



National Institutes of Health
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NHLBI, CDC Launch Surveillance and Research Program for Inherited Blood Diseases

Six States to Study Sickle Cell Disease and Thalassemias in National Pilot Project

Medical researchers are developing a new surveillance system to determine the number of patients diagnosed with a family of inherited blood disorders known as hemoglobinopathies, including sickle cell disease, thalassemias, and hemoglobin E disease.

The National Heart, Lung, and Blood Institute (NHLBI) of the National Institutes of Health is funding the four-year pilot project, which will involve the Centers for Disease Control and Prevention and six state health departments, to create ways to learn more about the extent of hemoglobinopathies in the United States.

Data collected from the \$27 million Registry and Surveillance System in Hemoglobinopathies (RuSH) project will help researchers determine the most effective plans for developing future hemoglobinopathy registries. Research findings based on data from disease registries may provide new ideas for drug therapies and can spur the development of tests that can determine severity of diseases over the lifespan.

To manage the surveillance efforts, the NHLBI has entered into an interagency agreement with the CDC's National Center on Birth Defects and Developmental Disabilities. As part of the project, the CDC has developed cooperative agreements to create surveillance programs with state health departments in California, Florida, Georgia, Michigan, North Carolina, and Pennsylvania.

Hemoglobinopathies involve problems with hemoglobin, the vital blood component responsible for transporting oxygen throughout the body. Production of abnormal hemoglobin, which occurs in the family of sickle cell diseases and hemoglobin E, or production of too little hemoglobin, which occurs in the thalassemias, can cause organ damage and shorten lifespan. While all states now test newborns for some of these diseases, there is no system to track the diseases nationally. In addition, patients born before screening programs began or those who have immigrated to the United States are not tracked. These statistical gaps make it difficult to know the true impact of hemoglobinopathies in this country. RuSH will help determine how many

people are affected by hemoglobinopathies. Such data are essential for public health agencies to allocate adequate resources to meet the medical and social service needs of hemoglobinopathy patients.

“While we have made great strides in developing treatments for patients with sickle cell disease and other hemoglobinopathies, RuSH stands as the first major surveillance and registry program to gather comprehensive demographic and other information on people with these life-threatening diseases,” said NHLBI Acting Director Susan B. Shurin, M.D., a hematology researcher.

Hemoglobinopathies cause health problems when abnormal hemoglobin genes are inherited from both parents. Individuals who inherit a single abnormal gene, which is called carrying a trait, have few of these health problems.

The hemoglobinopathies are most common in areas where malaria has been endemic. Sickle cell disease is the most common hemoglobinopathy in the United States and the condition affects millions worldwide. Of the estimated 70,000 to 100,000 people in the United States with sickle cell disease, most are thought to have African ancestry, although the gene also occurs among people from the Mediterranean and Middle East. The abnormal hemoglobin molecules of sickle cell disease deform red blood cells, causing them to clump together and block blood flow through blood vessels, leading to painful sickle cell crises, organ damage, anemia (lack of red blood cells), and premature death.

Life-threatening complications include infections, acute chest syndrome, stroke, and pulmonary hypertension (increased blood pressure in the lung arteries). Painful crises are the leading cause of emergency room visits and hospitalizations of people who have sickle cell disease. Life expectancy has increased dramatically with state newborn screening programs and early treatment, which can include daily penicillin treatment for patients age five and younger as well as immunizations for other diseases to prevent complications.

Patients with thalassemia syndromes produce less hemoglobin than normal, and the red blood cells that are produced are rapidly destroyed. Signs and symptoms of thalassemia can include severe anemia; slowed growth and delayed puberty; bone problems; and enlarged spleen, liver, and heart. Severely affected individuals require frequent and repeated blood transfusions and treatments to reduce the accumulation of iron in the body. Thalassemia genes are widespread across the Mediterranean, Middle East, Africa, the Indian subcontinent, and Southeast Asia.

Hemoglobin E diseases are most common among persons with ancestors from Southeast Asia. Affected individuals produce a smaller than normal number of red blood cells. Red blood cells in these individuals are smaller than normal and misshapen. These abnormal red blood cells carry less oxygen to organs. Milder forms of hemoglobin E disease may not need treatment, although affected individuals may have mild anemia. Severe forms of hemoglobin E disease can cause significant anemia, bone pain, and other complications.

Through surveillance under the initial phase of the RuSH pilot program, researchers hope to determine the prevalence of the hemoglobinopathies among screened newborns and patients not identified through newborn screening. The data should help determine the prevalence of the various conditions. The research will also help describe the demographic characteristics of individuals with these conditions as well as their geographic distribution. Researchers will also examine the existing health care resources available for patients with hemoglobinopathies.

“The data gathered through our RuSH surveillance efforts will provide critical knowledge about the current state of care available for patients who have hemoglobinopathies,” said Hani Atrash, M.D., M.P.H, director of the Division of Blood Disorders, National Center on Birth Defects and Developmental Disabilities at the CDC.

To schedule an interview with an NHLBI spokesperson, contact the NHLBI Communications Office at 301-496-4236 or at nhlbi_news@nhlbi.nih.gov.

To schedule an interview with a CDC spokesperson, contact the CDC’s Division of Media Relations at 404- 639-3286 or at media@cdc.gov.

Part of the National Institutes of Health, the National Heart, Lung, and Blood Institute (NHLBI) plans, conducts, and supports research related to the causes, prevention, diagnosis, and treatment of heart, blood vessel, lung, and blood diseases; and sleep disorders. The Institute also administers national health education campaigns on women and heart disease, healthy weight for children, and other topics. NHLBI press releases and other materials are available online at: www.nhlbi.nih.gov.

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Resources:

Diseases and Conditions Index: Sickle Cell Disease:
http://www.nhlbi.nih.gov/health/dci/Diseases/Sca/SCA_WhatIs.html

CDC: Sickle Cell Disease:
<http://www.cdc.gov/ncbddd/sicklecell/>

Diseases and Conditions Index: Thalassemias:
http://www.nhlbi.nih.gov/health/dci/Diseases/Thalassemia/Thalassemia_WhatIs.html

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