Summary of the 2010 International Consensus Conference from DBAF's Research Director, Steven Ellis, PhD:

The Annual DBA International Consensus Conference is widely acknowledged as the premier showcase for clinical and basic research on Diamond Blackfan anemia. And the 2010 conference held in March in New York City was no exception. What struck me most about this year's conference is the breath of DBA research drawing in more and more investigators of increasingly diverse backgrounds. The other thing that struck me about this meeting is the vital role that the DBAF plays in supporting research in these diverse areas and in supporting new investigators to the field. The DBAF logo was shown time and time again as speakers gratefully acknowledged the Foundation's support for their research.

One of the Foundation's philosophies is to support as much research as possible with the limited funds available with an emphasis on new and innovative research and supporting new investigators to the field. What this means in practical terms is that the Foundation's awards are typically much smaller than those from the Federal Government, but play vital roles in testing new ideas and allowing these investigators to generate preliminary data that will make them competitive for larger awards. What I think is evident from listening to awardees speak at this year's conference is how much they appreciate these funds and how important they have been to making progress in the field.

As a consequence of this funding philosophy, the DBAF's Research Portfolio is fairly broad from gene discovery to drug development, from zebrafish to mice, from the USA to the world, and from young investigators to established investigators. Thus, DBAF support continues to be a significant driving force for progress in the field. Researchers supported through the DBAF presenting at this year's meeting included:

- Dr. Hanna Gazda, Boston's Children's Hospital, Gene discovery efforts in DBA
- Dr. Johan Flygare, Whitehead Institute of Biomedical Research, DBA drug development
- Dr. Shuo Lin, UCLA, zebrafish model of DBA

- Dr. David Bodine, National Human Genome Research Institute, mouse models of DBA
- Dr. Fabrizio Loreni, Universita di Roma Tor Vergata, cellular models of DBA
- Dr. Irma Dianzani, Eastern Piedmont University, DBA mutation database

One of the major developments in the past few years in the field has been the development of animal models for DBA. There were five talks this year on different mouse models of DBA (Flygare, Mason, Bodine, Abkowitz, and Jaako) and two on zebrafish models (Taylor and Lin). Animal models with their complex physiology allow investigators to begin to address the fundamental question of how defects in ribosome synthesis affect erythropoiesis and other developmental pathways. These animal models are also crucial for drug development studies (Flygare). Cellular models of DBA continue to provide interesting insights into features of ribosome synthesis and pathways through which defects in ribosome synthesis trigger signaling pathways that lead to critical cell fate decisions. Numerous talks dealt with this fascinating topic (Fumagalli, Goldberg, Du, Moniz, Caywood, Horos, Loreni, Gleizes). There was even a talk on the prospect of using ribosome synthesis as a tool in DBA diagnosis (Leblanc). Gene discovery continues to play a vital role in our understanding of the basic biology of DBA. Two large scale sequencing efforts were reported by Gazda and Farrar, where it is hoped that the majority of genes giving rise to DBA in patients in North American will soon be identified. Genetic analysis will play an increasingly important role in DBA diagnosis as more genes are identified. It is also hoped that the genes affected may give insights into how patients may respond to various treatment and potentially also provide insights into other clinical parameters like cancer predisposition. In this regard, Alter gave a talk on cancer predisposition in DBA, while Ball discussed the need for regular updates to the DBA clinical consensus document. These topics were also front and center at a poster session where information from many DBA registries throughout the world was presented. Finally, Pospisilova, Vlachos, and Glader gave updates on clinical trials either beginning or underway to carefully analyze the value of leucine and lenalidomide as potential treatments for DBA.

As noted above, the DBA International Consensus Conference continues to be the premier venue for DBA research. The conference was hosted by the

Daniella Maria Arturi Foundation, and supported by the Diamond Blackfan Anemia Foundation, the Office of Rare Diseases at the National Institutes of Health, the National Heart Lung and Blood Institute, and Jack's Fight for a Cure. In addition to clinicians and scientists in attendance, the conference also includes various other interested constituencies included representatives from various Institutes within the National Institutes of Health and other National agencies. I hope the reader will get a sense from this brief summary of the 2010 conference that progress continues to be made on numerous research fronts giving us a better understanding of DBA pathophysiology. The quest now is to exploit this understanding to improve clinical care for DBA patients and identify more effective treatments for this disorder.