



Hematology Genetics Testing

**Expertise and experience
you can trust.**

Your hematology experts

BloodCenter of Wisconsin is a trusted laboratory partner and recognized leader in hematology, with more than 70 years of unparalleled experience across the continuum of care of patients with hematologic disorders. We provide premier service through integration of functional diagnostics and disease focused genetic sequencing, clearly reported to guide superior patient care.

Quick, Targeted & Tailored Testing

- Guided panel testing, customizable test panels and single gene analysis mean that you can request panels tailored to your patient's needs.
- Control costs through expert-guided test selection and utilization practices.
- Comprehensive testing menu streamlines your ordering process – ensuring the best care for your patients and their families.

Clear & Actionable Reporting

- Each report is easy to understand, and is driven by patient-specific bioinformatics analysis.
- You receive the most precise analysis through BloodCenter's multidisciplinary team approach to review and interpretation of results.
- Quality is our #1 goal – We rigorously classify variants according to ACMG guidelines.

Consultative Support & Service

- Our team of experts is on hand for clinical consulting and interpretation of your test results.
- We'll improve cost effectiveness by helping to determine which tests you should order for your patients, and which may be unnecessary.
- Take advantage of our superior customer service and support team.



Panel, Test and Code	
Panel	Genes Tested
aHUS Genetic Evaluation	<i>CFH, CFI, MCP(CD46), THBD, C4BPA, C4BPB, CFB, C3, LMNA, DGKE, ADAMTS13, CFHR1, CFHR3, CFHR4, CFHR5</i>
Coagulation Disorder Panel	<i>F2, F5, F7, F8, F9, F10, F11, F13A1, F13B, FGA, FGB, FGG, GGCX, LMAN1, MCFD2, SERPINA1, SERPINE1, SERPINF2, VKORC1, VWF</i>
Comprehensive Bleeding Disorder Panel	<i>ANO6, AP3B1, BLOC1S3, BLOC1S6(HPS9), DTNBP1(HPS7), F2, F5, F7, F8, F9, F10, F11, F13A1, F13B, FERMT3, FGA, FGB, FGG, FLI1, GGCX, GFI1B, GP1BA, GP1BB, GP6, GP9, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2B, ITGB3, LMAN1, LYST, MCFD2, NBEAL2, P2RY12, PLA2G4A, PLAU, PRKACG, RASGRP2, RUNX1, SERPINA1, SERPINE1(PAI1), SERPINF2, STIM1, TBXA2R, VIPAS39, VKORC1, VPS33B, VWF</i>
Comprehensive Platelet Disorder Panel	<i>ACTN1, ANKRD26, ANO6, AP3B1, BLOC1S3, BLOC1S6(HPS9), CYCS, DTNBP1(HPS7), ETV6, FERMT3, FLI1, GATA1, GFI1B, GP1BA, GPIBB, GP6, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2B, ITGB3, LYST, MASTL, MPL, MYH9, NBEAL2, P2RY12, PLA2G4A, PLAU, PRKACG, RASGRP2, RBM8A, RUNX1, STIM1, STXBP2, TBXA2R, TUBB1, VIPAS39, VPS33B, WAS</i>
Congenital Neutropenia Panel	<i>AP3B1, C16ORF57, CSF3R, CXCR4, ELANE, G6PC3, GATA1, GATA2, GFI1, HAX1, JAGN1, LAMTOR2, LYST, RAB27A, RAC2, SBDS, SLC37A4, TAZ, TCIRG1, VPS13B, VPS45, WAS, WIPF1</i>
Inherited Thrombocytopenia Panel	<i>ACTN1, ANKRD26, CYCS, ETV6, FLI1, GATA1, GP1BA, GP1BB, GP9, HOXA11, ITGA2B, ITGB3, MASTL, MPL, MYH9, NBEAL2, PRKACG, RBM8A, RUNX1, STXBP2, TUBB1, WAS</i>
Platelet Function Disorder Panel	<i>ANO6, AP3B1, BLOC1S3, BLOC1S6(HPS9), DTNBP1(HPS7), FERMT3, FLI1, GFI1B, GP1BA, GP1BB, GP6, GP9, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2B, ITGB3, LYST, NBEAL2, P2RY12, PLA2G4A, PLAU, PRKACG, RASGRP2, RUNX1, STIM1, TBXA2R, VIPAS39, VPS33B</i>
Thrombosis Panel	<i>ADAMTS13, F2, F5, FGA, FGB, FGG, HRG, KNG1, PLAT, PROC, PROS1, SERPINC1, THBD</i>
Custom Blood Disorder Panel	Select any genes from established panels. Two gene minimum, 10 gene maximum. If greater than 10 genes desired, please call 800-245-3117 ext 6250

Indications for testing

Specifically Designed Panels:

- Clarification and/or confirmation of diagnosis in a patient with clinical and laboratory findings of a hematologic disorder when patient's history suggests multiple disorders
- Identification of carriers with family history of unspecified bleeding disorders to provide accurate reproductive risk assessment and genetic counseling

Single gene sequencing or custom gene panel:

- Analysis of genes included in any specifically designed panel may also be ordered as a stand-alone single gene sequencing test or custom panel (2-10 genes) as dictated by the patient's clinical and laboratory phenotype

Targeted familial variant analysis:

- Targeted variant analysis for clinical diagnosis, carrier identification or prenatal diagnosis can also be performed on any gene in the panel when the pathogenic variant(s) is known in the family (test code: 4970)

Confidence begins with clarity

BloodCenter of Wisconsin has variant classification policies that are founded in established practice and are guided with unparalleled experience in hematology.

Our genetic tests are specifically designed and optimized for the detection of germline variants known to cause or contribute to numerous common and rare hematological disorders.

We adhere to the 5-tier classification system outlined in the American College of Medical Genetics (ACMG) Standards and Guidelines (Richards, et al, 2015), which are nationally recognized recommendations for clinical variant interpretation among genetic testing laboratories:

1. **Pathogenic:** Conclusive evidence demonstrates that the variant directly causes or contributes to disease
2. **Likely Pathogenic:** Strong evidence supports that the variant causes or contributes to disease
3. **Uncertain Significance:** Current evidence on the identified variant is insufficient or inconclusive
4. **Likely Benign:** Strong evidence supports that the variant does not cause or contribute to disease
5. **Benign:** Conclusive evidence that the variant does not cause or contribute to disease

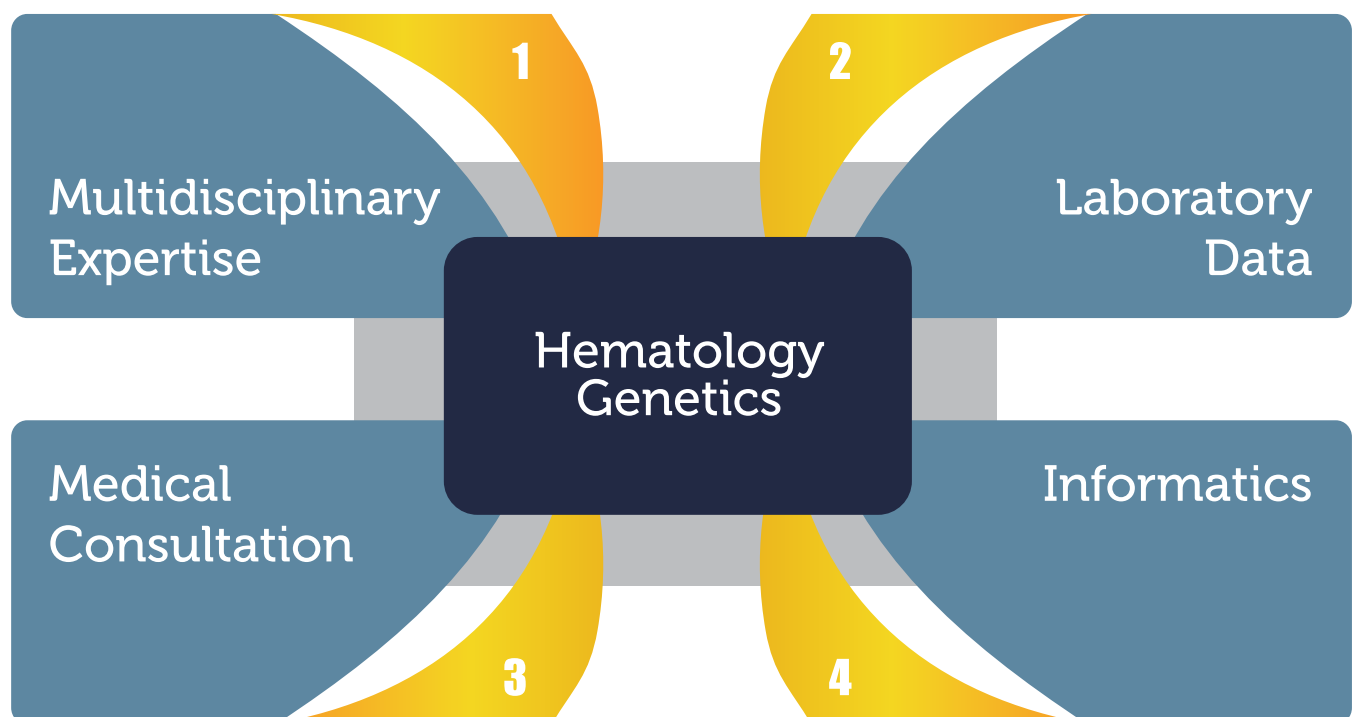
Disease-Focused for High Quality Analysis

With BloodCenter of Wisconsin, you can have confidence knowing that each panel is specifically designed for hematology diagnosis, with careful gene by gene selection for clinical validity and utility, with comprehensive coverage of relevant regions.

Other panels built upon an exome or genome platform to analyze data across the entire set of human genes, scale back data analysis to cover only designated genes. In this approach, breadth is offered at the expense of depth, resulting in gaps in coverage of potentially relevant genes.

Our NGS panels are not simply scaled-back analysis, but are custom-built and designed to include every clinically relevant part of each gene expertly selected for inclusion. Supplemental Sanger sequencing to optimize reliable and accurate detection of variants is included in our panel design. This approach to panel design maximizes diagnostic utility and minimizes the risk of off-target results or incidental findings more likely to be identified by a broader testing platform designed for a less specific purpose.

Panels designed for hematology challenges, by hematology experts.



Panel , Test Code and CPT Codes		
Panel	Test Code	CPT Codes
aHUS Genetic Evaluation	1200	81479
Coagulation Disorder Panel	4815	81240, 81241, 81332, 81355, 81400x5, 81401, 81405, 81407, 81408, 81479
Comprehensive Bleeding Disorder Panel	4825	81240, 81241, 81332, 81355, 81400x6, 81401x2, 81405, 81408x2, 81479
Comprehensive Platelet Disorder Panel	4830	81400x3, 81402, 81403, 81406, 81479
Congenital Neutropenia Panel	4845	81250, 81406x4, 81479
Inherited Thrombocytopenia Panel	4840	81400x3, 81402, 81403, 81406, 81479
Platelet Function Disorder Panel	4835	81400x3, 81479
Thrombosis Panel	4820	81400, 81479
Custom Blood Disorder Panel	4850	Varies by order

The CPT codes provided are subject to change. CPT codes are provided only as guidance to assist clients with billing.

Specimen requirements

Parental/Patient/Pediatric: 3-5 mL Whole blood (EDTA tube, lavender top), 2-5 mL Bone marrow (EDTA tube, lavender top), 3-4 Buccal swabs or $\geq 1\mu\text{g}$ of DNA at $\geq 50\text{ng}/\mu\text{L}$ of High Quality DNA.

Fetal: 7-15 mL Amniotic fluid, 5-10 mg Chorionic villi; back up culture of amniocytes or chorionic villi is highly recommended. Cultured: Two T25 flasks cultured amniocytes or cultured chorionic villi (2×10^6 minimum). Maternal blood sample of 3-5 mL Whole blood (EDTA tube, lavender top) is requested for all prenatal samples for maternal cell contamination studies.

If questions, please contact the laboratory to discuss sample requirements.



SHIP

Shipping requirements

Ship on an ice pack or at room temperature. Protect from freezing. Place the specimen and the requisition into plastic bags and seal. Insert into a Styrofoam container, seal and place into a sturdy cardboard box, and tape securely. Ship the package in compliance with your overnight carrier guidelines. Label with the following address:

**Client Services/Diagnostic Laboratory
BloodCenter of Wisconsin
638 N. 18th St.
Milwaukee, WI 53233**



ORDER

Required forms

Please complete all pages of the requisition form. Clinical history (including patient's ethnicity, clinical diagnosis, family history and relevant laboratory findings) is necessary for optimal interpretation of genetic test results and recommendations. Clinical and laboratory history can either be recorded on the requisition form or clinical and laboratory reports can be included with the sample.

For additional information related to shipping, billing or pricing, please contact, BloodCenter Client Services: (414) 937-6396 or 800-245-3117, Option 1, or LabInfo@bcw.edu.