

### **GENETICS AND GENOMICS DIAGNOSTIC LABORATORY**

For local courier service and/or inquiries, please contact 513-636-4474  $\cdot$  Fax: 513-636-4373 www.cincinnatichildrens.org/moleculargenetics  $\cdot$  Email: LabGeneticCounselors@cchmc.org

Mailing Address:
3333 Burnet Avenue, Room R1042
Cincinnati, OH 45229

## **HEMATOLOGY TEST REQUISITION**

All Information Must Be Completed Before Sample Can Be Processed

PATIENT INFORMATION	ON	ETHNIC/RACIAL B	ACKGROUND (Choose all that apply)
Address:  Home Phone:  MR# Date of Birth		□ European American (Wh □ Native American or Alas □ Pacific Islander □ Latino-Hispanic □ (specify country/region of □ Other □ (specify country/region of	Ashkenazi Jewish ancestry  f origin)
Sex Assigned at Birth:   Male  Female  Uncerta		PR	OVIDER INFORMATION
☐ REFERRING INSTITUTION		Physician Name (print):	
Institution:			
Address:			Fax: ( )
City/State/Zip:			
Accounts Payable Contact Name:		Genetic Counselor/Lab Co	ntact Name:
		Phone: ( )	Fax: ( )
Phone:Fax:		Email:	
Email:		Referring Physician Signat	Date:/
Linea.	SAMPLE/SPECIME		ine (including p)
Note: For post bone marrow transplant patients, we accept pre-transplant samples or Culturing of skin fibroblasts is done at an additional charge.  SPECIMEN TYPE: Amniotic fluid Blood Cytobrushes CVS Cord blood Bone marrow Tissue (specify): Specimen Date: // Time: Specimen Amount: DRAWN BY: *Phlebotomist must initial tube of specimen to confirm sample identity.		WE ARE UNABLE TO ACCEPT BLOOD SAMPLES COLLECTED WITHIN TWO WEEKS OF A TRANSFUSION. Please call before sending tissue samples.  • Single gene tests require at least 3mL whole blood in EDTA.  • Panels require at least 5 mL whole blood in EDTA.  • Hemoglobin Disorder tests require TWO tubes of whole blood in EDTA (preferably a 5mL and a 2mL, 7mL total). Ship refrigerated.	
	INDICATIONS/DIAGN	OSIS/ICD-10 CODE	
□ Abnormal bleeding □ Anemia □ Asymptomatic infant with abnormal newborn screen □ Carrier (Heterozygote) testing □ Concerns for: □HS, □HE, □HPP, □Stomatocytosis (HX & OHSt), □CDA □ Consideration to start treatment containing estrogens (such as OCPs) □ Diagnosis in symptomatic patient □ Easy bruising/spontaneous ecchymoses	☐ High Hemoglobin ☐ Hemolysis ☐ Iron overload ☐ Jaundice ☐ Microcytic anemia ☐ Microcytosis ☐ Persistently high platele evidence of inflammatic ☐ Platelet dysfunction/der ☐ Polycythemia/erythrocy	on) fect	Positive family history. Please specify relationship (e.g. cousin):  Prenatal testing (by previous arrangement only) Presymptomatic diagnosis of at-risk sibling Reticulocytosis Strong family history of thrombosis Unexplained Thrombocytopenia Unprovoked thrombosis Other:
	CLINICAL H	IISTORY	
☐ Hepatomegaly ☐ Skeletal abnormalities ☐ Other positive findings:	☐ Splenomgaly	If yes, date of bone marrov	e marrow transplant? ☐ Yes ☐ No v transplant

C	Cincinnati Children's changing the outcome together

Patient Name:	Date of Birth:

MEDICAL HISTORY	PEDIGREE OR FAMILY HISTORY
Clinic notes and laboratory data attachment, in lieu of writing in medical history, is also acceptable.  Patient's medical history:	Parental Consanguinity □ Y □ N
Date of last transfusion:  We are unable to accept blood samples collected within 2 weeks of a transfusion  Provisional Hb Diagnosis (for Hemoglobin Disorder tests):	
For Hemoglobin Disorders testing, please provide a copy of the following laboratory test results, as available: CBC, Hemoglobin electrophoresis, and iron studies.	
TEST(S) RE	QUESTED
Hemoglobin Disorders  Collect TWO tubes of whole blood: 5mL EDTA and 2mL EDTA. Ship refrigerated.  A hemoglobin electrophoresis is included as part of the Hemoglobin Disorders assays to aid the interpretation of genetic results. Charges may apply.  Alpha (HBA1/2) and Beta (HBB) Globin Gene Locus Analysis  HBA1 and HBA2 (α-globin) sequence analysis	Hemolytic Anemia  ☐ Hemolytic Anemia Panel (includes sequence analysis of ABCG5, ABCG8, AK1, ALAS2, ALDOA, ANK1, ATP11C, C15orf41, CDAN1, COL4A1, EPB41, EPB42, G6P, GATA1, GCLC, GPI, GPX1, GSR, GSS, GYPC, HK1, KCNN4, KIF23, KLF1, LPIN2, NT5C3A, PFKM, PGK1, PIEZO1, PKLR, RHAG, SEC23B, SLC2A1 (GLUT1), SLC4A SPTA1, SPTB, TPI1, XK)  ☐ Reflex to deletion/duplication of entire panel' ☐ Reflex to deletion/duplication of single gene(s) (specify):
<ul> <li>□ HBB (β-globin) sequence analysis</li> <li>□ HBB (β-globin) locus del/dup analysis (HBB, HBD, HBG1/2, &amp; HBE)</li> <li>□ HBD (Δ-globin) sequence analysis</li> <li>□ HPFH Gene Analysis</li> <li>□ HBG1 and HBG2 (γ-globin) sequence analysis</li> </ul>	□ Congenital Dyserythropoietic Anemia (CDA) Panel (includes sequence analysi of ALAS2, C15orf41, CDAN1, GATA1, KIF23, KLF1, LPIN2, SEC23B) □ Reflex to deletion/duplication of entire panel' □ Reflex to deletion/duplication of single gene(s) (specify):
	☐ Reflex to Hemolytic Anemia Panel reanalysis, if indicated
☐ HPFH SNP analysis (polymorphisms in <i>BCL11A</i> , <i>HBS1L-MYB</i> , & <i>KLF1</i> )  Thrombophilic Disorders	□ RBC Membrane Disorders Panel (includes sequence analysis of ABCG5, ABCG5 ANK1, ATP11C, COL4A1, EPB41, EPB42, GYPC, KCNN4, PIEZO1, RHAG, SLC2A1,
<ul> <li>□ Factor V (Leiden)</li> <li>□ Prothrombin (Factor II) G20210A genotype</li> <li>□ Thrombophilic polymorphism panel (Factor V- Leiden and Factor II-Prothrombin G20210A)</li> </ul>	SLC4A1, SPTA1, SPTB, XK)  Reflex to deletion/duplication of entire panel* Reflex to deletion/duplication of single gene(s) (specify):
Platelet Disorders	☐ Reflex to Hemolytic Anemia Panel reanalysis, if indicated
□ Platelet Disorders Gene Sequencing Panel Please include CBC with platelet count, mean platelet volume, family history of bleeding disorders, bleeding assessment tool (type) and score, von Willebrand testing, Platelet Function Analysis (PFA) results, platelet aggregation testing and mean platelet volume	□ RBC Enzymopathy Panel (includes sequence analysis of AK1, ALDOA, G6PD, GCLC, GPI, GPX1, GSR, GSS, HK1, NT5C3A, PFKM, PGK1, PKLR, TPI1) □ Reflex to deletion/duplication of entire panel <sup>†</sup> □ Reflex to deletion/duplication of single gene(s) (specify):
(MPV) & platelet distribution width (PDW) (if available) for comprehensive analysis (ABCG5, ABCG8, ACTB, ACTN1, ANKRD26, ANO6, AP3B1, AP3D1, ARPC1B, BLOC1S3, BLOC1S6, CDC42, CYCS, DIAPH1, DTNBP1, ETV6, FERMT3, FLI1,	Reflex to Hemolytic Anemia Panel reanalysis, if indicated  Erythrocytosis
FLNA, FYB1, GALE, GATA1, GF11B, GNE, GP1BA, GP1BB, GP6, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, IKZF5, ITGA2, ITGA2B, ITGB3, KDSR, LYST, MASTL, MECOM, MPIG6B, MPL, MYH9, NBEA, NBEAL2, ORAI1, P2RX1, P2RY1, P2RY12, PLA2G4A, PRKACG, PTGS1, PTPRJ, RASGRP2, RBM8A, RUNX1, SLFN14, SRC, STIM1, STX11, STXBP2, TBXA2R, TBXAS1, THPO, TPM4, TUBB1, UNC13D, VIPAS39, VPS33B, VPS45, WAS)	□ Erythrocytosis Gene Sequencing Panel (BHLHE41, BPGM, CALR, CYB5R3, EGLN1 (PHD2), EGLN2 (PHD1), EGLN3 (PHD3), EPAS1 (HIF2A), EPO, EPOR, GFI1B, HBA1, HBA2, HBB, HIF1A, HIF1AN (FIH), HIF3A, JAK2, KDM6A, MPL, OS9, PIEZO1, PKLR, SH2B3, SLC30A10, VHL, ZNF197)
$\square$ Reflex to deletion/duplication of entire panel $^{^{\dagger}}$ $\square$ Reflex to deletion/duplication of single gene(s) (specify):	☐ Reflex to deletion/duplication of entire panel <sup>†</sup> ☐ Reflex to deletion/duplication of single gene(s) (specify):
Reflex to Whole Exome Sequencing*	☐ Reflex to Whole Exome Sequencing*

\*Whole exome sequencing (WES) orders require completion of the WES Test Requisition. Also, inclusion of biological parental samples is strongly encouraged to assist with the analysis of WES and to increase test yield. Please visit our website at <a href="www.cincinnatichildrens.org/exome">www.cincinnatichildrens.org/exome</a> to obtain the required documents. WES testing will <a href="MOS">MOS</a> to be started until all forms are completed and received by the lab.



Patient Name:	Date of Birth:
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### **TEST(S) REQUESTED, CONTINUED**

# Thrombocytosis ☐ Thrombocytosis Gene Sequencing Panel (CALR, JAK2, MPL, THPO) ☐ Reflex to deletion/duplication of entire panel<sup>†</sup> ☐ Reflex to deletion/duplication of single gene(s) (specify):

<sup>†</sup>CGH Deletion/Duplication analysis of *ACTB, BHLHE41, CDC42, CYB5R3, EGLN2 (PHD1), EGLN3 (PHD3), FERMT3, GALE, GNE, GP6, GP9, HBA1, HBA2, HBB, HIF1A, HIF1AN (FIH), HIF3A, HPS5, HPS6, IKZF5, KDM6A, KDSR, MPIG6B, OS9, P2RY1, PTGS1, PTPRJ, SH2B3, SLC30A10, SRC, TPM4 and ZNF197* is not available at this time.

**Note:** Any gene in the panels listed may be ordered individually through custom gene sequencing.

Targeted (family specific) variant analysis for
gene If testing was <u>not</u> performed at CCHMC, please include proband's
report and at least 100ng of proband's DNA to use as a positive control.
Proband's name:
Proband's DOB:
Proband's variant:
Patient's relation to proband:
Patient's phenotype/test indication:

Please call 513-636-4474 to discuss any family-specific variant analysis with genetic counselor prior to shipment.

CUSTOM GENE SEQUENCING		
Gene(s) to be sequenced (specify):		
Only genes with clear published functional relationship to rare diseases are accepted.		
Suspected syndrome/ condition:		
Please choose one of the following:		
☐ Full gene(s) sequencing		
☐ Full gene(s) sequencing with reflex to deletion and duplication analysis, if indicated (please see list of genes available for del/dup at		
www.cincinnatichildrens.org/deldup)		
☐ Familial variant analysis		
Proband's name:		
Proband's variant:		
Patient's relation to proband:		
Patient's phenotype/test indication:		
If testing was $\underline{\text{not}}$ performed at Cincinnati Children's, please include proband's		
report and at least 100ng of proband's DNΔ to use as a positive control		

#### **DELETION AND DUPLICATION ASSAY**

Gene(s) to be analyzed (specify):
Please see list of available genes at: www.cincinnatichildrens.org/deldup

# Suspected syndrome/ condition: \_\_\_\_\_ Please choose one of the following:

- ☐ Deletion and duplication analysis of gene(s) specified above
- ☐ Deletion and duplication analysis of gene(s) specified above with reflex to sequencing, if indicated
- ☐ Analysis of gene(s) specified above from previously analyzed deletion and duplication
- $\square$  Familial deletion analysis

Proband's name: \_\_\_\_\_

Patient's phenotype/test indication:\_\_

Proband's DOB: \_\_\_\_

Proband's variant:

Patient's relation to proband: \_

If testing was  $\underline{not}$  performed at Cincinnati Children's, please include proband's

report and at least 100ng of proband's DNA to use as a positive control.

### **Deciding Which Hemoglobin Disorders Test to Order**

- If your patient's clinical symptoms and hematology testing do not suggest a specific diagnosis, order the Alpha (HBA1/2) and Beta (HBB) Globin Gene Locus Analysis. This includes PCR-based sequencing of HBA and HBB as well as deletion/duplication (del/dup) analysis by MLPA of both genes.
- If a common alpha thalassemia deletion is suspected, order the HBA1/HBA2 (alpha globin) deletion analysis.
  - If a suspected alpha globin deletion is not detected, order the HBA1/HBA2 (alpha globin) sequence analysis reflexively.
- If a structural alpha globin variant is suspected, order the HBA1/HBA2 (alpha globin) sequence analysis.
- If a structural beta globin defect or beta thalassemia mutation(s) are suspected, order the HBB (beta globin) sequence analysis.
- If a specific globin mutation has been identified in a family member, order Targeted (family specific) variant analysis (top left of this page).

  These tests detect only the specified mutation/deletion.