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GENETICS AND GENOMICS DIAGNOSTIC LABORATORY
For local courier service and/or inquiries, please contact 513-636-4474 • Fax: 513-636-4373
www.cincinnatichildrens.org/moleculargenetics • 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

Patient Name: _____ (Last), _____ (First), _____ (MI)

Address: _____

_____

Home Phone: _____

MR# _____ Date of Birth _____ / _____ / _____

Sex Assigned at Birth: ☐ Male ☐ Female ☐ Uncertain/Other:_____

## ETHNIC/RACIAL BACKGROUND (Choose All)

☐ European American (White)
☐ African-American (Black)
☐ Native American or Alaskan
☐ Asian-American
☐ Pacific Islander
☐ Ashkenazi Jewish ancestry
☐ Latino-Hispanic _____
(specify country/region of origin)
☐ Other _____
(specify country/region of origin)

## BILLING INFORMATION

☐ **REFERRING INSTITUTION**

Institution: _____

Address: _____

City/State/Zip: _____

Accounts Payable Contact Name: _____

Phone: _____

Fax: _____

Email: _____

## PROVIDER INFORMATION

Physician Name (print): _____

Address: _____

Phone: ( _____ ) _____ Fax: ( _____ ) _____

Email: _____

Genetic Counselor/Lab Contact Name: _____

Phone: ( _____ ) _____ Fax: ( _____ ) _____

Email: _____

_____ Date: ____/ _____/ ____

**Referring Physician Signature (REQUIRED)**

## SAMPLE/SPECIMEN INFORMATION

**Note:** For post bone marrow transplant patients, we accept pre-transplant samples or post-transplant skin fibroblasts **ONLY** (blood, saliva, and cytobrushes are not accepted). 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/DIAGNOSIS/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 platelet count (without evidence of inflammation)
☐ Platelet dysfunction/defect
☐ Polycythemia/erythrocytosis
☐ 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 HISTORY

☐ Hepatomegaly ☐ Skeletal abnormalities ☐ Splenomgaly
☐ Other positive findings: _____

**Has patient received a bone marrow transplant?** ☐ Yes ☐ No
If yes, date of bone marrow transplant _____
Percent engraftment _____

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Patient Name: _____ Date of Birth: _____

## MEDICAL HISTORY

**Clinic notes and laboratory data attachment, in lieu of writing in medical history, is also acceptable.**

Patient's medical history: _____

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.

## PEDIGREE OR FAMILY HISTORY

Parental Consanguinity ☐ Y ☐ N

## TEST(S) REQUESTED

### 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
  - ☐ *HBA1* and *HBA2* (α-globin) locus del/dup analysis *(HBA1/2 & HBZ)*
  - ☐ *HBB* (β-globin) sequence analysis
  - ☐ *HBB* (β-globin) locus del/dup analysis *(HBB, HBD, HBG1/2, & HBE)*
- ☐ *HBD* (Δ-globin) sequence analysis
- ☐ HPFH Gene Analysis
  - ☐ *HBG1* and *HBG2* (γ-globin) sequence analysis
  - ☐ *HBB* (β-globin) locus del/dup analysis *(HBB, HBD, HBG1/2, & HBE)*
  - ☐ HPFH SNP analysis (polymorphisms in *BCL11A, HBS1L-MYB, & KLF1*)

### Thrombophilic Disorders

- ☐ Factor V (Leiden)
- ☐ Prothrombin (Factor II) G20210A genotype
- ☐ Thrombophilic polymorphism panel (Factor V- Leiden and Factor II-Prothrombin G20210A)

### Platelet Disorders

- ☐ 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 (MPV) & platelet distribution width (PDW) (if available) for comprehensive analysis**

*(ABCG5, ABCG8, ACBD5, ACTB, ACTN1, ANKRD26, ANO6, AP3B1, AP3D1, ARPC1B, BLOC1S3, BLOC1S6, CDC42, CYCS, DIAPH1, DTNBP1, ETV6, FERMT3, FLI1, FLNA, FYB1, GALE, GATA1, GFI1B, 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, RAB27A, RASGRP2, RBM8A, RUNX1, SLFN14, SRC, STIM1, STX11, STXBP2, TBXA2R, TBXAS1, THPO, TPM4, TUBB1, UNC13D, VIPAS39, VPS33B, VPS45, WAS)*

- ☐ Reflex to deletion/duplication of entire panel†
- ☐ Reflex to deletion/duplication of single gene(s)† (specify): _____
- ☐ **Reflex to Whole Exome Sequencing***

### Hemolytic Anemia

- ☐ Hemolytic Anemia Panel (includes sequence analysis of *ABCG5, ABCG8, AK1, ALAS2, ALDOA, ANK1, ATP11C, C15orf41, CDAN1, COL4A1, EPB41, EPB42, G6PD, GATA1, GCLC, GPI, GPX1, GSR, GSS, GYPC, HK1, KCNN4, KIF23, KLF1, LPIN2, NT5C3A, PFKM, PGK1, PIEZO1, PKLR, RHAG, SEC23B, SLC2A1 (GLUT1), SLC4A1, SPTA1, SPTB, TPI1, XK)*
  - ☐ Reflex to deletion/duplication of entire panel†
  - ☐ Reflex to deletion/duplication of single gene(s) (specify): _____
- ☐ Congenital Dyserythropoietic Anemia (CDA) Panel (includes sequence analysis 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
- ☐ RBC Membrane Disorders Panel (includes sequence analysis of *ABCG5, ABCG8, ANK1, ATP11C, COL4A1, EPB41, EPB42, GYPC, KCNN4, PIEZO1, RHAG, SLC2A1, SLC4A1, SPTA1, SPTB, XK)*
  - ☐ Reflex to deletion/duplication of entire panel†
  - ☐ Reflex to deletion/duplication of single gene(s) (specify): _____
  - ☐ Reflex to Hemolytic Anemia Panel reanalysis, if indicated
- ☐ 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†
  - ☐ Reflex to deletion/duplication of single gene(s) (specify): _____
  - ☐ Reflex to Hemolytic Anemia Panel reanalysis, if indicated

### Erythrocytosis

- ☐ 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 )*

- ☐ Reflex to deletion/duplication of entire panel†
- ☐ Reflex to deletion/duplication of single gene(s) (specify): _____
- ☐ **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 www.cincinnatichildrens.org/exome to obtain the required documents. WES testing will **NOT** be started until all forms are completed and received by the lab.

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Patient Name: _____ Date of Birth: _____

## TEST(S) REQUESTED, CONTINUED

**Thrombocytosis**

☐ Thrombocytosis Gene Sequencing Panel
*(CALR, JAK2, MPL, THPO)*
- ☐ Reflex to deletion/duplication of entire panel†
- ☐ Reflex to deletion/duplication of single gene(s) (specify): _____

_____

†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 **not** 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 DOB: _____

Proband's variant: _____

Patient's relation to proband: _____

Patient's phenotype/test indication: _____

**If testing was 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.**

## 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

☐ Familial deletion analysis

Proband's name: _____

Proband's DOB: _____

Proband's variant: _____

Patient's relation to proband: _____

Patient's phenotype/test indication: _____

**If testing was 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.