Upcoming Discovery Lecture:

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The Genetics of Congenital Heart Disease: Murmurs and Hiss Tones

Congenital heart disease (CHD) comprises a variety of cardiovascular malformations, which, collectively, affects approximately 2% of newborns. Over the past few decades, there has been stunning progress in devising effective interventions for CHD. Despite that, CHD remains the leading cause of newborn deaths due to birth defects, and survivors continue to suffer from co-morbidities such as neurodevelopmental delays as well as early mortality. Elaborating the genetic causes of CHD, believed to account for the majority of its etiology, is of importance, both for devising prevention and therapeutic strategies. For this Flexner Discovery Lecture, I will review the genetic epidemiology of CHD as well as ongoing efforts, principally through the use of next generation sequencing-based approaches, to elaborate the genetic architecture of CHD as a complex genetic trait. In addition, the implications of these findings as they related to outcomes for CHD survivors will be discussed. Finally, opportunities ahead to explain the missing heritability of CHD will be described.

Bruce D. Gelb, M.D. is the Gogel Family Professor and Director of the Mindich Child Health and Development Institute at the Icahn School of Medicine at Mount Sinai in New York City. He is also Professor of Pediatrics and Genetics & Genomic Sciences. Dr. Gelb received his M.D. degree from the University of Rochester. He trained in pediatrics at the Babies’ Hospital, Columbia-Presbyterian Medical Center in New York City and then in pediatric cardiology at Texas Children’s Hospital in Houston, Texas. During the latter, he also received training in molecular genetics at the Institute for Molecular Genetics at the Baylor College of Medicine. Following his training, Dr. Gelb joined the faculty at Mount Sinai where he has remained throughout his career. Clinically, he is the founder and Co-Director of the Cardiovascular Genetics Program, a multi-disciplinary program caring for patients and families with genetic-based cardiovascular disorders. For his research, Dr. Gelb has focused on using genetic approaches to understand the causes of cardiac defects. His group identified the first gene for Noonan syndrome (PTPN11) and has made several other gene discoveries for the RASopathies. The Gelb group continues to search for new RASopathy genes, to explore the biology of those disorders and to search for potential therapies for them. Dr. Gelb is also a site Principal Investigator for the NHLBI-funded Pediatric Cardiac Genomics Consortium, which is using ‘omic’ approaches to understand the genetic causes of congenital heart defects. Dr. Gelb is the President of the International Pediatric Research Foundation, the President-Elect of the American Pediatric Society, and the Treasurer-Elect for the American Society of Human Genetics. He is an elected member of the National Academy of Medicine and the American Society for Clinical Investigation.