Society for Pediatric Research

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Desch, Karl

I graduated from Washington University in St. Louis School of Medicine in 1997 and from my residency in pediatrics at St. Louis Children's Hospital in 2000. I completed my fellowship training in neonatal-perinatal medicine at the University of Michigan in 2005. I am currently an Assistant Professor in the Department of Pediatrics and Communicable Disease at the University of Michigan and have both clinical, research and educational effort. I am board certified in Neonatal-Perinatal Medicine and I am an active member in the American Society of Hematology, The American Society of Human Genetics and the International Society of Thrombosis and Haemostasis.

My laboratory focuses on the functional characterization of human genetic variants that play critical roles in the regulation of hemostasis and thrombosis. For example, as part of an international collaboration, I directed a genomic study of venous thromboembolic disease using whole-exome sequencing in 400 cases and 6000 controls. Using a gene-collapsing analysis, we identified an excess of rare damaging mutations in the coding sequence of 4 genes leading to the identification of a new candidate gene in thrombophilia risk. My investigative approach is to use non-biased genetic screening studies for variant discovery followed by mechanistic examination of gene variants in model systems. Through basic research, I hope to improve the understanding of the molecular pathogenesis of blood related disorders affecting children and adults.