Children's Hospital of Philadelphia℠

LAB-1514
Rev. 11/18

LAST NAME | FIRST NAME

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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 Diagnostics Laboratory, 3615 Civic Center Blvd., Abramson Research Center, 714, 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:

☒ Blood
- ☒ Purple Top Tube (EDTA)
- ☐ Green Top Tube (Sodium Heparin)
- ☐ Other Tube (Specify): _____

Source of Blood:
- ☐ Peripheral
- ☐ Cord Blood
- ☐ Other (Specify): _____

☐ Cerebral Spinal Fluid
☐ *Cultured CVS Cells
☐ *Cultured Amniotic Fluid
☐ *Cheek Brush ☐ *Saliva
☐ DNA: Specify Tissue of Origin: _____
☐ *Fresh Tissue
☐ *Frozen Tissue ☐ OCT ☐ Snap or Flash Frozen

Please contact the lab if sending this sample type

☐ Other (Specify): _____

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

Kelsen, Judith, MD
Name (Last, First, Degree)

(215) 590-7423
Phone

(267) 426-9001
Fax

kelsen@email.chop.edu
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; 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): _____

_____

_____

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 Cells from Amniocentesis ☐ 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

SEQUENTIAL TESTING

If sequential testing is indicated for this patient, list the order of testing here, or number the tests in the menu:

Initial Test: _____ if ☐ Positive/ ☐ Negative then:

Reflex to: _____ if ☐ Positive/ ☐ Negative then:

Reflex to: _____ if ☐ Positive/ ☐ Negative then:

Reflex to: _____

*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: _____

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:
- ☐ Please submit Exome requisition and consent documents available separately

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

Alagille Syndrome:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Alport Syndrome Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Angelman/Rett-Like Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Branchio-Oto-Renal Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Cancer (Germline) Panel Testing:

Comprehensive Hereditary Cancer Testing:
- ☐ Comprehensive Hereditary Cancer Panel
- ☐ Hereditary Brain Tumor Panel
- ☐ Hereditary Breast, Ovarian, and Endometrial Cancer Panel
- ☐ Hereditary High-Risk Breast Cancer Panel
- ☐ Hereditary Colorectal and Gastrointestinal Cancer Panel
- ☐ Hereditary High-Risk Colorectal Cancer Panel
- ☐ Hereditary Neuroendocrine Cancer Panel
- ☐ Hereditary Kidney Panel
- ☐ Hereditary Melanoma Panel
- ☐ Hereditary Pancreatic Cancer Panel
- ☐ Hereditary Paraganglioma/Pheochromocytoma Panel
- ☐ Hereditary Prostate Cancer Panel
- ☐ Reflex to Comprehensive Hereditary Cancer Panel

Panel Testing (cont'd)
For a current list of genes on each panel please see pages 5, 6, 7

Cancer (Germline) Panel Testing (cont'd):

Bone Marrow Failure/Myelodysplastic Syndrome/Leukemia Testing:
- ☐ Comprehensive Bone Marrow Failure (BMF)/Myelodysplastic Syndrome (MDS)/Leukemia Predisposition Panel
- ☐ Bone Marrow Failure Panel
- ☐ Fanconi Anemia/Rare Chromosomal Breakage Disorders Panel
- ☐ Hereditary Myelodysplastic Syndrome/Leukemia Predisposition Panel
- ☐ Inherited Neutropenia Panel
- ☐ Inherited Red Blood Cell Disorder Panel
- ☐ Inherited Thrombocytopenia Panel
- ☐ Telomere Disorder Panel
- ☐ Reflex to Comprehensive BMF/MDS/Leukemia Predisposition Panel

Cholestasis Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Congenital Diarrhea Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Connective Tissue Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Cornelia de Lange Syndrome Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Craniofacial Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Craniosynostosis Panel Testing:

Craniosynostosis Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

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

Epilepsy Panel Testing:

Epilepsy Panel:
- ☐ Targeted exome based sequence and copy number analysis of epilepsy related genes

Rapid Epilepsy Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Glycogen Storage Disease Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Hearing Loss Panel (AUDIOME): Hearing loss phenotype checklist must be completed on page 6
- ☐ Tier 1: DFNB1 locus testing (GJB2 sequence analysis and targeted ΔGJB6-D13S1830 testing)
- ☐ Tier 2: Targeted exome and aCGH based sequence and copy number analysis of hearing loss related genes
- ☐ Tier 1, Reflex to Tier 2 – If Tier 1 is inconclusive or negative, reflex to Tier 2

Humoral Dysfunction Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

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Panel Testing (cont'd)

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

Kabuki Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Ketotic Hypoglycemia Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Neonatal Respiratory Distress Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Noonan Spectrum Disorders Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Osteogenesis Imperfecta Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Pancreatitis Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Pneumothorax Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Primary Ciliary Dyskinesia Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Pulmonary Arterial Hypertension Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Pulmonary-Vascular Panel, Comprehensive:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Rubinstein-Taybi Syndrome Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Severe Combined Immunodeficiency (SCID) Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Stickler Syndrome Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication Analysis

Very Early Onset Inflammatory Bowel Disease:
- ☒ Targeted exome based sequence and copy number analysis of VEO-IBD related genes

Waardenburg Syndrome Panel:
- ☐ Sequence Analysis
- ☐ Deletion/Duplication 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
- ☐ 22q11.2 Deletion/Duplication Analysis, if normal reflex to microarray

Angelman Syndrome:
- ☐ Chromosme 15 Methylation Analysis

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

Blephrophemosis-Ptosis-Epicanthosis Syndrome:
- ☐ FOXL2 Sequence Analysis
- ☐ FOXL2 Deletion/Duplication Analysis

Brooke Spiegler Syndrome:
- ☐ CYLD Sequence Analysis

Single Gene and Region Specific Testing (cont'd)

CADASIL:
- ☐ NOTCH3 Sequence Analysis

CHARGE Syndrome:
- ☐ CHD7 Sequence Analysis
- ☐ CHD7 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 Analysis
- ☐ CFTR Deletion/Duplication Analysis

Fragile X Syndrome:
- ☐ Triplet Repeat Analysis

Gilbert's Syndrome:
- ☐ UGT1A1 Sequence Analysis of the promoter region

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 Analysis
- ☐ FBN1 Deletion/Duplication Analysis

Marshall Syndrome:
- ☐ COL11A1 Sequence Analysis
- ☐ COL11A1 Deletion/Duplication Analysis

Mental Retardation and Microcephaly with Pontine and Cerebellar Hypoplasia:
- ☐ CASK Sequence Analysis

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

Neuroblastoma:
- ☐ ALK Sequence Analysis
- ☐ PHOX2B Sequence Analysis (Not suitable for tumor samples)

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

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

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

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

Thalassemia:
- ☐ Alpha Thalassemia: HBA1 and HBA2 Deletion/Duplication Analysis
- ☐ Alpha Thalassemia: HBA1 and HBA2 Sequence Analysis
- ☐ Beta Thalassemia: HBB Sequence Analysis
- ☐ Beta Thalassemia: HBB Deletion/Duplication Analysis
- ☐ Hereditary Persistence of Fetal Hemoglobin: Beta Globin Gene Locus Deletion/Duplication Analysis
- ☐ Sickle Cell Anemia: HBB Sequence Analysis

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Single Gene and Region Specific Testing (cont'd)

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

- ☐ Known Variant Testing

Please fill out the known genetic abnormality section on page 2

PANEL TESTING: GENE CONTENT

For panels with both sequencing and deletion/duplication options: * Designates panels/genes with analysis by sequencing only; ** Designates genes with analysis by deletion/duplication only
For CPT codes and turn around times, please visit our website at https://www.testmenu.com/chop.

PANEL NAME	GENE CONTENT
Alagille Syndrome	*JAG1, NOTCH2*
Alport Syndrome Panel	*COL4A3, COL4A4, COL4A5*
Angelman/Rett-Like Panel	*ARX, ATRX, CDKL5, CNTNAP2*, DYRK1A, EHMT1, FOLR1, FOXG1, MBD5, MECP2, MEF2C, NRXN1*, OPHN1, PCDH19, PNKP, SLC9A6, TCF4, UBE3A, ZEB2*
Branchio-Oto-Renal Panel	*EYA1, SIX1, SIX5*
Cancer (Germline) Panel Testing: Comprehensive Hereditary Cancer Testing:	
Comprehensive Hereditary Cancer Panel	*ABCB11, ALK, APC, ARID5B, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CBL, CDC73, CDH1, CDK4, CDKN1C, CDKN1B, CDKN2A, CEBPA, CEBPE, CHEK2, COL7A1, CYLD, DDB2, DDX41, DICER1, DIS3L2, DKC1, DOCK8, EGFR, ELANE, EPCAM, ERCC2, ERCC3, ERCC4, ERCC5, ETV6, EXT1, EXT2, FAH, FANCA, FANCC, FANCG, FH, FLCN, G6PC3, GATA2, GBA, GJB2, GPC3, HABP2, HAX1, HFE, HMBS, HRAS, IKZF1, ITK, KIT, MAX, MEN1, MET, MLH1, MPL, MSH2, MSH6, MTAP, MUTYH, NBN, NF1, NF2, PALB2, PAX5, PDGFRA, PHOX2B, PMS2, POLD1, POLE, POLH, PRKAR1A, PRSS1, PTCH1, PTEN, PTPN11, RAD51, RAD51C, RAD51D, RB1, RECQL4, RET, RHBDF2, RMRP, RUNX1, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SERPINA1, SH2D1A, SLC25A13, SMAD4, SMARCA4, SMARCB1, SMARCE1, SOS1, SRY, STAT3, STK11, SUFU, TERT, TGFBR1, TMEM127, FAS, TP53, TRIM37, TSC1, TSC2, UROD, VHL, WAS, WRN, WT1, XPA, XPC*
Hereditary Brain Tumor Panel	*ALK, APC, ATM, MEN1, MLH1, MSH2, MSH6, PMS2, NBN, NF1 NF2, PALB2, PHOX2B, PTCH1, SMARCB1, SUFU, TP53, VHL*
Hereditary Breast, Ovarian, and Endometrial Cancer Panel	*ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, PMS2, FANCA, ITK, MEN1, MUTYH, NBN, NF1, PALB2, PTEN, RAD50, RAD51C, RAD51D, SMARCA4, STK11, TP53, WRN*
Hereditary High-Risk Breast Cancer Panel	*BRCA1, BRCA2, CDH1, PALB2, PTEN, STK11, TP53*
Hereditary Colorectal and Gastrointestinal Cancer Panel	*APC, ATM, BMPR1A, BRCA1, BRCA2, CDH1, CDK4, CDKN2A, CHEK2, EPCAM, MLH1, MSH2, MSH6, PMS2, KIT, MUTYH, PALB2, POLD1, POLE, PTEN, SMAD4, STK11, TP53*
Hereditary High-Risk Colorectal Cancer Panel	*APC, BMPR1A, EPCAM, MLH1, MSH2, MSH6, PMS2, MUTYH, PTEN, SMAD4, STK11, TP53*
Hereditary Endocrine Cancer Panel	*CDC73, CDKN1B, MAX, MEN1, PRKAR1A, PTEN, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, TP53, and VHL*
Hereditary Kidney Panel	*BAP1, CDC73, CDKN1C, FH, FLCN, GPC3, MET, MLH1, MSH2, MSH6, PTEN, SDHA, SDHB, SDHC, SDHD, SMARCB1, TSC1, TSC2, VHL, WT1*
Hereditary Melanoma Panel	*BAP1, BRCA1, BRCA2, CDK4, CDKNA2, PTEN, RB1, TERT, TP53, WRN*
Hereditary Pancreatic Cancer Panel	*APC, ATM, BMPR1A, BRCA1, BRCA2, CDK4, CDKNA2, EPCAM, MLH1, MSH2, MSH6, PMS2, PALB2, PRSS1, SMAD4, STK11, TP53, VHL*
Hereditary Paraganglioma/ Pheochromocytoma Panel	*FH, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL*
Hereditary Prostate Cancer Panel	*BRCA1, BRCA2, CHEK2, NBN, TP53*
Hereditary Cancer Panels: Bone Marrow Failure/Myelodysplastic Syndrome/Leukemia Testing:	
Comprehensive Bone Marrow Failure (BMF)/Myelodysplastic Syndrome (MDS)/ Leukemia Predisposition Panel	*AK2, ABCB7, ACD, ALAS2, ANKRD26, APC, ATM, ATR, BLM, BRCA1, BRCA2, C15ORF41, CBL, CDAN1, CDKN2A, CEBPA, CHEK2, CSF3R, CTC1,CXCR4, DDX11, DDX41, DKC1, ELANE, EPCAM, ERCC4, ESCO2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCJ, FANCL, FANCM, G6PC3, GATA1, GATA2, GFI1, GLRX5, HAX1, KIF23, KLF1, KRAS, LAMTOR2, LIG4, LYST, MLH1, MPL, MSH2, MSH6, NBN, NF1, NF2, NHEJ1, NHP2, NOP10, PALB2, PARN, PAX5, PMS2, PTPN11, RAB27A, RAC2, RAD51, RAD51C, RAF1, RBM8A, RECQL, RMRP, RPL11, RPL15, RPL19, RPL26, RPL27, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS27, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMDL9, SBDS, SH2B3, SEC23B, SLC25A38, SLC37A4, SLX4, SOS1, SRP72, TAZ, TERC, TERT, TINF2, TP53, USB1, VPS13B, VPS45, WAS, WIPF1, WRAP53*
Bone Marrow Failure Panel	*ABCB7, ACD, AK2, ALAS2, ANKRD26, ATM, ATR, BLM, BRCA1, BRCA2, C15ORF41, CBL, CDAN1, CSF3R, CTC1, CXCR4, DDX11, DKC1, ELANE, ERCC4, ESCO2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCJ, FANCL, FANCM, G6PC3, GATA1, GATA2, GFI1, GLRX5, HAX1, KIF23, KLF1, LAMTOR2, LYST, MPL, NBN, NHP2, NOP10, PALB2, PARN, RAB27A, RAC2, RAD51, RAD51C, RBM8A, RMRP, RPL11, RPL15, RPL19, RPL26, RPL27, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS27, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMDL9, SBDS, SEC23B, SLC25A38, SLC37A4, SLX4, SRP72, TAZ, TERC, TERT, TINF2, USB1, VPS13B, VPS45, WAS, WIPF1, WRAP53*
Fanconi Anemia/Rare Chromosomal Breakage Disorders Panel	*ATM, BLM, BRCA1 (FANCS), BRCA2 (FANCD1), DDX11, ERCC4 (FANCQ), ESCO2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG (XRCC9), FANCI, FANCJ (BRIP1), FANCL, FANCM, LIG4, NBN, NHEJ1, PALB2 (FANCN), RAD51 (FANCR), RAD51C (FANCO), SLX4 (FANCP)*

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PANEL TESTING: GENE CONTENT (cont'd)

For panels with both sequencing and deletion/duplication options: * Designates panels/genes with analysis by sequencing only; ** Designates genes with analysis by deletion/duplication only
For CPT codes and turn around times, please visit our website at https://www.testmenu.com/chop.

PANEL NAME	GENE CONTENT
Hereditary Myelodysplastic Syndrome/ Leukemia Predisposition Panel	*ACD, AK2, ANKRD26, APC, ATM, ATR, BLM, BRCA1, BRCA2, CBL, CDKN2A, CEBPA, CHEK2, CSF3R, CTC1, DDX11, DDX41, DKC1, EPCAM, ERCC4, ESCO2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCJ, FANCL, FANCM, GATA1, GATA2, KRAS, LIG4, MLH1, MPL, MSH2, MSH6, NBN, NF1, NF2, NHEJ1, NHP2, NOP10, PALB2, PARN, PAX5, PMS2, PTPN11, RAD51, RAD51C, RAF1, RECQL, RMRP, RTEL1, RUNX1, SAMD9, SAMDL9, SBDS, SH2B3, SLX4, SOS1, SRP72, TERC, TERT, TINF2, TP53, WAS, WIPF1, WRAP53*
Inherited Neutropenia Panel	*AP3B1, CSF3R, CXCR4, ELANE, G6PC3, GATA1, GATA2, GFI1, HAX1, LAMTOR2, LYST, RAB27A, RAC2, RMRP, SBDS, SLC37A4, TAZ, USB1, VPS13B, VPS45, WAS, WIPF1*
Inherited Red Blood Cell Disorder Panel	*ABCB7, ABCG5, ABCG8, AK1, ALAS2, ALDOA, ANK1, C15ORF41, CDAN1, EPB41, EPB42, G6PD, GATA1, GCLC, GLRX5, GPI, GPX1, GSR, GSS, HK1, KCNN4, KIF23, KLF1, NT5C3A, PFKM, PGK1, PIEZO1, PKLR, RHAG, RPL11, RPL15, RPL19, RPL26, RPL27, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS27, RPS29, RPS7, SEC23B, SLC25A38, SLC2A1, SLC4A1, SPTA1, SPTB, TPI1, XK*
Inherited Thrombocytopenia Panel	*ACTN1, ABCG5, ABCG8, ANKRD26 including 5'UTR, AP3B1, CYCS, DIAPH1, ETV6, FLI1, FLNA, GATA1, GFI1B, GP1BA, GP1BB, GP9, HOXA11, ITGA1, ITGA2B, MPL, MYH9, NBEAL2, PRKACG, RBM8A, RUNX1, SRC, SRP72, TPM4, TRPM7, TUBB1, WAS*
Telomere Disorder Panel	ACD, CTC1, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, WRAP53
Reflex to Comprehensive Bone Marrow Failure (BMF)/Myelodysplastic Syndrome (MDS)/Leukemia Predisposition Panel	*AK2, ABCB7, ACD, ALAS2, ANKRD26, APC, ATM, ATR, BLM, BRCA1, BRCA2, C15ORF41, CBL, CDAN1, CDKN2A, CEBPA, CHEK2, CSF3R, CTC1,CXCR4, DDX11, DDX41, DKC1, ELANE, EPCAM, ERCC4, ESCO2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCJ, FANCL, FANCM, G6PC3, GATA1, GATA2, GFI1, GLRX5, HAX1, KIF23, KLF1, KRAS, LAMTOR2, LIG4, LYST, MLH1, MPL, MSH2, MSH6, NBN, NF1, NF2, NHEJ1, NHP2, NOP10, PALB2, PARN, PAX5, PMS2, PTPN11, RAB27A, RAC2, RAD51, RAD51C, RAF1, RBM8A, RECQL, RMRP, RPL11, RPL15, RPL19, RPL26, RPL27, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS27, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMDL9, SBDS, SH2B3, SEC23B, SLC25A38, SLC37A4, SLX4, SOS1, SRP72, TAZ, TERC, TERT, TINF2, TP53, USB1, VPS13B, VPS45, WAS, WIPF1, WRAP53*
Cholestasis Panel	*ABCB11, ABCB4, ABCC2, AKR1D1, ATP8B1, BAAT, CFTR, CLDN1, HNF1A*, HNF1B*, HSD3B7, JAG1, LIPA, MYO5B, NOTCH2, NPC1, NPC2, NR1H4, SERPINA1, SLC25A13, TJP2, VIPAS39, VPS33B*
Congenital Diarrhea Panel	*APOB, CFTR, DGAT1, EPCAM, FLNA, GUCY2C, LCT, MTTP, MYO5B, NEUROG3, PCSK1, PNLIP, SAR1B, SI, SKIV2L, SLC26A3 (DRA), SLC5A1 (SGLT1), SLC9A3, SPINT2, STX3, TTC37*
Connective Tissue Panel	*ACTA2, B3GAT3, CBS, COL1A1, COL1A2, COL3A1, COL4A5, COL5A1, COL5A2, EFEMP2, ELN, 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, EDN1, EFNB1, EFTUD2, ERF, EVC, EVC2, EYA1, FGFR1*, FGFR2*, FGFR3*, GLI3, GNAI3*, IFT122, IFT43, MEGF8, MIR17HG**, MSX2, MYCN, PDE4D*, PLCB4, POLR1A, POLR1C, POLR1D, POR, PRKAR1, RAB23, RECQL4, RUNX2, SALL1, SF3B4, SIX1, SIX5, SKI, SNRPB, SPECC1L, TCF12, TCOF1, TWIST1, WDR19, WDR35, ZIC1, ZSWIM6*
Craniosynostosis Panel	*CD96, EFNB1, ERF, FGFR1*, FGFR2*, FGFR3*, IFT122, IFT43, MEGF8, MSX2, POR, RAB23, RECQL4, RUNX2, SKI, SPECC1L, TCF12, TWIST1, WDR35, ZIC1*
Epilepsy Panel Testing:	
Epilepsy Panel	*ALDH7A1, ALG13, ARHGEF9, ARX, ASAH1, ATP1A2, ATP1A3, CACNA1A, CACNA1D, CASK, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, CLN3, CLN5, CLN6, CLN8, CNKSR2, CSTB, CTSD, DEPDC5, DNM1, DYNC1H1, EEF1A2, EFHC1, EPM2A, FOLR1, FOXG1, GABRA1, GABRB3, GABRG2, GNAO1, GOSR2, GRIN1, GRIN2A, GRIN2B, HCN1, HDAC4, HNRNPU, IQSEC2, KCNA1, KCNA2, KCNB1, KCNC1, KCNJ10, KCNQ2, KCNQ3, KCNT1, KCTD7, LGI1, MECP2, MEF2C, MFSD8, NHLRC1, PCDH19, PIGA, PIGO, PIGT, PLCB1, PNKP, PNPO, POLG, PPT1, PRICKLE1, PRRT2, PURA, QARS, RELN, RYR3, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A8, SPTAN1, ST3GAL3, STX1B, STXBP1, SYN1, SYNGAP1, SZT2, TBC1D24, TPP1, TSC1, TSC2, WDR45, WWOX, UBE3A, ZEB2*
Rapid Epilepsy Panel	*ALDH7A1, CSTB, EPM2A, GAMT, GATM, GRIN2A, GRIN2B, GRIN2D*, KCNB1, KCNQ2, KCNT1*, NHLRC1, PNPO, POLG, SCN1A, SCN2A, SCN8A, SLC2A1, SLC6A8*
Glycogen Storage Disease Panel	*ACAT1, AGL, FBP1, G6PC, GAA, GBE1, GYS2, PCK1, PFKM, PHKA1, PHKA2, PHKB, PHKG2, PYGL, SLC16A1, SLC2A2, SLC37A4*
Hearing Loss Panel (AUDIOME v.2)	Tier 1: GJB2 sequence analysis and targeted GJB6 variant (ΔGJB6-D13S1830)
	Tier 2: ABHD12, ACTG1*, ADGRV1, ALMS1, ATP6V1B1, ATP6V1B2, BCS1L, BSND, CABP2, CACNA1D*, CCDC50, CDC14A*, CDH23, CEACAM16, CEP78*, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH*, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL9A1, COL9A2, COL9A3, DIABLO*, DIAPH1, DMXL2*, EDN3, EDNRB, EPS8, ESPN, ESRP1*, ESRRB, EYA1, EYA4, FGF3, GIPC3, GJB2, GJB6, GPSM2, GRHL2, GRXCR1, GRXCR2, GSDME (DFNA5), HARS, HARS2, HGF, HOMER2*, HSD17B4, ILDR1, KCNE1, KCNQ1, KCNQ4, LARS2, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MITF, MSRB3, MT-RNR1 (m.1555A>G), MT-TS1 (m.7445A>G), MYH14*, MYH9, MYO15A, MYO3A, MYO6, MYO7A, OPA1*, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2*, PAX3, PCDH15, PDZD7, PJVK (DFNB59), POU3F4, POU4F3, PRPS1, PTPRQ, RDX, RIPOR2* (FAM65B), S1PR2, SERPINB6, SIX1, SLC17A8, SLC26A4, SLC4A11, SLC52A2, SLC52A3, SLITRK6, SMPX, SNAI2, SOX10, STRC, SUCLA2, SYNE4, TBC1D24, TECTA, TIMM8A, TMC1, TMIE, TMPRSS3, TPRN, TRIOBP, TSPEAR, USH1C, USH1G, USH2A, WFS1, WHRN
Humoral Dysfunction Panel	*ICDA, BTK, CD27, CD40LG, CECR1, CR2, CTLA4, CXCR4, GATA2, ICOS, IKBKG*, IKZF1, IL21, IL21R, LRBA, NFKB1, NFKB2, PIK3CD*, PIK3R1, PRKCD, SH2D1A, STAT1, STAT3, UNG, XIAP*
Kabuki Syndrome Panel	*KDM6A, KMT2D*
Ketotic Hypoglycemia Panel	*ACAT1, AGL, G6PC, GYS2, PYGL, PHKA2, PHKB, PHKG2, SLC16A1, SLC37A4*
Neonatal Respiratory Distress Panel	*ABCA3, COPA, CSF2RA, CSF2RB, DKC1, FLNA, FOXF1, GATA2, JAG1, MARS, NKX2-1, NOTCH2, SFTPB, SFTPC*, SLC7A7, TERC, TERT*
Noonan Spectrum Disorders Panel*	*A2ML1, BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1*
Osteogenesis Imperfecta Panel	*COL1A1, COL1A2, IFITM5**

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PANEL TESTING: GENE CONTENT (cont'd)

For panels with both sequencing and deletion/duplication options: * Designates panels/genes with analysis by sequencing only; ** Designates genes with analysis by deletion/duplication only
For CPT codes and turn around times, please visit our website at https://www.testmenu.com/chop.

PANEL NAME	GENE CONTENT
Pancreatitis Panel	*CASR, CFTR, CTRC, PRSS1*, SPINK1*
Pneumothorax Panel	*COL3A1, FBN1, FLCN, TGFBR1, TGFBR2*
Primary Ciliary Dyskinesia Panel	*ARMC4, C21ORF59, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CFTR, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH6, DNAH8, DNAI1, DNAI2, DNAL1, DRC1 , DYX1C1, GAS8, HYDIN, INVS, LRRC6, MCIDAS, NME8, OFD1, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, ZMYND10*
Pulmonary Arterial Hypertension Panel	*ACVRL1, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, GDF2, KCNA5, KCNK3, RASA1, SMAD4, SMAD9, FOXF1*
Pulmonary-Vascular Panel, Comprehensive	*ABCA3, ACVRL1, ARMC4, BMPR1B, BMPR2, C21ORF59, CAV1, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CFTR, COPA, CSF2RA, CSF2RB, DKC1, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH6, DNAH8, DNAI1, DNAI2, DNAL1, DRC1, DYX1C1, EIF2AK4, ENG, FLNA, FOXF1, GAS8, GATA2, GDF2, HYDIN, INVS, JAG1, KCNA5, KCNK3, LRRC6, MARS, MCIDAS, NKX2.1, NME8, NOTCH2, OFD1, RASA1, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SFTPB, SFTPC*, SLC7A7, SMAD4, SMAD9, SPAG1, TERC, TERT, ZMYND10*
Rapid Epilepsy Panel	*ALDH7A1, CSTB, EPM2A, GAMT, GATM, GRIN2A, GRIN2B, GRIN2D*, KCNB1, KCNQ2, KCNT1*, NHLRC1, PNPO, POLG, SCN1A, SCN2A, SCN8A, SLC2A1, SLC6A8*
Rubinstein-Taybi Syndrome Panel	*CREBBP, EP300*
Severe Combined Immune Deficiency (SCID) Panel	*ADA, ATM, CHD7, DCLRE1C, DOCK8, FOXN1, IKBKG*, IL2RG, IL7R, JAK3, LIG4, NHEJ1, PNP, PRKDC, RAG1, RAG2, TBX1, WAS*
Stickler Syndrome Panel	*COL11A1, COL11A2, COL2A1, COL9A1, COL9A2*
Waardenburg Syndrome Panel	*EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10*

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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:

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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.

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

For 22q11.2 Deletion/Duplication Analysis, if normal reflex to array, we require 3-5mL of blood in an EDTA (purple top) tube **AND** 3-5mL in a Sodium Heparin (green top) tube.

Bone Marrow – For testing on a bone marrow aspirate, 3-5mL is needed in an EDTA (purple top) tube.

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 number to use for return shipment along with the return address.

Shipping Instructions

Samples should be shipped by overnight carrier to arrive Monday-Saturday.

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: (215) 590-4452

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. 11/18

LAST NAME _____ FIRST NAME _____

MR# _____ DOB _____

PLACE PATIENT LABEL HERE *OR* COMPLETE ABOVE

DO NOT HANDWRITE PATIENT INFORMATION HERE

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Billing Options (For Non-CHOP patients only)

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

For Lab use only: