

# GENETIC TESTING: HEMATOLOGIC CONDITIONS (NON-CANCEROUS)

## OVERVIEW

Genetic testing for hematologic (non-cancerous) conditions may be used to confirm a diagnosis in a patient who has signs and/or symptoms of a specific hematologic condition. Confirming the diagnosis may alter aspects of management and may eliminate the need for further diagnostic workup. This document addresses genetic testing for common hematologic (non-cancerous) conditions.

## POLICY REFERENCE TABLE

Below is a list of higher volume tests and the associated laboratories for each coverage criteria section. This list is not all inclusive.

<a href="#">Coverage Criteria Sections</a>	Example Tests (Labs)	Common CPT Codes	Common ICD Codes	<a href="#">Ref</a>
<b><a href="#">Known Familial Variant Analysis for Hematologic Conditions (non-cancerous)</a></b>				
<a href="#">Known Familial Variant Analysis for Hematologic Conditions (non-cancerous)</a>	Targeted Mutation Analysis for a Known Familial Variant	81403, 81258, 81362		11
<b><a href="#">Inherited Thrombophilia</a></b>				
<a href="#">Factor V Leiden (F5) and Prothrombin (F2) Variant Analysis for Inherited Thrombophilia</a>	Factor V (Leiden) Mutation Analysis (Quest Diagnostics)	81241	D68.51, D68.2, D68.59, R79.1, Z86.2, I82.90	1, 5
	Prothrombin (Factor II) 20210G→ A Mutation Analysis (Quest Diagnostics)	81240	D68.52, D68.2,	

			D68.59, R79.1, Z86.2, I82.90	
<b>Hemoglobinopathies</b>				
<a href="#">HBA1/HBA2 and/or HBB Variant Analysis</a>	HBA1 Deletion/Duplication Analysis	81257, 81259, 81269, S3845,	D56.0, D56.9, D53.9, R70.1,	2, 3, 4, 6
	HBA2 Deletion/Duplication Analysis	S3850	D56.3, D56.8, Z86.2	
	HBA1 Sequencing Analysis			
	HBA2 Sequencing Analysis			
	HBB Sequencing Analysis	81361, 81363,	D57.00-	
	HBB Deletion/Duplication Analysis	81364, S3846	D57.819, D56.1, D64.9	
<b>Hemophilia</b>				
<a href="#">F8 and/or F9 Variant Analysis</a>	F8 Deletion/Duplication Analysis	81403, 81406,	D66, D67,	8, 9
	F8 Sequencing Analysis	81407	I62.9, M25,	
	F9 Deletion/Duplication Analysis	81238, 81479	N92.2, R04.0,	
	F9 Sequencing Analysis		R31	
<b>Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency</b>				
<a href="#">G6PD Variant Analysis</a>	G6PD Targeted Mutation Analysis	81247, 81248,	D55.0	7
	G6PD Sequencing Analysis	81249		
<b>von Willebrand Disease</b>				
<a href="#">GP1BA and/or VWF Variant Analysis</a>	GP1BA Sequencing Analysis	81401, 81403,	D68.0	10
	VWF Targeted Mutation Analysis	81404, 81405,		
	VWF Sequencing Analysis	81406, 81408, 81479		
<b>Other Covered Hematologic Conditions (non-cancerous)</b>				
<a href="#">Other Covered Hematologic Conditions (non-cancerous)</a>	See list below	81400-81408		12, 13, 14

## OTHER RELATED POLICIES

This policy document provides coverage criteria for Genetic Testing for Hematologic Conditions (Non-Cancerous). Please refer to:

- **Oncology: Molecular Analysis of Solid Tumors and Hematologic Malignancies** for coverage criteria related to exome and genome sequencing of solid tumors and hematologic malignancies.
- **Genetic Testing: Prenatal and Preconception Carrier Screening** for coverage criteria related to carrier screening in the prenatal, preimplantation, and preconception setting.
- **Genetic Testing: Prenatal Diagnosis (via amniocentesis, CVS, or PUBS) and Pregnancy Loss** for coverage related to prenatal and pregnancy loss diagnostic genetic testing for tests intended to diagnose genetic conditions following amniocentesis, chorionic villus sampling or pregnancy loss.
- **Genetic Testing: Multisystem Inherited Disorders, Intellectual Disability, and Developmental Delay** for coverage criteria related to diagnostic genetic testing for conditions affecting multiple organ systems.
- **Genetic Testing: Metabolic, Endocrine, and Mitochondrial Disorders** for coverage criteria related to genetic testing for *MTHFR*.
- **Genetic Testing: General Approach to Genetic Testing** for coverage criteria related to genetic testing for non-cancerous hematologic disorders that are not specifically discussed in this or another non-general policy.

## COVERAGE CRITERIA

### KNOWN FAMILIAL VARIANT ANALYSIS FOR HEMATOLOGIC CONDITIONS (NON-CANCEROUS)

- I. Targeted mutation analysis for a known familial variant (81403, 81258, 81362) for a non-cancerous hematologic condition is considered **medically necessary** when:

- A. The member has a [close relative](#) with a known pathogenic or likely pathogenic variant causing the condition.
- II. Targeted mutation analysis for a known familial variant (81403, 81258, 81362) for a non-cancerous hematologic condition is considered **investigational** for all other indications.

[back to top](#)

## INHERITED THROMBOPHILIA

### Factor V Leiden (*F5*) and Prothrombin (*F2*) Variant Analysis for Inherited Thrombophilia

- I. *F5* (81241) and *F2* (81240) variant analysis to confirm or establish a diagnosis of an inherited thrombophilia may be considered **medically necessary** when:
  - A. The member meets at least one of the following:
    - 1. A first unprovoked venous thromboembolism (VTE) younger than 50 years old, **OR**
    - 2. VTE at unusual sites (such as hepatic portal, mesenteric, and cerebral veins), **OR**
    - 3. Recurrent VTE, **OR**
    - 4. Personal history of VTE with at least one of the following:
      - a) Two or more family members with a history of VTE, **OR**
      - b) One [first-degree relative](#) with VTE at a young age, **OR**
    - 5. Low activated protein C (APC) resistance activity, **OR**
    - 6. The member is a female under the age of 50 who smokes tobacco and has a history of acute myocardial infarction, **OR**
    - 7. A [first-degree relative](#) known to be homozygous for factor V Leiden or factor II c.\*97G>A, **OR**

8. The member is an asymptomatic pregnant female or female contemplating pregnancy, with a [first-degree relative](#) with unprovoked VTE or VTE provoked by pregnancy or contraceptive use, **OR**
  9. The member is a pregnant female or female contemplating pregnancy or estrogen use who has a [first-degree relative](#) with both of the following:
    - a) A history of VTE, **AND**
    - b) The member is a known carrier for factor V Leiden and/or factor II c.97\*G>A variant, **OR**
  10. The member is a pregnant female or female contemplating pregnancy with a previous non-estrogen-related VTE or VTE provoked by a minor risk factor.
- II. *F5* (81241) and *F2* (81240) variant analysis to confirm or establish a diagnosis of an inherited thrombophilia is considered **investigational** for all other indications, including:
- A. Fetal loss or adverse pregnancy outcomes (e.g., placental abruption, fetal growth restriction, or preeclampsia).

[back to top](#)

## HEMOGLOBINOPATHIES

### ***HBA1/HBA2* and/or *HBB* Variant Analysis**

- I. *HBA1/HBA2* variant analysis (81257, 81259, 81269, S3845, S3850), and/or *HBB* (81361, 81363, 81364, S3846) to confirm or establish a diagnosis of a hemoglobinopathy (alpha-thalassemia, beta-thalassemia, or sickle cell disease) is considered **medically necessary** when:
  - A. The member's hematologic screening results (examples: MCV, MCH, CBC, hemoglobin electrophoresis, or dichlorophenol indophenol (DCIP)) are positive for a hemoglobinopathy, **OR**

- B. The member's hematologic screening results (examples: MCV, MCH, CBC, hemoglobin electrophoresis, or dichlorophenol indophenol (DCIP)) do not conclusively diagnose or rule out a hemoglobinopathy.
- II. *HBA1/HBA2* variant analysis (81257, 81259, 81269, S3845, S3850), and/or *HBB* (81361, 81363, 81364, S3846) to confirm or establish a diagnosis of a hemoglobinopathy (alpha-thalassemia, beta-thalassemia, or sickle cell disease) is considered **investigational** for all other indications.

[back to top](#)

## HEMOPHILIA

### ***F8* and/or *F9* Variant Analysis**

- I. *F8* variant analysis (81403, 81406, 81407) and/or *F9* variant analysis (81238, 81479) to confirm or establish a diagnosis of hemophilia A or B is considered **medically necessary** when:
  - A. The member has any of the following clinical features of hemophilia:
    1. Hemarthrosis (especially with mild or no antecedent trauma), **OR**
    2. Deep-muscle hematomas, **OR**
    3. Intracranial bleeding in the absence of major trauma, **OR**
    4. Neonatal cephalohematoma or intracranial bleeding, **OR**
    5. Prolonged oozing or renewed bleeding after initial bleeding stops following tooth extractions, mouth injury, or circumcision, **OR**
    6. Prolonged, delayed bleeding, or poor wound healing following surgery or trauma, **OR**
    7. Unexplained GI bleeding or hematuria, **OR**
    8. Heavy or prolonged menstrual bleeding (especially with onset at menarche), **OR**
    9. Prolonged nosebleeds, especially recurrent and bilateral, **OR**

10. Excessive bruising (especially with firm, subcutaneous hematomas),  
**OR**

B. The following laboratory features:

1. Normal platelet count, **AND**
2. Prolonged activated partial thromboplastin time (aPTT), **AND**
3. Normal prothrombin time (PT).

C. *F8* variant analysis (81403, 81406, 81407) and/or *F9* variant analysis (81238, 81479) to confirm or establish a diagnosis of hemophilia A or B is considered **investigational** for all other indications.

[back to top](#)

## GLUCOSE-6-PHOSPHATE DEHYDROGENASE (G6PD) DEFICIENCY

### **G6PD Variant Analysis**

- I. *G6PD* variant analysis (81247, 81248, 81249) to confirm or establish a diagnosis\* of glucose-6-phosphate dehydrogenase deficiency is considered **investigational**.

\* Diagnosis of *G6PD* can be achieved by quantitative spectrophotometric analysis or, more commonly, by a rapid fluorescent spot test detecting the generation of NADPH from NADP.

[back to top](#)

## VON-WILLEBRAND DISEASE

### **GP1BA and VWF Variant Analysis**

- I. *GP1BA* and/or *VWF* variant analysis (81401, 81403, 81404, 81405, 81406, 81408, 81479) to confirm or establish a diagnosis\* of von-Willebrand disease is considered **investigational**.

\* Diagnosis of von-Willebrand disease can be achieved by standard laboratory and biochemical testing.

[back to top](#)

## OTHER COVERED HEMATOLOGIC CONDITIONS (NON-CANCEROUS)

The following is a list of conditions that have a known genetic association. Due to their relative rareness, it may be appropriate to cover these genetic tests to establish or confirm a diagnosis.

- I. Genetic testing to establish or confirm one of the following hematologic conditions (non-cancerous) to guide management is considered **medically necessary** when the member demonstrates clinical features\* consistent with the disorder (the list is not meant to be comprehensive, see II below):
  - A. [Atypical Hemolytic-Uremic Syndrome \(aHUS\)](#)
  - B. [Complete Plasminogen Activator Inhibitor 1 Deficiency \(PAI-1\)](#)
  - C. [Diamond-Blackfan Anemia \(DBA\)](#)
  - D. [Hereditary Spherocytosis](#)
  - E. Factor VII Deficiency
  - F. Factor X Deficiency
  - G. Factor XI Deficiency (Hemophilia C)
  - H. Factor XII Deficiency
  - I. Factor XIII Deficiency
  
- II. Genetic testing to establish or confirm the diagnosis of all other non-cancerous hematologic conditions not specifically discussed within this or another medical policy will be evaluated by the criteria outlined in *General Approach to Genetic Testing* (see policy for coverage criteria).

\*Clinical features for a specific disorder may be outlined in resources such as [GeneReviews](#), [OMIM](#), [National Library of Medicine](#), [Genetics Home Reference](#), or other scholarly source.

[back to top](#)



## NOTES AND DEFINITIONS

1. Close relatives include first, second, and third degree blood relatives on the same side of the family:
  - a. **First-degree relatives** are parents, siblings, and children
  - b. **Second-degree relatives** are grandparents, aunts, uncles, nieces, nephews, grandchildren, and half siblings
  - c. **Third-degree relatives** are great grandparents, great aunts, great uncles, great grandchildren, and first cousins

[back to top](#)

## BACKGROUND AND RATIONALE

### Known Familial Variant Analysis for Hematologic Conditions (non-cancerous)

#### *Genetic Support Foundation*

The Genetic Support Foundation's Genetics 101 information on inheritance patterns says the following about testing for familial pathogenic variants:

Genetic testing for someone who may be at risk for an inherited disease is always easier if we know the specific genetic cause. Oftentimes, the best way to find the genetic cause is to start by testing someone in the family who is known or strongly suspected to have the disease. If their testing is positive, then we can say that we have found the familial pathogenic (harmful) variant. We can use this as a marker to test other members of the family to see who is also at risk.

### Factor V Leiden (F5) and Prothrombin (F2) Variant Analysis for Inherited Thrombophilia

#### *American College of Medical Genetics and Genomics (ACMG)*

The American College of Medical Genetics and Genomics (Zhang, 2018) published updated technical standards for genetic testing for variants associated with VTE, with a focus on

factor V Leiden and factor II. Testing is recommended for factor V Leiden and factor II c.\*97G>A for the following indications:

- 1.) A first unprovoked VTE, especially <50 years old
- 2.) VTE at unusual sites (such as hepatic portal, mesenteric, and cerebral veins)
- 3.) Recurrent VTE
- 4.) Personal history of VTE with (a) two or more family members with a history of VTE or (b) one first-degree relative with VTE at a young age
- 5.) Patients with low activated protein C (APC) resistance activity (p. 1492)

In addition, this testing “may be considered” for the following indications:

- 1.) Females under the age of 50 who smoke tobacco and have a history of acute myocardial infarction
- 2.) Siblings of individuals known to be homozygous for factor V Leiden or factor II c.\*97G>A, because they have a 1 in 4 chance of being a homozygote
- 3.) Asymptomatic pregnant female or female contemplating pregnancy, with a first-degree relative with unprovoked VTE or VTE provoked by pregnancy or contraceptive use
- 4.) Pregnant female or female contemplating pregnancy or estrogen use who has a first-degree relative with a history of VTE and is a known carrier for factor V Leiden and/or factor II c.\*97G>A variant
- 5.) Pregnant female or female contemplating pregnancy with a previous non-estrogen-related VTE or VTE provoked by a minor risk factor, because knowledge of the factor V Leiden or factor II c.\*97G>A status may alter pregnancy-related thrombophylaxis (p. 1492-1493)

*American College of Obstetricians and Gynecologists (ACOG)*

ACOG also published Practice Bulletin 197 (2018) on Inherited Thrombophilias in Pregnancy which states that “...screening for inherited thrombophilias is not recommended for women with a history of fetal loss or adverse pregnancy outcomes including abruptio, preeclampsia, or fetal growth restriction because there is insufficient clinical evidence that antepartum prophylaxis with unfractionated heparin or low-molecular-weight-heparin prevents recurrence in these patients, and a causal association has not been established.” (p. e23).

## **Hemoglobinopathies - *HBA1/HBA2* and/or *HBB* Variant Analysis**

GeneReviews is an expert-authored review of current literature on a genetic disease, and goes through a rigorous editing and peer review process before being published online. The recommended hemoglobinopathy evaluation testing for Alpha-Thalassemia, Beta-Thalassemia, and Sickle Cell Disease is as follows:

*GeneReviews: Alpha-Thalassemia*

Hemoglobin Bart hydrops fetalis (Hb Bart) syndrome, which is caused by deletion or inactivation of all four alpha globin genes, exhibits the following hematologic findings: severe macrocytic hypochromic anemia (in the absence of ABO or Rh blood group incompatibility), reticulocytosis (may be >60%), and peripheral blood smear with large, hypochromic red cells, severe anisopoikilocytosis, and numerous nucleated red cells. In addition, hemoglobin analysis will typically display decreased amounts or complete absence of hemoglobin A and increased amounts of Hb Bart.

Hemoglobin H disease (HbH disease), which is caused by deletion or inactivation of three alpha globin genes, exhibits the following hematologic findings: mild-to-moderate (rarely severe) microcytic hypochromic hemolytic anemia, moderate reticulocytosis (3%-6%), Peripheral blood smear with anisopoikilocytosis, and very rarely nucleated red blood cells, Red blood cell supravital stain showing HbH inclusions ( $\beta_4$  tetramers) in 5%-80% of erythrocytes following incubation of fresh blood smears with 1% brilliant cresyl blue for one to three hours. In addition, hemoglobin analysis will typically display the presence of 0.8%-40% HbH and 60%-90% hemoglobin A.

*GeneReviews: Beta-Thalassemia*

Beta-Thalassemia typically displays the following hematologic findings of microcytic hypochromic anemia, anisopoikilocytosis with nucleated red blood cells on peripheral blood smear, and hemoglobin analysis that reveals decreased amounts or complete absence of hemoglobin A and increased amounts of hemoglobin F.

*GeneReviews: Sickle Cell Disease*

Laboratory features of sickle cell disease include: normocytic anemia; sickle cells, nucleated red blood cells, target cells, and other abnormal red blood cells on peripheral blood smear; Howell-Jolly bodies indicate hyposplenism; presence of hemoglobin S (HbS) on a hemoglobin assay (e.g., high-performance liquid chromatography [HPLC], isoelectric focusing, cellulose acetate electrophoresis, citrate agar electrophoresis) with an absence or diminished amount of HbA.

*Viprakasit V, Ekwattanakit S. Clinical classification, screening and diagnosis for thalassemia*

Viprakasit and Ekwattanakit (2018) published a clinical classification, screening and diagnosis for thalassemia article that states:

“In general, these mutation analyses would be critical for the confirmation of thalassemia diagnoses in only a few selected cases for whom the basic hematology and Hb analysis described could not provide a conclusive diagnosis. However, these molecular analyses would be indispensable in a program for the prevention and control of thalassemia syndromes because the mutation data would be required for genetic counseling, genetic risk calculation in the offspring, and prenatal and preimplantation genetic diagnosis. In addition, DNA analysis could help in predicting the clinical severity and guiding clinical management; milder b-globin mutations (b1-thal) usually are associated with milder phenotypes, as has been shown in HbE/b-thalassemia.” (p. 207)

### **Hemophilia - F8 and/or F9 Variant Analysis**

GeneReviews is an expert-authored review of current literature on a genetic disease, and goes through a rigorous editing and peer review process before being published online. The recommended hemoglobinopathy evaluation testing for Hemophilia A and Hemophilia B is as follows:

*GeneReviews: Hemophilia A and Hemophilia B*

Individuals with Hemophilia A (factor VIII deficiency) or Hemophilia B (factor IX deficiency) can exhibit the following clinical symptoms:

- Hemarthrosis, especially with mild or no antecedent trauma
- Deep-muscle hematomas
- Intracranial bleeding in the absence of major trauma
- Neonatal cephalohematoma or intracranial bleeding
- Prolonged oozing or renewed bleeding after initial bleeding stops following tooth extractions, mouth injury, or circumcision
- Prolonged or delayed bleeding or poor wound healing following surgery or trauma
- Unexplained GI bleeding or hematuria
- Menorrhagia, especially with onset at menarche

- Prolonged nosebleeds, especially recurrent and bilateral
- Excessive bruising, especially with firm, subcutaneous hematomas

The following are laboratory findings in individuals with Hemophilia A or Hemophilia B:

- Normal platelet count
- Prolonged activated partial thromboplastin time (aPTT)
- Normal prothrombin time (PT)

### **Glucose6-Phosphate Dehydrogenase Deficiency - G6PD Variant Analysis**

*American Academy of Family Physicians*

Frank (2005) published guidelines in *American Family Physician* for evaluating individuals for *G6PD* deficiency, including specific laboratory tests which notably do not include genetic testing: “The diagnosis of *G6PD* deficiency is made by a quantitative spectrophotometric analysis or, more commonly, by a rapid fluorescent spot test detecting the generation of NADPH from NADP. The test is positive if the blood spot fails to fluoresce under ultraviolet light.” (p. 1278)”

### **von Willebrand Disease - *GP1BA* and/or *VWF* Variant Analysis**

*Centers for Disease Prevention and Control (CDC), via the National Heart Lung and Blood Institute, National Institutes of Health (NHLBI-NIH)*

Guidelines for diagnosis and management of von Willebrand disease (VWD) were developed for practicing primary care and specialist clinicians—including family physicians, internists, obstetrician-gynecologists, pediatricians, and nurse-practitioners—as well as hematologists and laboratory medicine specialists, which included recommendations for laboratory tests to aid in the diagnosis of VWD, which notably do not include genetic testing.

[back to top](#)

## REFERENCES

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[back to top](#)