# Jill M. Johnsen, MD
# 2019 HTRS Mid-Career Research Award (MCRA) Recipient
# Supported by an educational grant from Genentech

Dr. Johnsen is a physician scientist with expertise in classical (benign) hematology. She is an Associate Member at the Bloodworks Northwest (BWNW) Research Institute and Associate Professor in the Division of Hematology, Department of Medicine at the University of Washington (UW) in Seattle. Dr. Johnsen's research program focus is in hemostasis and blood groups. She also sees patients in the Washington Center for Bleeding Disorders (WACBD) at Bloodworks Northwest (BWNW), where she cares for patients with bleeding disorders and their families.

Dr. Johnsen has devoted her career to benign hematology research studying the molecular mechanisms of hemostasis using insights from genetics and molecular biology. She began her training studying a model of von Willebrand Disease in the laboratory of Dr. David Ginsburg (University of Michigan). During her Assistant Professorship in Seattle, she used these skills to pivot her research towards basic and translational studies of human hemostasis, including participating in the NHLBI ESP-GO project and joining the My Life, Our Future (MLOF) hemophilia genotyping project. She has gone on to expand projects studying the genetic basis of variant hemostasis, including serving as an Investigator in the NHLBI TOPMed Program on behalf of MLOF. Dr. Johnsen's lab has become expert in multiple genetic methods and hemostasis assays, and her group has also developed new methods for testing coagulation factors. Dr. Johnsen's goal is to bring together her clinical interests with her laboratory expertise to better understand the molecular basis of how inherited bleeding disorders uniquely impact females, particularly in hemophilia and von Willebrand disease.

**The HTRS MCRA Program**

Dr. Johnsen is the recipient of the 2019 HTRS Mid-Career Research Award (MCRA) for her project titled: "Study of Skewed Factor Levels and Bleeding in Female Genetic Carriers of Hemophilia." The goals of this project are to further the understanding of the biology of factor levels and bleeding in female genetic carriers of hemophilia and to provide the foundations for a larger-scale research program that investigates the causes of factor variation and bleeding in female genetic carriers of hemophilia.

Hemophilia A and B are inherited X-linked bleeding disorders caused by deficiencies in coagulation factor VIII (FVIII) or factor IX (FIX), respectively. Female genetic carriers of hemophilia are women and girls that have DNA changes that cause hemophilia in males. In females, being a genetic carrier of hemophilia is associated with a wide range of factor levels and bleeding symptoms, although a substantial portion of this variation remains unexplained.

In this HTRS MCRA, Dr. Johnsen hypothesizes that specific characteristics of the hemophilia-causing DNA changes (genotypes) are responsible for variable factor levels and contribute to excessive bleeding risk in female genetic carriers of hemophilia. She will study the role of hemophilia genotype, X-chromosome inactivation, the performance of different coagulation factor laboratory assays relative, and further assess von Willebrand Factor as a modifier of bleeding in 400 female genetic carriers who participated in My Life, Our Future (MLOF), a national hemophilia genotyping project. This work will significantly advance our understanding of variation in factor levels and bleeding in females genetic carriers of hemophilia, and will lay the groundwork for expanding a program of research to better understand the biology of bleeding in female genetic carriers of hemophilia.

Dr. Johnsen's goal is that her MCRA-funded project will develop into a large-scale study of female hemophilia carriers. "It is a tremendous honor to receive this HTRS Mid-Career Research Award, I am grateful for the significant support this award will provide me to advance my research on female genetic carriers of hemophilia. This MCRA work will form the basis of applications for large-scale NIH funding, and will enable me to fully transition to an established senior scientist position."

The HTRS Mid-Career Research Award (MCRA) Program provides financial support for mid-career investigators pursuing clinical, translational, or basic science research projects in hemostasis and/or thrombosis.

The goals of the MCRA Program are to:

- Provide financial support for talented mid-career physician-scientists to enable them to **transition from mentored research to full research independence**.
- **Combat the shortage of skilled academic researchers in benign hematology** by supporting mid-career physician-scientists in the U.S. and Canada at a critical juncture in their academic research careers. Upon the completion of a project funded by the MCRA Program, the recipient should be ready to apply for a large-scale grant to expand their research in hemostasis and/or thrombosis such as an NIH R01, NIH R34, or the equivalent from another agency such as the American Heart Association or the Canadian Institutes of Health Research (CIHR).
- **Advance the science underlying the clinical management of hemostasis and thrombosis disorders** by supporting research to improve the health and well-being of people living with these disorders in the U.S. and Canada.

The 2019 HTRS Mid-Career Research Award Program is supported by a medical education grant to HTRS from Genentech. According to Dr. Richard Ko, Senior Medical Director at Genentech, "Genentech is proud to support the Hemostasis and Thrombosis Research Society 2019 Mid-Career Research Award (MCRA). With an ever-changing therapeutic landscape and the approval of new hemophilia treatment options, we recognize the importance of ensuring all patients with hemophilia who can benefit from these advances are able to take advantage of them and that investigators are able to study all populations, especially those who may have been neglected in the past."