKCG, PRPS1, PRX, PTF1A, PTRF, PYGM, RAB7A, RAPSIN, REEP1, RNF216, RRM2B, RTN2, RUBCN, RYR1, RYR2, SACS, SBF2, SCN4A, SCN9A, SEPN1, SETX, SGCA, SGCB, SGCD, SGCE, SGCG, SH3TC2, SIL1, SLC12A6, SLC16A2, SLC1A3, SLC33A1, SLC39A4, SLC52A2, SLC9A1, SLC9A6, SMN1, SNX14, SOD1, SPAST, SPG11, SPG20, SPG21, SPG7, SPTBN2, SPTLC1, SPTLC2, STAC3, STUB1, SUCLA2, SYNE1, SYNE2, SYT14, TBP, TCAP, TDP1, TECPR2, TGM6, TK2, TMEM240, TNNT2, TNNT1, TPM2, TPM3, TPP1, TRIM32, TRPV4, TTBK2, TTN, TTPA, TTR, TUBB4A, TYMP, VAMP1, VCP, VLDLR, VPS13A, VPS37A, VRK1, WFS1, WNK1, WWOX, XK, YARS, ZFYVE26, ZFYVE27, ZNF592

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Epilepsy Panel

AARS, ABCA13, ABCB11, ADGRV1, ADSL, ALDH7A1, ALG13, ARHGEF15, ARHGEF9, ARX, ASAH1, ATP1A2, ATP6AP2, CACNA1A, CASK, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNA7, CHRN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNTNAP2, COG1, COL4A4, CSTB, CTSD, DCX, DEPDC5, DLG3, DNAJC5, DNMT1, DNMT3, 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, L2HGDH, LAMA3, LIG1, MAGI2, MBD5, MECP2, MEF2C, MFSD8, NDUFV3, NECAP1, NHLRC1, NPHS2, NR2F1, NRXN1, PCDH19, PDP1, PIGA, PIGO, PIQ, PIGV, PLCB1, PNKP, PNPO, POLG, PPT1, PRICKLE1, PRICKLE2, PRRT2, QARS, RELN, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SERPIND1, SLC12A6, SLC13A5, SLC25A22, SLC2A1, SLC35A2, SLC6A8, SLC9A6, SMS, SOX6, SPTAN1, SRPX2, SRY, ST3GAL3, STXBP1, SUMF1, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TCF4, TDRD7, TPP1, TSC1, TSC2, UBE3A, VCAN, WDR45, WNK1, WWOX, ZEB2

Alzheimer-Parkinson-Dementia Panel

AARS, ABCA13, ABCA7, ABCB11, ADCY5, ANG, ANO3, APP, ATP13A2, ATP1A3, ATP7B, C19orf12, CACNA1B, CHCHD10, CHMP2B, CIZ1, COG1, COL4A4, COL6A3, DCTN1, DNMT1, EVC, FERMT1, FREM2, FUS, GBA, GCH1, GNAL, GNAO1, GRM1, GRN, HPCA, HPSE2, IL12RB2, KCTD17, KMT2B, L2HGDH, LAMA3, LRRK2, MAPT, MECP2, NDUFV3, NPHS2, PANK2, PARK7, PDP1, PINK1, PLA2G6, PNKD, PRKN, PRKRA, PRNP, PRRT2, PSEN1, PSEN2, RELN, SERPIND1, SGCE, SIGMAR1, SLC12A6, SLC19A3, SLC2A1, SLC30A10, SLC6A3, SNCA, SORL1, SOX6, SQSTM1, SRY, SUMF1, TAF1, TARDBP, TBK1, TDRD7, TH, THAP1, TIMM8A, TOR1A, TREM2, TUBA4A, TUBB4A, VAC14, VCAN, VCP, VPS13A, WNK1

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Short Stature Panel

AARS, ABCA13, ABCB11, ACTA2, ADAMTS10, ADAMTS2, ADAMTSL4, AGPS, ALPL, 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, 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, FOXE3, FREM2, GH1, GHR, GHRHR, GLI2, GLI3, GNAS, GNPAT, GRM1, HESX1, HPSE2, HRAS, HSPG2, IFITM5, IFT80, IGF1, IGF1R, IL12RB2, INPPL1, KCNJ2, KCNJ8, KDM6A, KMT2D, KRAS, L2HGDH, LAMA3, LBR, LHX3, LIFR, LOX, LTBP2, LZTR1, MAP2K1, MAP2K2, MAT2A, MATN3, MED12, MFAP5, MYH11, MYLK, NBAS, NBN, NDUFV3, NEK1, NF1, NIPBL, NPHS2, NRAS, NSDHL, OBSL1, ORC1, ORC4, ORC6, P3H1, PCNT, PDP1, PEX7, PHEX, PLOD1, POR, POU1F1, PPIB, PPP1CB, PRKG1, PROP1, PTPN11, PYCR1, RAF1, RIN2, RIT1, RMRP, ROR2, RPS6KA3, RUNX2, SBDS, SERPIND1, SERPINH1, SHOC2, SKI, SLC12A6, SLC26A2, SLC2A10, SLC34A3, SLC35D1, SLC39A13, SMAD3, SMARCAL1, SMC1A, SMC3, SMS, SOS1, SOS2, SOX3, SOX6, SOX9, SPRED1, SRCAP, SRY, SUMF1, TDRD7, TGFB1, TGFB2, TGFB3, TGFB1, TGFB2, THRB, TRIM37, TRIP11, TRPS1, TRPV4, TTC21B, VCAN, WDR19, WDR35, WNK1, WRN

Solid Tumor Panel

ABL1, AKT1, ALK, APC, ATM, BRAF, BRCA1, BRCA2, CDH1, CDKN2A, CSF1R, CTNNA1, DLC1, EGFR, ERBB2, ERBB4, ESR1, FBXW7, FGFR1, FGFR2, FGFR3, FTSJ3, GNA11, GNAQ, GNAS, HNF1A, HRAS, IDH1, IDH2, JAK2, JAK3, KCNB2, KDR, KRAS, MET, MLH1, MYC, MYCN, NOTCH1, NRAS, NRXN1, PDGFRA, PIK3CA, PTEN, PTPN11, RB1, RBAK, RET, ROS1, SMAD4, SMARCB1, SMO, SMURF1, SRC, SSFA2, STK11, TP53, VHL, ZNF594

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Skin Disorders Panel

ABCA12, ABCB6, ABCC6, ABHD5, ADAMTS2, ADAR, ALAD, ALAS2, ALDH3A2, ALOX12B, ALOXE3, AP1S1, ATM, ATP2A2, ATP2C1, ATP6V0A2, BLM, CARD14, CDH3, CDSN, CLDN1, COL17A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL7A1, CPOX, CTC1, 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, GPR143, GSN, GTF2H5, HFE, HMBS, HR, IL36RN, ITGA3, ITGA6, ITGB4, JUP, Panel, 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, NOP10, NSDHL, OCA2, PKP1, PLEC, PLOD1, PNPLA1, POFUT1, POGLUT1, POLH, POMP, PPOX, PRKAR1A, PTCH1, PTCH2, PYCR1, RECQL4, RTEL1, SLC27A4, SLC39A4, SLC45A2, SLURP1, SNAP29, SPINK5, SPRED1, ST14, STAT3, STS, SUFU, TERC, TERT, TGM1, TGM5, TINF2, TNXB, TRPV3, TSC1, TSC2, TTR, TYK2, TYR, TYRP1, UROD, UROS, WAS, WRAP53, XPA, XPC, ZMPSTE24

Lymphoid Leukemia Panel

AARS, ABCA13, ABCB11, ABL1, BRAF, BTG1, CDKN2A, COG1, COL4A4, CREBBP, CRLF2, DNMT2, DNMT1, DNMT3A, EP300, ETV6, EVC, EZH2, FBXW7, FERMT1, FLT3, FREM2, GATA3, GRM1, HPSE2, IDH1, IDH2, IKZF1, IL12RB2, IL7R, JAK1, JAK2, JAK3, KDM6A, KMT2A, KMT2D, KRAS, L2HGDH, LAMA3, LEF1, LMO1, MAPK1, NDUFV3, NF1, NOTCH1, NPHS2, NRAS, NSD2, NT5C2, NUDT15, PAX5, PDP1, PHF6, PTEN, PTPN11, RB1, RUNX1, SERPIND, SETD2, SH2B3, SLC12A6, SOX6, SRY, STAG2, STAT3, STAT5B, SUMF1, TBL1XR1, TCF3, TDRD7, TP53, TPMT, VCAN, WNK1, WT1

Lymphoma Panel

AARS, ABCA13, ABCB11, ALK, ATM, B2M, BCL6, BIRC3, BRAF, BTK, CARD11, CD79A, CD79B, COG1, COL4A4, CREBBP, CXCR4, DNMT1, EGR2, EP300, EVC, EZH2, FAS, FAT4, FBXO11, FERMT1, FREM2, GRM1, HPSE2, ID3, IDH2, IKBKB, IKZF1, IL12RB2, JAK3, KLF2, L2HGDH, LAMA3, MYC, MYD88, NDUFV3, NFKBIE, NOTCH1, NOTCH2, NPHS2, PDP1, PLCG1, PLCG2, POT1, PRDM1, RHOA, RPS15, RRAGC, SERPIND1, SF3B1, SLC12A6, SOCS1, SOX6, SRY, STAT3, STAT5B, SUMF1, TBL1XR1, TCF3, TDRD7, TET2, TNFAIP3, TNFRSF14, TP53, TP63, TRAF3, UBR5, VCAN, WNK1, XPO1

Thrombocytopenia Panel

ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, CD36, CYCS, ETV6, FLI1, FLNA, FYB1, GATA1, GFI1B, GP1BB, GP9, HOXA11, ITGA2B, ITGB3, MASTL, MECOM, MPL, MYH9, NBEAL2, PRKACG, RBM8A, RUNX1, TUBB1, WAS

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|--------------------------|--|--|
| <input type="checkbox"/> | H. Haemolytic Anemia - N. Hyperbilirubinemia Panel | ALDOA, ANK1, EPB42, EPB41, G6PD, GPI, GSR, GSS, HK1, KCNN4,NT5C3A,PFKL,PFKM,PGK1,PIEZO1, PKLR, RHAG, SLC4A1, SLCO1B3, SPTA1, SPTB, TPI1, UGT1A1 |
| <input type="checkbox"/> | aHUS SEMD RMP Panel | ADAMTS13, C3, CD46, CFB, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, THBD, ACAN,CHST3,COL2A1,DDR2,DYM,EIF2AK3,MATN3,NKX 3-2,SLC39A13,TRAPPC2,TRPV4,WISP3, C11ORF80, MEI1, KHDC3L, NLRP7, PADI6 |
| <input type="checkbox"/> | CMT Hereditary Neuropathy Panel | GDAP1, GJB1, HINT1, MFN2, MPZ, PMP22, SH3TC2, AARS1, ABHD12, AIFM1, ARHGEF10, ATP1A1, ATP7A, BAG3, BSCL2, CNTNAP1, COA7, DCTN1, DCTN2, DGAT2, DHTKD1, DNAJB2, DNMT1, DNM2, DRP2, DYNC1H1, EGR2, FGD4, FIG4, GARS1, GNB4, HARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MARS1, MCM3AP, MED25, MME, MORC2, MPV17, MTMR2, NAGLU, NDRG1, NEFH, NEFL, PDK3, PLEKHG5, PMP2, PRPS1, PRX, PTRH2, RAB7A, SBF1, SBF2, SCO2, SETX, SIGMAR1, SGPL1, SPG11, SPTLC1, TRIM2, TRPV4, TTR, VCP, WARS, YARS1 |
| <input type="checkbox"/> | Hypogonadotropic hypogonadism Panel | CHD7, DMXL2, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GNRH1, GNRHR, HS6ST1, IL17RD, ANOS1, KISS1, KISS1R, LHB, NR0B1, NSMF, POLR3A, POLR3B, PROK2, PROKR2, RAB18, RAB3GAP1, RAB3GAP2, RNF216, SEMA3A, SOX10, SPRY4, TAC3, TACR3, TBC1D20, WDR11 |
| <input type="checkbox"/> | Inherited Diseases Panel | 582 Gen içerir |
| <input type="checkbox"/> | Pan Cancer 405 Panel | 405 Gen ve Gen Füzyonları |
| <input type="checkbox"/> | Clinical Exome Panel | 5300 Gen ve Hot Spot Bölgeler |
| <input type="checkbox"/> | Tüm Ekzom | Yaklaşık 22.000 Gen |
| <input type="checkbox"/> | Tüm Genom | Tüm Genom Dizi analizi |

Hastanın Adı Soyadı

İmzası