PhenX Measures for Blood Sciences Research
Special Report

Funded by the National Heart, Lung, and Blood Institute (NHLBI), PhenX Measures for Blood Sciences Research includes collections of protocols to support Sickle Cell Disease (SCD) and Hemophilia Inhibitor research. The PhenX Blood Sciences Research Collections are the result of a collaborative, consensus effort by the extramural scientific community, the NHLBI and the National Human Genome Research Institute (NHGRI). Consistent use of these standard measurement protocols will establish a common currency to help researchers better understand the etiology, progression, and treatment of SCD and hemophilia, promote collaboration, and facilitate cross-study data analysis.

Blood Sciences Research

PhenX Measures for Sickle Cell Disease (SCD) Research

The NHLBI launched the PhenX Measures for SCD Research project in 2014 to establish a framework for data sharing across different SCD research projects. In 2019, the NHLBI funded the Expansion of PhenX Measures for SCD Research project to extend this framework by updating and expanding SCD protocols in the Toolkit. Ellen Werner, PhD, MA, served as the NHLBI Project Scientist from 2014 until her retirement in 2019. William P. Tonkins Jr, DrPH, is the current NHLBI Project Scientist.

The recent addition of the SCD Pregnancy Collection completes the Expansion of PhenX Measures for SCD Research project. There are now nine PhenX SCD Collections of protocols in the PhenX Toolkit:

1. SCD Core: Tier 1

2. SCD Core: Tier 2
3. SCD Cardiovascular, Pulmonary, and Renal
4. SCD Neurology, Quality of Life, and Health Services
5. SCD Curative Therapies
6. SCD Pain
7. SCD Psychosocial and Social Determinants of Health
8. SCD Genitourinary
9. SCD Pregnancy

SCD Core Tier 1 and Tier 2 Collections

The SCD Core Collections are recommended for use by all NHLBI funded investigators performing human subjects research in SCD. These protocols are designed to create a common framework to facilitate cross-study data aggregation and to address questions related to the effects of a key set of SCD-related factors on relevant outcomes.

The SCD Core Collections were developed by the SCD Research and Scientific Panel (SRSP) as part of the PhenX Measures for SCD Research project and released in 2015. The SCD Core Collections were updated by the SRSP 2023.

SCD Specialty Collections

The seven SCD Specialty Collections are complementary to the SCD Core Collections and are deemed relevant and essential within these more specialized research domains. Two Specialty Collections (SCD Cardiovascular, Pulmonary, and Renal, and SCD Neurology, Quality of Life, and Health Services) were added by Working Groups (WG) under the PhenX Measures for Sickle Cell Disease Research project and released in 2015. The remaining five Specialty Collections were completed by WGs under the Expansion of PhenX Measures for SCD Research project and released between 2021 and 2024.

PhenX Measures for Hemophilia Inhibitor Research

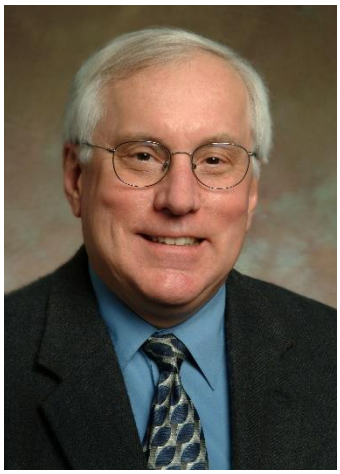
The NHLBI launched the PhenX Measures for Hemophilia Inhibitor Research project in 2017 to establish a framework for data sharing across different hemophilia research projects. The NHLBI project scientists were Dr. Ellen Werner and Dr. Donna DiMichele. Released in 2019, the Hemophilia Inhibitor Research Collection includes 17 measures and 38 protocols to determine levels of Factor VIII and Factor IX, quantify inhibitors to

Factor VIII and Factor IX, measure the biological response to therapeutics, and capture key outcomes.

Recommended Collection: Social Determinants of Health Core

The Social Determinants of Health (SDOH) Core Collection is recommended for use by all Toolkit users. Social Determinants of Health play an integral role in human health and well-being and affect a wide range of health risks and outcomes. SDoH are particularly important for those with life-long, chronic health conditions such as SCD and hemophilia.

PhenX Measures for SCD Research Leadership



James Eckman, MD, SRSP Co-chair

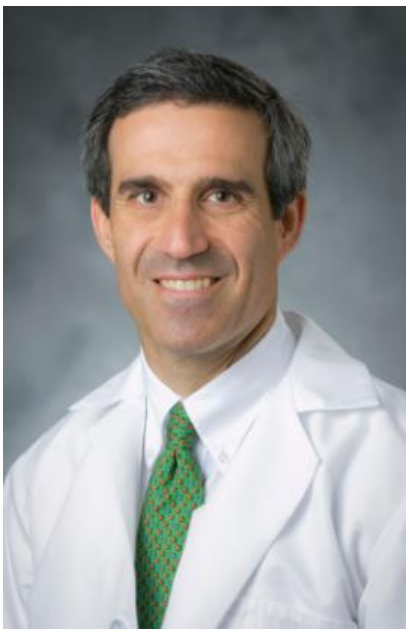
Dr. Eckman is Emeritus Professor of the Department of Hematology and Medical Oncology and held appointments as Professor of Medicine and Adjunct Professor of Pediatrics in Medical Genetics at Emory University School of Medicine. In 1978 He established the adult sickle cell sickle cell program at Grady Memorial Hospital, and in 1984, he helped establish and became Medical Director of the world's first 24-hour comprehensive acute care sickle cell center that presently serves a patient population of more than 1,000 active patients from a population of over 5,500 registered patients. This program serves a model for many day clinics that are providing improved care for individuals with acute complications of sickle cell disease.

He has broad research experience. Early studies focused on methods for newborn screening and the psychological impact of sickle cell disease. His basic and translational research focused on characterizing interactions between sickle red cells and endothelial cells and activation of coagulation in sickle cell disease. He collaborated in clinical studies of leg ulcers, preop transfusion, acute chest syndrome, MSH trial of hydroxyurea, bone marrow transplantation, and many other clinical trials. He was Chair of the Oversight Steering Committee for HRSA/MCHB Sickle Cell Disease Treatment Demonstration Program (SCDTDP).

He chairs the Hemoglobin Subcommittee of the Georgia Newborn Screening Advisory Committee working to improve follow-up of infants detected by newborn screening. He is also working on methods of transitioning youth with sickle cell disease from pediatric centered to adult centered medical homes that will work in many different care settings throughout the state. He was a

consultant on the recently completed PCORI ST3P-UP Transition Study. His current focus is improving access to and utilization of primary care medical home for adults with sickle cell disease.

John J. Strouse, MD, PhD, SRSP Co-chair



Dr. Strouse's research has focused on the epidemiology, risk factors, and prevention of the pulmonary and central nervous system complications of sickle cell disease and includes retrospective and prospective cohort studies and clinical trials. Dr. Strouse received his Ph.D. in clinical investigation from the Johns Hopkins Bloomberg School of Public Health for a series of studies to identify predictors of cognitive function in children with sickle cell disease. This work has expanded to the evaluation of the interaction between environment and disease in both children and adults and the functional evaluation of adults with sickle cell disease. Dr. Strouse's other research interests include the application of large clinical, research, and administrative databases to the study of rare hematological diseases and interventions to improve quality of and access to care for sickle cell disease. Dr. Strouse serves on the American Society of Hematology Sickle Cell Taskforce and Sickle Cell Pain Guideline Panel and is co-chair of the American Society of Hematology Healthcare Professional Education and Training Work Group.

NHLBI Leadership

William P. Tonkins, DrPH, JD

Dr. Tonkins has served as the project scientist for the Expansion of PhenX Measures for Sickle Cell Disease Research project since 2019. He is a program officer in the Blood Epidemiology and Clinical Therapeutics branch at the NHLBI.

Ellen M. Werner, PhD, MA

Dr. Werner (retired) served as the NHLBI project scientist for the PhenX Blood Sciences Research Collections between 2014 and 2019. She was a program director in the Blood Epidemiology and Clinical Therapeutics Branch at the NHLBI.

Donna DiMichele, MD

Dr. DiMichele (retired) served as the project scientist for the PhenX Measures for Hemophilia Inhibitor Research Project. She was deputy director of the Division of Blood Diseases and Resources at the NHLBI.