

**NIH-NHLBI/NIDDK Thalassemia Workshop:
Clinical Priorities and Clinical Trials
Fishers Lane Conference Center
Rockville, MD
May 20-21, 2009**

The hemoglobinopathies, thalassemia and sickle cell disease, are the most common genetic diseases worldwide, affecting millions of people. However, hemoglobinopathies are orphan conditions in the U.S. with sickle cell disease and thalassemia major affecting approximately 80,000 and 1,000 persons, respectively. Increased immigration to the U.S. is increasing the prevalence and broadening the demographics of the hemoglobinopathies.

On May 20 and 21, 2009, the National Heart, Lung, and Blood Institute (NHLBI) and the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK) convened a workshop to discuss priorities for thalassemia research and clinical trials. The overall goals of the workshop were to identify clinical research needs and trials to reduce the burden of disease.

Prior to the meetings, subcommittees formulated opinions and representatives of the subcommittees presented their recommendations at the workshop on several topics:

- Iron Overload: Clinical Monitoring and Chelation Therapy
- Fetal Hemoglobin: Investigation of Globin Chain Switching
- Stem Cell Transplantation
- Gene Therapy
- Endocrine Disorders/Bone Disease/Growth and Development

The subcommittees were asked to consider the questions:

1. What are the clinical research priorities currently facing the thalassemia community?
2. What are the most important clinical trials (Phase I, II, and III) that should be undertaken?
3. How can successful study completion and patient recruitment be optimized?
4. What is the role of international collaborations in clinical research in thalassemia?

Experts in thalassemia clinical trial research presented the current status of clinical trials conducted in the U.S., U.K, and Thailand. Speakers uniformly recommended that international studies are critical to achieve adequate enrollment in thalassemia clinical trials. There was broad support to keep the goals, protocols and organizational structure as simple as possible and facilitate cooperation among the investigators designing and conducting the studies and clinical research organizations with expertise in regulatory issues for each country. Protocol design can be enhanced with input from the thalassemia patient population. It is important to develop a framework that rewards investigators for attainment of study milestones in a timely manner. An executive summary of the workshop will be posted on the NHLBI and NIDDK websites and used by the NIH to develop future initiatives in thalassemia clinical research.