Children's Hospital of Philadelphia℠

LAB-1514
Rev. 1/25

LAST NAME | FIRST NAME

MR# | DOB

PLACE PATIENT LABEL HERE *OR* COMPLETE ABOVE

DO NOT HANDWRITE PATIENT INFORMATION HERE

Division of Genomic Diagnostics
GENOMIC DIAGNOSTICS TEST REQUISITION

*For Cancer tests please use the Cancer Test Requisition.

SHIP TO: Children's Hospital of Philadelphia, Genomic Diagnostic Laboratory, 3615 Civic Center Blvd., Abramson Research Center, 714J, Philadelphia, PA 19104-4302 - Phone: (267) 426-1447

PATIENT INFORMATION

Patient Name (Last, First, Middle):

Ordering Facility MRN:

☐ Fetus of above patient (Check if prenatal sample)

Sex: ☐ Male ☐ Female ☐ Unknown ☐ Other: _____

Date of Birth (Month/Day/Year): _____

Street Address: _____

City/State: _____

Zip/Country: _____

Phone Number: (_____)_____

Race/Ethnicity:

☐ Amish ☐ Asian ☐ Black/African-American
☐ Caucasian ☐ East Indian ☐ French Canadian
☐ Hispanic ☐ Jewish-Ashkenazi ☐ Jewish-Sephardic
☐ Mediterranean ☐ Native American
☐ Other: _____

SAMPLE INFORMATION

Collection Date (Month/Day/Year): _____

Did the patient previously have a bone marrow transplant? ☐ Yes ☐ No

Sample Type (Please see Sample Requirements Page):

☐ Blood — Source of Blood:
- ☐ Purple Top Tube (EDTA) — ☐ Peripheral
- ☐ Green Top Tube (Sodium Heparin) — ☐ +Cord Blood
- ☐ Other Tube (Specify): _____ — ☐ Other (Specify): _____

☐ Cerebral Spinal Fluid
☐ *Cultured CVS Cells ☐ *Cultured Amniotic Fluid
☐ *Cheek Brush ☐ *Saliva

For the sample types listed below, please specify tissue of origin (required): _____

☐ *Frozen Tissue ☐ OCT ☐ Snap or Flash Frozen
☐ *Fresh Tissue ☐ Cultured Fibroblasts ☐ **DNA:
☐ Other (Specify): _____

**Please contact the lab if sending this sample type*

*** Were the nucleic acids (DNA or RNA) extracted from a CAP or CLIA certified laboratory (as required)? ☐ Yes ☐ No*

+ Two samples and an order for Maternal Cell Contamination (Page 4) are required for this specimen type:
- 3-5mL patient cord blood in a purple top (EDTA) tube
- 3-5mL maternal blood in a purple top (EDTA) tube

ORDERING PROVIDER

Name (Last, First, Degree)

(_____) _____ Phone

(_____) _____ Fax

Institution

Street Address

City State

Zip Country

Email

ORDERING LABORATORY

Name (Last, First, Degree)

(_____) _____ Phone

(_____) _____ Fax

Institution

Street Address

City State

Zip Country

Email

OTHER ORDERING PROVIDER / GENETIC COUNSELOR

Name (Last, First, Degree)

(_____) _____ Phone

(_____) _____ Fax

Email

For Lab Use Only
Type of billing: ☐ Institutional ☐ CHOP ☐ Self-pay ☐ Institution called
Comments:

Received by: Received Date: Received Time: Sample:

Phone: (267) 426-1447; Fax: (215) 590-3514; Email: dgdgeneticcounselor@chop.edu;
Website: http://www.chop.edu/centers-programs/division-genomic-diagnostics

*Testing will not be initiated until all necessary samples and clinical information are submitted to the laboratory.

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INDICATION FOR TESTING

Reason for Testing (please include pertinent history and findings, including pathology report): _____

_____

_____

Family History (Attach Pedigree if available): _____

_____

_____

Previous Relevant Genetic Testing:

Previous Relevant Genetic Testing Performed (Attach a copy): ☐ Yes ☐ No

If yes, sample type: ☐ Cultured cells from Chorionic Villus ☐ Cultured Amniocytes ☐ Blood ☐ Bone Marrow

☐ Tissue ☐ Other: _____

Results: _____

Known Genetic Abnormality/Familial Testing:

Is the current patient who is being tested similarly affected to proband (person originally tested)? ☐ Yes ☐ No

If yes, please describe:_____

Attach proband (originally tested family member) report if available.

Gene/Transcript/Region: _____ Alteration (c., p., and/or g.): _____

Proband (originally tested family member) Name (Last, First): _____

Proband (originally tested family member) Date of Birth (Month/Day/Year):_____

Current patient's relationship to the proband (originally tested family member): _____

Please Note: If family member samples are submitted for testing to clarify the interpretation of a variant of uncertain significance for a proband, the family member(s) results will be reported as an addendum on the proband's report. In the case that the provider for the family member is not the same as for the proband, a CHOP HIPAA release is required for release of the results.

CHOP HIPAA release included? ☐ Yes ☐ No

*Please contact the laboratory to request expedited testing.

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TEST MENU

**For known genetic abnormality/familial testing for any gene offered by the lab, please refer to the Known Genetic Abnormality section under "Indication for Testing."*

Chromosome Analysis

Chromosome Analysis:
- ☐ Karyotype
- ☐ Mosaicism Screen
- ☐ Parental Study

Fluorescence In Situ Hybridization (FISH):
- ☐ Indicate specific syndrome or region of interest _____

_____

**Contact the lab to confirm availability of FISH probe. There are over 100 probes available.*

Parental/Familial study:
- ☐ Indicate cytogenetic finding in family member and include a copy of the family member's report: _____

_____

Tissue Culture Services

- ☐ Thaw and Expansion
- ☐ Tissue Culture and Storage (fibroblast)

DNA/RNA Extraction

- ☐ DNA Extraction
- ☐ RNA Extraction

Nuclear Genome Wide Testing

Chromosomal Microarray Analysis:
- ☐ Chromosomal SNP Microarray
- ☐ Parental/Familial Studies (Genome Wide SNP Array)

Medical Exome Analysis:
- ☐ Medical Exome*

Medical Exome + MitoGenome Analysis:
- ☐ Medical Exome*
- ☐ MitoGenome Sequencing + Deletion Analysis

Medical Exome Reanalysis:
- ☐ Medical Exome Reanalysis*

Reflex to Medical Exome from Exome Panel
- ☐ Medical Exome from Exome Panel*

**Please submit Exome requisition and consent documents separately*

Panel Testing

For a current list of genes on each panel please see pages 5, 6, 7

Rare Disease Panel Testing:
- ☐ Alagille Syndrome Panel
- ☐ Alport Syndrome Panel
- ☐ Branchiootorenal Spectrum Disorder Panel
- ☐ Cholestasis Panel
- ☐ Congenital Diarrhea Panel
- ☐ Connective Tissue Panel
- ☐ Cornelia de Lange Syndrome Panel
- ☐ Craniofacial Panel
- ☐ Craniosynostosis Panel
- ☐ Epilepsy Panel
- ☐ Hearing Loss Panel (Comprehensive AUDIOME)
- ☐ Hemophagocytic Lymphohistiocytosis (HLH) Panel
- ☐ Rapid Hemophagocytic Lymphohistiocytosis (HLH) Panel
- ☐ Hereditary Pancreatitis Panel
- ☐ Kabuki Syndrome Panel
- ☐ Ketotic Hypoglycemia Panel

Panel Testing (cont'd)

For a current list of genes on each panel please see pages 5, 6, 7

- ☐ Neonatal Respiratory Distress Panel
- ☐ Noonan Spectrum Disorders Panel
- ☐ Osteogenesis Imperfecta Panel
- ☐ Pneumothorax Panel
- ☐ Primary Ciliary Dyskinesia Panel
- ☐ Pulmonary Arterial Hypertension Panel
- ☐ Pulmonary-Vascular Panel, Comprehensive
- ☐ Rubinstein-Taybi Syndrome Panel

Sickle Cell Disease Globin Panel
- ☐ *HBB* Sequence Analysis
- ☐ Copy Number Analysis of *HBA1, HBA2,* and *HBB*

- ☐ Stickler Syndrome Panel
- ☐ Very Early Onset Inflammatory Bowel Disease (VEO-IBD) Panel
- ☐ Waardenburg Syndrome Panel

Cancer (Germline) Panel Testing:
- ☐ Comprehensive Hereditary Cancer Panel
- ☐ *ALK*/*PHOX2B* Germline Analysis (Hereditary Neuroblastoma)

Bone Marrow Failure/Myelodysplastic Syndrome/Leukemia Testing:
- ☐ Bone Marrow Failure Panel
- ☐ Fanconi Anemia NGS Panel
- ☐ Inherited Red Blood Cell Disorder Panel
- ☐ Platelet Disorder Panel
- ☐ Telomere Disorder Panel

Mitochondrial Genome Testing

- ☐ MitoGenome Sequencing and Deletion Analysis*
- ☐ Rapid MitoGenome Sequencing and Deletion Analysis*
- ☐ mtDNA content Analysis

**Please fill out maternal relative information section on page 8 if submitting a maternal relative as part of this analysis.*

Single Gene and Region Specific Testing

22q11.2 Deletion/Duplication (VCFS, Cat Eye Syndrome, Congenital Heart Defect, DiGeorge syndrome):
- ☐ 22q11.2 Deletion/Duplication Analysis

Angelman Syndrome:
- ☐ Chromosome 15 Methylation Analysis

Birt-Hogg-Dube Syndrome:
- ☐ *FLCN* Sequence Analysis
- ☐ *FLCN* Deletion/Duplication Analysis

Blepharophimosis-Ptosis-Epicanthus Inversus Syndrome:
- ☐ *FOXL2* Sequence Analysis
- ☐ *FOXL2* Deletion/Duplication Analysis

Brooke Spiegler Syndrome:
- ☐ *CYLD* Sequence Analysis

CADASIL:
- ☐ *NOTCH3* Sequence Analysis

CASK-Related Disorders:
- ☐ *CASK* Sequence Analysis

CHARGE Syndrome:
- ☐ *CHD7* Sequence And Deletion/Duplication Analysis

Congenital Central Hypoventilation Syndrome:
- ☐ *PHOX2B* Sequence Analysis including polyalanine repeat analysis

Craniofrontonasal Syndrome:
- ☐ *EFNB1* Sequence Analysis
- ☐ *EFNB1* Deletion/Duplication Analysis

Cystic Fibrosis:
- ☐ *CFTR* Sequence And Deletion/Duplication Analysis

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Single Gene and Region Specific Testing (cont'd)

DFNB1-Related Hearing Loss:
- ☐ DFNB1 locus testing (*GJB2* sequence analysis and targeted deletion testing for the ΔGJB6-D13S1830 variant)
- ☐ *GJB2* sequencing
- ☐ Targeted deletion testing for the ΔGJB6-D13S1830 variant

Fragile X Syndrome:
- ☐ Triplet Repeat Analysis

Gilbert's Syndrome:
- ☐ *UGT1A1* Promoter Analysis

Hereditary Leiomyomatosis and Renal Cell Carcinoma:
- ☐ *FH* Sequence Analysis
- ☐ *FH* Deletion/Duplication Analysis

Li Fraumeni:
- ☐ *TP53* Sequence Analysis
- ☐ *TP53* Deletion/Duplication Analysis

Marfan Syndrome:
- ☐ *FBN1* Sequence And Deletion/Duplication Analysis

Multiple Endocrine Neoplasia, Type 2:
- ☐ *RET* Sequence Analysis

Neuroblastoma (Germline Analysis- For somatic testing, see Cancer Test Requisition):
- ☐ *ALK*/*PHOX2B* Panel (Sequencing Analysis of *ALK* and *PHOX2B* and Deletion/Duplication Analysis of *PHOX2B*)
- ☐ *ALK* Sequence Analysis
- ☐ *PHOX2B* Sequence Analysis

Opitz G/BBB Syndrome:
- ☐ *MID1* Sequence Analysis
- ☐ *MID1* Deletion/Duplication Analysis

***OTOF*-related Hearing Loss:**
- ☐ *OTOF* Seq + Del/Dup

***PMS2*-related Lynch Syndrome:**
- ☐ *PMS2* Sequencing and Del/Dup Analysis via Long Range PCR (LR-PCR) + NGS

PTEN Related Autism/PTEN Hamartoma Tumor Syndrome:
- ☐ *PTEN* Sequence Analysis
- ☐ *PTEN* Deletion/Duplication Analysis

Prader-Willi Syndrome:
- ☐ Chromosome 15 Methylation Analysis

Rett Syndrome:
- ☐ *MECP2* Sequence Analysis
- ☐ *MECP2* Deletion/Duplication Analysis

Saethre Chotzen Syndrome Panel:
- ☐ *TWIST1* Sequence Analysis
- ☐ *TWIST1* Deletion/Duplication Analysis and *FGFR3* Analysis for p.Pro250Arg

SMARCB1 Related Disorders:
- ☐ *SMARCB1* Sequence Analysis
- ☐ *SMARCB1* Deletion/Duplication Analysis

Single Gene and Region Specific Testing (cont'd)

SHOX Related Disorders:
- ☐ *SHOX* Deletion/Duplication Analysis
- ☐ *SHOX* Sequence Analysis

Spinal Muscular Atrophy:
- ☐ SMN1 & SMN2 Copy Number Analysis

STRC-Related Hearing Loss:
- ☐ *STRC* Deletion/Duplication Analysis
- ☐ *STRC* Sequence Analysis

Swachman-Diamond Syndrome Testing:
- ☐ *SBDS* Sequencing and Del/Dup Analysis via long range PCR (LR-PCR) + NGS

Thalassemia/Sickle Cell Disease:
- ☐ Sickle Cell Disease Globin Panel (sequencing of *HBB* and Deletion/Duplication Analysis of *HBB, HBA1, HBA2*)
- ☐ Alpha Globin (*HBA1* and *HBA2*) Deletion/Duplication Analysis
- ☐ Alpha Globin (*HBA1* and *HBA2*) Sequence Analysis
- ☐ Beta Globin (*HBB*) Sequence Analysis
- ☐ Beta Globin (*HBB*) Deletion/Duplication Analysis

Thrombophilia:
- ☐ Factor II Sequence Analysis for c.*97G>A
- ☐ Factor V Sequence Analysis for c.1601G>A

Von Hippel Lindau:
- ☐ *VHL* Sequence Analysis
- ☐ *VHL* Deletion/Duplication Analysis

Pharmacogenomic Testing

Mercaptopurine Intolerance:
- ☐ *NUDT15* Sequence Analysis
- ☐ *TPMT* Sequence Analysis

Maternal Cell Contamination

Maternal Cell Contamination:
- ☐ Child/Fetal sample assessment
- ☐ Maternal comparative sample assessment

*Paternity testing not performed.

Known Variant Testing

Please fill out the known genetic abnormality section on page 2

Sequencing Variant*
- ☐ Sequence Variant

Copy Number Variant*
- ☐ Copy Number Variant

Mitochondrial Genome Variant*
- ☐ Targeted mtDNA Variant

*Please check if any of the following apply:
☐ Lab Requested ☐ Research ☐ Positive Control

PANEL TESTING: GENE CONTENT

The following panels include both sequence and copy number analysis, unless otherwise indicated. * Designates panels/genes with analysis by sequencing only; ** Designates genes with copy number analysis only. For CPT codes and turnaround times, please visit our website at https://www.testmenu.com/chop.

Alagille Syndrome Panel	*JAG1, NOTCH2*
Alport Syndrome Panel	*COL4A3, COL4A4, COL4A5, COL4A6*
Branchiootorenal Spectrum Disorder Panel	*EYA1, SIX1, SIX5*
Cholestasis Panel	*ABCB4, ABCB11, ABCC2, AKR1D1, ATP8B1, BAAT, CFTR[φ], CLDN1, HNF1A, HNF1B, HSD3B7, JAG1, KIF12, LIPA, MYO5B, NOTCH2, NPC1, NPC2, NR1H4, SERPINA1, SLC25A13, TJP2, UNC45A, VIPAS39, VPS33B* [φ]Deep intronic variants c.3718-2477 and c.1680-886A>G and the intron 8 poly T tract in *CFTR* are included in the analysis.
Congenital Diarrhea Panel	*APOB, ARX, CD55, CFTR[φ], DGAT1, EPCAM, FLNA, GUCY2C, LCT, LIPA, MTTP, MYO5B, NEUROG3, PCSK1, PLVAP, PNLIP, RFX6, SAR1B, SBDS, SI, SKIV2L, SLC10A2, SLC26A3, SLC39A4, SLC51B, SLC5A1, SLC9A3, SPINT2, STX3, TTC37, UNC45A, WNT2B* [φ]Deep intronic variants c.3718-2477 and c.1680-886A>G and the intron 8 poly T tract in *CFTR* are included in the analysis.

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Connective Tissue Panel	*ACTA2, B3GAT3, BGN, C1R, C1S, COL1A1, COL1A2, COL3A1, COL4A5, COL5A1, COL5A2, EFEMP2, ELN, EPHB4, FBLN5, FBN1, FBN2, FLNA, FOXE3, GATA5, LOX, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PLOD3, PRKG1*, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2*
Cornelia de Lange Syndrome Panel	*AFF4, ANKRD11, ASXL1, ASXL3, HDAC8, NIPBL, PACS1, RAD21, SMC1A, SMC3*
Craniofacial Panel	*ALPL, ALX1, ALX3, ALX4, CD96, CHD7, DHODH, EFNB1, EFTUD2, ERF, EVC, EVC2, EYA1, FGFR1, FGFR2, FGFR3, GLI3, IFT122, IFT43, MEGF8, MIR17HG**, MSX2, MYCN, PDE4D, PLCB4, POLR1C, POLR1D, POR, PRKAR1A, RAB23, RECQL4, RUNX2, SALL1, SF3B4, SIX1, SIX5, SKI, SPECC1L, TCF12, TCOF1, TWIST1, WDR19, WDR35, ZIC1*
Craniosynostosis Panel	Gene list V2: *ALPL, ASXL1, CD96, CDC45, CYP26B1, EFNB1, ERF, FGFR1, FGFR2, FGFR3, IFT122, IFT43, IL11RA, MASP1, MEGF8, MSX2, POR, RAB23, RECQL4, RUNX2, SKI, SLC25A24, SMAD6, SPECC1L, TCF12, TGFBR1, TGFBR2, TWIST1, WDR35, ZIC1, ZNF462.*
Epilepsy Panel	*ALDH5A1, ALDH7A1, ALG13, ANKRD11, AP3B2, ARHGEF9, ARID1B, ARX, ASAH1, ASXL3, ATP1A2, ATP1A3, CACNA1A, CACNA1E, CACNA1I, CARS2, CASR, CDKL5, CERT1, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNKSR2, CTSD, CTSF, DCX, DDX3X, DEPDC5, DNM1, DYNC1H1, DYRK1A, EEF1A2, EPM2A, FGF12, FOLR1, FOXG1, FRRS1L, GABRA1, GABRB2, GABRB3, GABRG2, GAMT, GNB1, GRIN1, GRIN2A, GRIN2B, HCN1, HNRNPU, IQSEC2, ITPA, KCNA1, KCNA2, KCNB1, KCNC1, KCNH1, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCTD7, LGI1, MBD5, MECP2, MEF2C, MFSD8, MTOR, NBEA, NEXMIF, NHLRC1, NPRL2, NPRL3, PACS2, PCDH19, PGAP3, PIGA, PIGN, PIGT, PIGV, PNKP, PNPO, POLG, PPP3CA, PPT1, PRICKLE1, PRRT2, PURA, QARS1, ROGDI, RORB, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SERPINI1, SLC12A5, SLC13A5, SLC19A3, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A8, SMC1A, SNAP25, SPATA5, ST3GAL5, SYNJ1, STX1B, STXBP1, SYN1, SYNGAP1, TBC1D24, TBCK, TCF4, TPK1, TPP1, TSC1, TSC2, UBA5, UBE3A, UGDH, WDR45, WWOX, ZEB2*
Hearing loss Panel (Comprehensive AUDIOME)	*ABHD12, ACTG1*, ADCY1, ADGRV1, AFG2B (SPATA5L1), AIFM1, ALMS1, ARSG, ATP2B2, ATP6V1B1, ATP6V1B2, BCS1L, BSND, CABP2, CACNA1D, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CEP250, CEP78, CIB2, CISD2, CLDN14, CLDN9, CLIC5, CLPP, CLRN1, COCH*, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6**, COL9A1, COL9A2, COL9A3, CRYM*, DIABLO*, DIAPH1, DIAPH3*, EDN3, EDNRB, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, EYA1, EYA4, FGF3, GATA3, GIPC3, GJB2‡, GJB6**, GPSM2, GRHL2, GRXCR1, GRXCR2, GSDME, HARS2, HGF, HOMER2*, HSD17B4, ILDR1, KARS1, KCNQ1, KCNQ4, KITLG, LARS2, LHFPL5, LMX1A, LOXHD1, LRTOMT, MARVELD2, MCM2, MIR96*, MITF, MPZL2, MSRB3, MT-RNR1 (m.1555A>G)φ, MT-TS1 (m.7445A>G)φ, MYH14*, MYH9, MYO15A, MYO3A, MYO6, MYO7A, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2*, PAX3, PCDH15, PDZD7, PJVK, PLS1*, PNPT1, POU3F4‡, POU4F3, PRPS1, PTPRQ, RDX, RIPOR2, S1PR2, SERPINB6, SIX1, SLC12A2*, SLC17A8, SLC26A4, SLC52A2, SLC52A3, SLITRK6, SMPX, SNAI2, SOX10, STRC, SYNE4, TBC1D24, TECTA, TIMM8A, TJP2, TMC1, TMEM132E, TMEM43, TMIE, TMPRSS3, TPRN, TRIOBP, TUBB4B, TWNK, USH1C, USH1G, USH2A, WFS1, WHRN* φTargeted variant analysis only is performed for these genes. ‡The ROI also includes non-coding regions in the *GJB2* and *POU3F4* genes for CNV analysis.
Hemophagocytic Lymphohistiocytosis (HLH) Panel	Gene list V2: *ADA, AP3B1, AP3D1, BLOC1S6, BTK, CARMIL2, CD27, CD70, CDC42, CORO1A, CTPS1, CYBA, CYBB, CYBC1, DPP9, FAAP24, FADD, FAS, FASLG, GATA2‡, HAVCR2, HPS6, IFNGR1, IFNGR2, IKBKG*#, IKZF2, IL12RB1, IL2RA, IL2RG, IRF8, ITK, LAMP1, LIPA, LYST, MADD, MAGT1, MCM4, MEFV, MVK, NBAS, NCF2, NCF4, NCKAP1L, NFKB1, NLRC4, NLRP3, PIK3CD*, PIK3R1, PNP, PRF1, PRKCD, RAB27A, RASGRP1, RC3H1, RHOG, SH2D1A, SLC7A7, STAT1, STAT2, STX11, STXBP2, STXBP3, TBXAS1, TNFRSF1A, UNC13D‡, WAS, XIAP* #*IKBKG* is NOT covered on the Rapid HLH Panel analysis ‡The ROI includes a non-coding region in *GATA2* for SNV analysis, as well as the pathogenic recurrent 253kb inversion variant in *UNC13D* [PMID: 21931115].
Hemophagocytic Lymphohistiocytosis (HLH), Rapid	*ADA, AP3B1, AP3D1, BLOC1S6, BTK, CARMIL2, CD27, CD70, CDC42, CORO1A, CTPS1, CYBA, CYBB, CYBC1, DPP9, FAAP24, FADD, FAS, FASLG, GATA2‡, HAVCR2, HPS6, IFNGR1, IFNGR2, IKZF2, IL12RB1, IL2RA, IL2RG, IRF8, ITK, LAMP1, LIPA, LYST, MADD, MAGT1, MCM4, MEFV, MVK, NBAS, NCF2, NCF4, NCKAP1L, NFKB1, NLRC4, NLRP3, PIK3CD*, PIK3R1, PNP, PRF1, PRKCD, RAB27A, RASGRP1, RC3H1, RHOG, SH2D1A, SLC7A7, STAT1, STAT2, STX11, STXBP2, STXBP3, TBXAS1, TNFRSF1A, UNC13D‡, WAS, XIAP* ‡The ROI includes a non-coding region in *GATA2* for SNV analysis, as well as the pathogenic recurrent 253kb inversion variant in *UNC13D* [PMID: 21931115].
Kabuki Syndrome Panel	*KMT2D, KDM6A*
Ketotic Hypoglycemia Panel	*ACAT1, AGL, G6PC, GYS2, OXCT1, PHKA2, PHKB, PHKG2, PYGL, SLC16A1, SLC37A4*
Neonatal Respiratory Distress Panel	*ABCA3, AP3B1, COPA, CSF2RA, CSF2RB, CTC1, DKC1, FARSB, FLNA, FOXF1§, HPS1, HPS4, ITGA3, JAG1, MARS, NKX2-1, NOTCH2, OAS1, PARN, RTEL1, SFTPB, SFTPC, SLC7A7, TERC, TERT, TINF2* §The upstream regulatory region of the *FOXF1* gene is included in the analysis.
Noonan Spectrum Disorders Panel	*A2ML1*, BRAF*, CBL*, HRAS*, KRAS*, LZTR1, MAP2K1*, MAP2K2*, MRAS*, NF1*, NRAS*, PPP1CB*, PTPN11*, RAF1*, RASA2*, RIT1*, RRAS*, SHOC2*, SOS1*, SOS2*, SPRED1*
Osteogenesis Imperfecta Panel	*COL1A1, COL1A2, IFITM5**

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Hereditary Pancreatitis Panel	*CASR, CFTR[φ], CTRC, PRSS1, SPINK1* [φ]Deep intronic variants c.3718-2477 and c.1680-886A>G and the intron 8 poly T tract in *CFTR* are included in the analysis.
Pneumothorax Panel	*COL3A1, FBN1, FLCN, TGFBR1, TGFBR2*
Primary Ciliary Dyskinesia Panel	*CCDC39, CCDC40, CCDC65, CCDC103, CCNO, CFAP298, CFAP300, CFTR[φ], DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAAF11, DNAH1, DNAH5, DNAH6, DNAH8, DNAH9, DNAH11, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, GAS8, INVS, MCIDAS, NME8, ODAD1, ODAD2, ODAD3, ODAD4, OFD1, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, ZMYND10* [φ]Deep intronic variants c.3718-2477 and c.1680-886A>G and the intron 8 poly T tract in *CFTR* are included in the analysis.
Pulmonary Arterial Hypertension Panel	*ABCC8, ACVRL1, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, FOXF1[§], GDF2, KCNA5, KCNK3, RASA1, SMAD4, SMAD9, SOX17, TBX4* [§]The upstream regulatory region of the *FOXF1* gene is included in the analysis.
Comprehensive Pulmonary-Vascular Panel	*ABCA3, ABCC8, ACVRL1, AP3B1, BMPR1B, BMPR2, CAV1, CCDC39, CCDC40, CCDC65, CCDC103, CCNO, CFAP298, CFAP300, CFTR[φ], COPA, CSF2RA, CSF2RB, CTC1, DKC1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAAF11, DNAH1, DNAH5, DNAH6, DNAH8, DNAH9, DNAH11, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, EIF2AK4, ENG, FARSB, FLNA, FOXF1[§], GAS8, GDF2, HPS1, HPS4, INVS, ITGA3, JAG1, KCNA5, KCNK3, MARS1, MCIDAS, NKX2-1, NME8, NOTCH2, OAS1, ODAD1, ODAD2, ODAD3, ODAD4, OFD1, PARN, RASA1, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, RTEL1, SCNN1A, SCNN1B, SCNN1G, SFTPB, SFTPC, SLC7A7, SMAD4, SMAD9, SOX17, SPAG1, TBX4, TERC, TERT, TINF2, ZMYND10* [φ]Deep intronic variants c.3718-2477 and c.1680-886A>G and the intron 8 poly T tract in *CFTR* are included in the analysis. [§]The upstream regulatory region of the *FOXF1* gene is included in the analysis.
Rubinstein-Taybi Syndrome Pane	*CREBBP, EP300*
Sickle Cell Disease Globin Panel	*HBB*, *HBA1***, *HBA2***
Stickler Syndrome Pane	Gene list V2: *BMP4, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, GZF1, LOXL3, LRP2, PLOD3, VCAN*
Very Early Onset Inflammatory Bowel Disease (VEO-IBD) Panel	Gene list V2: *ADA, ADAM17, AICDA, AIRE, ARPC1B, BTK, CARMIL2, CD19, CD40, CD40LG, CD55, CD81, CIITA, COL7A1, CR2, CTLA4, CYBA, CYBB, DCLRE1C, DKC1, DOCK8, FERMT1, FOXP3, FUT2, G6PC3, HPS1, HPS4, HPS6, ICOS, IKBKB, IKBKG*, IKZF1, IL10, IL10RA, IL10RB, IL21, IL23R, IL2RA, IL2RG, IL7R, ITCH, ITGB2, ITK, LCK, LIG4, LRBA, LYST, MALT1, MEFV, MVK, MYO5A, NCF2, NCF4, NFAT5, NFKB1, NFKB2, NLRC4, NLRP12, NOP10, PIK3R1, PLCG2, PRF1, PTEN, RAB27A, RAC1, RAC2, RAG1, RAG2, RET, RFX5, RFXANK, RFXAP, RIPK1, RTEL1, SH2D1A, SKIV2L, SLC37A4, STAT1, STAT3, STAT5A, STAT5B, STX3, STXBP2, STXBP3, TAP1, TAP2, TERC[‡], TERT, TINF2, TNFAIP3, TNFAIP6, TNFRSF13B, TRAF3, TTC37, TTC7A, UNC13D[‡], UNG, WAS, XIAP, ZAP70* [‡]The ROI also includes a non-coding region in *TERC* for CNV analysis, as well as the pathogenic recurrent 253kb inversion variant in *UNC13D* [PMID: 21931115].
Waardenburg Syndrome Panel	*EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10*
Cancer (Germline) Panel Testing: Comprehensive Hereditary Cancer Testing	
Comprehensive Hereditary Cancer Panel	*ABCB11, AIP, ALK*, APC[‡], ARID5B, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CBL*, CDC73, CDH1, CDK4*, CDKN1B, CDKN1C, CDKN2A, CEBPA, CEBPE, CHEK2, COL7A1, CTNNA1, CYLD, DDB2, DDX41, DICER1, DIS3L2, DKC1, DOCK8, EGFR, ELANE, EPCAM**, ERCC2, ERCC3, ERCC4, ERCC5, ETV6, EXT1, EXT2, FAH, FANCA, FANCC, FANCG, FAS, FH, FLCN, G6PC3, GATA2[‡], GBA, GPC3, GREM1, HABP2, HAX1, HFE, HMBS, HRAS*, IKZF1, ITK, KIT*, KRAS*, LZTR1, MAX, MEN1, MET*, MLH1, MLH3, MPL, MSH2, MSH3, MSH6, MTAP, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PAX5, PDGFRA*, PHOX2B, PMS2, POLD1*, POLE*, POLH, POT1, PRKAR1A, PRSS1*, PTCH1, PTEN, PTPN11*, RAD50, RAD51C, RAD51D, RB1, RECQL4, RET*, RHBDF2, RMRP[‡], RNF43, RUNX1, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SERPINA1, SH2B3, SH2D1A, SLC25A13, SMAD4, SMARCA4, SMARCB1, SMARCE1, SOS1*, STAT3, STK11, SUFU, TERC[‡], TERT, TGFBR1, TINF2, TMEM127, TP53, TRIM37, TSC1, TSC2, UROD, VHL, WAS, WRN, WT1, XPA, XPC* [‡]The ROI includes promoter and/or non-coding regions of *APC* (promoter and non-coding regions for SNV/CNV analysis), *GATA2* (non-coding region for SNV analysis), *RMRP* (promoter region and noncoding region for SNV analysis), and *TERC* (promoter and non-coding regions for SNV/CNV analysis).

Division of Genomic Diagnostics
GENOMIC DIAGNOSTICS TEST REQUISITION

*For Cancer tests please use the Cancer Test Requisition.

PANEL TESTING: GENE CONTENT

The following panels include both sequence and copy number analysis, unless otherwise indicated. * Designates panels/genes with analysis by sequencing only; ** Designates genes with copy number analysis only. For CPT codes and turnaround times, please visit our website at https://www.testmenu.com/chop.

Hereditary Cancer Panels: Bone Marrow Failure/Myelodysplastic Syndrome/Leukemia Testing	
Bone Marrow Failure Panel	Gene list V2: *ABCB7, ABCG5, ABCG8, ACD, ACTN1*, ADA2, AK2, ALAS2, ANKRD26*‡, AP3B1, ATM, ATR, BLM, BRCA1, BRCA2, BRIP1, CARD11, CBL*, CDAN1, CDIN1, CDKN2A, CEBPA, CHEK2, CLPB, CSF3R, CTC1, CXCR2, CXCR4*, CYCS, DBF4, DDX11, DDX41, DKC1, DNAJC21, DNMT3A, EFL1, EIF2AK3, ELANE, EPCAM**, ERCC4, ERCC6L2, ESCO2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, G6PC3, GATA1, GATA2‡, GFI1*, GLRX5, GP1BA, GP1BB, GP9, HAX1, HEATR3, HOXA11, IKZF1, ITGA2B, ITGB3, JAGN1, KCNN4, KIF23, KLF1, KRAS*, LAMTOR2, LIG4, LYST, MAD2L2, MECOM, MLH1, MPL, MSH2, MSH6, MYH9, MYSM1, NAF1, NBN, NF1, NHEJ1, NHP2, NOP10, PALB2, PARN, PAX5, PMS2, POT1, PTPN11*, RAB27A, RAC2, RAD51, RAD51C, RBM8A, RMRP‡, RPA1*, RPL11, RPL15, RPL19, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL4, RPL5, RPL9, RPS10, RPS15A, RPS17, RPS19, RPS20, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SASH3, SBDS, SEC23B, SEC61A1, SH2B3, SH2D1A, SLC25A38, SLC37A4, SLX4, SOS1*, SRP19, SRP54, SRP68, SRP72, SRPRA, STAT3, STIM1, STK4, STN1, TAFAZZIN, TCIRG1, TERC‡, TERT, THPO, TINF2, TP53, TSR2, TUBB1, UBE2T, USB1, VPS13B, VPS45, WAS, WIPF1, WRAP53, XRCC2, ZCCHC8* ‡The ROI also includes promoter and/or non-coding regions of *ANKRD26* (non-coding region for SNV analysis), *GATA2* (non-coding region for SNV analysis), *RMRP* (promoter and non-coding region for SNV analysis), and *TERC* (promoter and noncoding regions for SNV/CNV analysis).
Fanconi Anemia NGS Panel	Gene list V2: *ATM, BLM, BRCA1, BRCA2, BRIP1, DDX11, ERCC4, ESCO2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, LIG4, MAD2L2, NBN, NHEJ1, PALB2, RAD51, RAD51C, SLX4, UBE2T, XRCC2*
Inherited Red Blood Cell Disorder Panel	Gene list V2: *ABCB7, ABCG5, ABCG8, AK1, ALAS2, ALDOA, ANK1, CDAN1, CDIN1, COL4A1, EPB41, EPB42, G6PD, GATA1, GCLC, GLRX5, GPI, GPX1, GSR, GSS, HEATR3, HK1, KCNN4, KIF23, KLF1, MYSM1, NT5C3A, PFKM, PGK1, PIEZO1, PKLR, RHAG, RPL11, RPL15, RPL19, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL4, RPL5, RPL9, RPS10, RPS15A, RPS17, RPS19, RPS20, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, SEC23B, SLC25A38, SLC2A1, SLC4A1, SPTA1, SPTB, TPI1, TSR2, XK*
Platelet Disorder Panel:	*ABCG5, ABCG8, ACTN1*, ADA2, ADAMTS13, ANKRD26*‡, ANO6, AP3B1, AP3D1, ARPC1B, BLOC1S3, BLOC1S6, C3, CARD11, CASP10, CD46, CDC42, CFB, CFH, CFHR1, CFHR3, CFHR4, CFI, COL4A1, CTLA4, CYCS, DGKE, DIAPH1, DTNBP1, ETV6, FADD, FAS, FASLG, FERMT3, FLI1, FLNA, FYB1, GATA1, GATA2‡, GFI1B, GNE, GP1BA, GP1BB, GP6, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, IKZF1, IKZF5, ITGA2, ITGA2B, ITGB3, ITK, KDSR, KRAS*, LIG4, LRBA, LYST, MAGT1, MECOM, MPIG6B, MPL, MYH9, MYO5A, NBEAL2, P2RY12, PLA2G4A, PLAU, PLG, PRKACG, PRKCD, PTGS1, PTPN11*, PTPRJ, RAB27A, RASGRP2, RBM8A, RUNX1, SH2D1A, SLC4A1, SLFN14, SRC*, SRP72, STAT3, STIM1, STX11, STXBP2, TBXA2R, TBXAS1, TCN2, THBD, THPO, TPM4, TRPM7, TUBB1, UNC13D‡, VIPAS39, VPS33B, WAS, WIPF1, XIAP* ‡The ROI also includes non-coding regions of *ANKRD26* and *GATA2* for SNV analysis, as well as the pathogenic recurrent 253kb inversion variant in *UNC13D* [PMID: 21931115].
Telomere Disorder Panel	Gene list V2: *ACD, CTC1, DKC1, NAF1, NHP2, NOP10, PARN, POT1, RPA1*, RTEL1, STN1, TERC‡, TERT, TINF2, USB1, WRAP53, ZCCHC8* ‡The ROI also includes the promoter and non-coding regions of *TERC.*

LAST NAME | FIRST NAME

MR# | DOB

PLACE PATIENT LABEL HERE *OR* COMPLETE ABOVE

DO NOT HANDWRITE PATIENT INFORMATION HERE

Division of Genomic Diagnostics
GENOMIC DIAGNOSTICS TEST REQUISITION

*For Cancer tests please use the Cancer Test Requisition.

HEARING LOSS HISTORY *(required if Hearing Loss Panel requested on page 3)*

Age of onset: ☐ Congenital ☐ Childhood (pre-lingual) ☐ Childhood/Adolescence (post-lingual) ☐ Adulthood

Type of hearing loss: ☐ Sensorineural ☐ Conductive ☐ Auditory neuropathy/dyssynchrony ☐ Mixed

Laterality: ☐ Bilateral ☐ Unilateral-R ☐ Unilateral-L

Progression: ☐ Stable ☐ Progressive ☐ Fluctuating ☐ Unknown

Severity (PTA): *Please send audiogram if performed outside CHOP

Left Ear (if sloping, check all that apply):
☐ Mild (15-30dB) ☐ Moderate (31-50dB) ☐ Moderately-severe (51-70dB) ☐ Severe (71-90dB)
☐ Profound (>90db)

Right Ear (if sloping, check all that apply):
☐ Mild (15-30dB) ☐ Moderate (31-50dB) ☐ Moderately-severe (51-70dB) ☐ Severe (71-90dB)
☐ Profound (>90db)

Audiogram shape/frequencies:

Left Ear: ☐ Flat (all frequencies) ☐ Sloping (high frequency) ☐ Saucer-shaped (mid frequency)
☐ Rising (low frequency)

Right Ear: ☐ Flat (all frequencies) ☐ Sloping (high frequency) ☐ Saucer-shaped (mid frequency)
☐ Rising (low frequency)

Exposure to aminoglycoside antibiotics (e.g gentamicin, neomycin, tobramycin, amikacin):
☐ No ☐ Yes ☐ Unknown

Visual differences: ☐ No ☐ Yes

Specify differences:

Maternal Relative Information for Mitochondrial Testing *(required if Mitochondrial Testing requested on page 4)*

A maternal relative may be submitted as part of this analysis at the time of proband specimen submission. If you are submitting a maternal relative please fill out the following information:

Name: _____

DOB: _____

Relationship to proband: _____

Specimen Source: ☐ Blood ☐ Saliva ☐ Muscle ☐ Other: _____

Is this individual affected? ☐ Yes ☐ No

If affected, what are this individual's symptoms?

Division of Genomic Diagnostics
GENOMIC DIAGNOSTICS TEST REQUISITION

*For Cancer tests please use the Cancer Test Requisition.

Sample Requirements

Blood – The Panel Testing and Single Gene and Region Specific Testing menus require 3-5mL of blood in an EDTA (purple top) tube.

Chromosomal Microarray Analysis requires 3-5mL of blood in EDTA (purple top) tube and 3-5 mL of blood in a Sodium Heparin (dark green) tube.

The Chromosome Analysis testing menu requires 3-5mL of blood in Sodium Heparin (green top) tube.

Bone Marrow – For testing on a bone marrow aspirate, 3-5mL in an EDTA (purple top) tube is the preferred minimum. Please contact the lab for exceptions.

DNA – Most tests require 5-10ug of DNA with a minimum concentration of 50ng/uL. Panel testing requires 8ug of DNA. Please refer to the divisional website for specific amounts of DNA required for each test.

Cultured CVS Cells or Cultured Amniotic Fluid – This sample type requires 2 nearly confluent T-25 flasks. Please contact the laboratory if ordering prenatal testing.

Fresh or Frozen Tumor Tissue – For Cancer Testing, 0.5cm^3 of tissue is needed.

FFPE Sample – The lab prefers to receive tumor block but can also receive 4-6 scrolls cut 50 microns thick. Slides alone cannot be used for testing, but are useful to determine tumor percentage if scrolls are sent. If sending an FFPE block that needs to be returned please provide the FedEx or UPS account number to use for return shipment along with the return address. Please reach out to the GDL to confirm the testing schedule if sending cut formalin-fixed, paraffin embedded (FFPE) scrolls or slides.

Shipping Instructions

Samples should be shipped by overnight carrier to arrive Monday – Friday (9am-5pm) only.

Most samples should be sent at room temperature. Frozen tumor samples should be sent on dry ice.

Shipping address –

Children's Hospital of Philadelphia
Genomic Diagnostics Laboratory
3615 Civic Center Blvd.
Abramson Research Center, 714J
Philadelphia, PA 19104-4302
Phone: (267) 426-1447

Necessary Documents

Each sample should be sent with a completed **Test Requisition Form**, including billing information, and **Consent Form** (if applicable).

Previous Family Reports – Please include family member test reports if ordering known genetic abnormality/familial testing.

Results from Prior Testing – Please include results from prior genetic testing or other related reports (such as hemoglobin electrophoresis for thalassemia testing, pathology reports for any testing on tumors, or other testing that may provide necessary clinical information for genetic testing).

Pedigree – Please include a copy of the patient's family pedigree, especially if other family members are similarly affected. The pedigree should include genotype and phenotype information for all tested family members.

Children's Hospital of Philadelphia℠

LAB-1514
Rev. 1/25

LAST NAME | FIRST NAME

MR# | DOB

PLACE PATIENT LABEL HERE *OR* COMPLETE ABOVE

DO NOT HANDWRITE PATIENT INFORMATION HERE

Division of Genomic Diagnostics
GENOMIC DIAGNOSTICS TEST REQUISITION

*For Cancer tests please use the Cancer Test Requisition.

Billing Options (For Non-CHOP patients only)

***By using and sending this Requisition Form to CHOP Outreach Lab for laboratory testing, you, the sender, acknowledge and agree that you have read and agree to the CHOP Terms and Conditions posted at www.chop.edu/labs and agree to pay CHOP the rates in CHOP's fee schedule in effect on the date the specimen is received.**

Institutional Billing Option

ICD-10 Diagnosis Codes for Billing: _____

Bill to Institution/Department: _____

Address: _____

_____

Billing Contact: _____

Phone: _____ Fax: _____

Email: _____

Self Pay Option

Total Cost Approved: _____ Credit Card: ☐ Visa ☐ American Express ☐ Discover ☐ MC

Name on Card: _____

Cardholder Date of Birth: _____
Month/Day/Year

Card Number: _____

Expiration Date: _____ CCV (security # on back): _____
Month/Year

Billing Address: _____

_____

Phone: _____ Email: _____

_____
Cardholder Signature | Printed Name | Date (Month/Day/Year) | Time

*Cardholders signature indicates authorization to bill Credit Card