

Workshop Summary

A one-day workshop on "**Inherited bone marrow failure syndromes (IBMFS): Definitions and diagnostic criteria**" was held at the *Cloisters* building, NIH main campus, on Friday, January 26, 2007. This workshop was co-sponsored by the Hematology Research Program of NIDDK, the Blood Division of NHLBI, and the Office of Rare Diseases, OD, NIH. The workshop was planned and organized as a direct result of concerns raised at a prior *Interagency Coordinating Committee Meeting for Hematology on Inherited Marrow Failure Disorders* organized by the NIDDK Hematology Research Program and held on March 10, 2006.

The specific objectives of this workshop were to establish a current and comprehensive list of inherited bone marrow failure syndromes (IBMFS) that can be recognized as distinct entities by phenotypic and genotypic features, and to define specific diagnostic criteria that must be met to assign affected individuals into one or another IBMFS category (or sub-category). The workshop also aimed to develop widely applicable diagnostic algorithms for IBMFS.

Co-organizers of this workshop were: Drs. **Daniel Wright** (NIDDK), **Blanche Alter** (NCI), and **Susan Shurin** (NHLBI) of the National Institutes of Health, together with Drs. **Akiko Shimamura** of Boston Children's Hospital, Harvard Medical School and **Mohandas Narla** of the New York Blood Center. In addition to these workshop organizers, 15 international IBMFS experts attended the workshop as invited participants: Drs. **Sarah Ball** (St George's Hospital Medical School, London, UK), **Matthias Ballmaier** (Hannover Medical School, Hannover, Germany), **Monica Bessler** (Wash. Univ., St. Louis, MO), **Larry Boxer** (Univ. Michigan, Ann Arbor, MI), **David Dale** (Univ. Washington, Seattle, WA), **Inderjeet Dokal** (Queen Mary's School of Medicine and Dentistry, London, UK), **Peter Durie** (Hospital for Sick Children, Univ of Toronto, Toronto, Canada), **Bert Glader** (Stanford Univ., Palo Alto, CA), **Thierry Leblanc** (Hematologie Hopital St. Louis, Paris, France), **Jeff Lipton** (Schneider Children's Hosp., NY), **Charlotte Niemeyer** (University Children's Hospital, Freiburg, Germany), **Zora Rogers** (Univ. Texas, Southwestern, Dallas, TX), **Philip Rosenberg** (NCI, NIH, Bethesda, MD), **Hannah Tamary** (Schneider Children's Med Cntr of Israel, Tel Aviv, Israel), and **David Williams** (Univ. Cincinnati, Cincinnati, OH).

The workshop was divided into two segments. The first segment was devoted to achieving consensus on a comprehensive list of inherited bone marrow failure syndromes (IBMFS) that can be defined by phenotypic and genotypic features, and the second segment was devoted to defining diagnostic criteria and diagnostic algorithms for these disorders. Individual participants presented summaries and led general discussion for each topic area, as outlined below:

Sessions I - IBMFS categories and sub-categories: (1) IBMFS associated with multi-lineage cytopenias (*Blanche Alter*), (2) IBMFS associated with single lineage cytopenias (*Mohandas Narla*)

Session II - IBMFS definitions and diagnostic criteria: (*At-large Discussants: Charlotte Niemeyer, Thierry Leblanc, Phil Rosenberg*)

(A) IBMFS associated with multi-lineage cytopenias:

Fanconi Anemia (*Akiko Shimamura, David Williams*), **Dyskeratosis Congenita** (*Inderjeet Dokal, Monica Bessler*), **Schwachman-Diamond Syndrome** (*Peter Durie, Akiko Shimamura*), **Amegakaryocytic Thrombocytopenia** (*Matthias Ballmaier, Ellis Neufeld*), **Non-Fanconi Anemia aplasias** (*Blanche Alter, Zora Rogers*).

(B) IBMFS associated with single lineage cytopenias:

Diamond-Blackfan Anemia (*Sarah Ball, Jeff Lipton*), **Severe Congenital Neutropenias** (*Larry Boxer, David Dale*), **Thrombocytopenia "absent radii"** (*Matthias Ballmaier, Ellis Neufeld*), **Congenital Dyserythropoietic Anemias** (*Bert Glader, Hannah Tamary*)

Session III - IBMFS definitions and diagnostic criteria - Summary and Conclusions: *Susan Shurin*

An intended outcome of this workshop is that a detailed summary report of the proceedings and conclusions of workshop, prepared and edited by the organizers and participants, be published during 2007. It is hoped that this report will promote the development and coordination of *IBMFS* patient registries and facilitate the creation of productive *IBMFS* clinical research consortia.