15th Diamond Blackfan Anemia International Consensus Conference

Sponsored by:

SATURDAY, MARCH 10th

6:30PM 2018 Kick-Off Dinner ~ The Plaza Ballroom (Second Floor)

SUNDAY, MARCH 11th

7:25 – 8:25 Breakfast ~ Grand Ballroom – Salons III and IV (Lower Level)

Meetings will be held in The Grand Ballroom Salons I and II (Lower Level)

8:30 – 8:50 Welcome Remarks and Introductions
Dawn M. Baumgardner
Executive Director, Diamond Blackfan Anemia Foundation, Inc.

8:50 – 9:00 Opening Remarks
Steven R. Ellis
Research Director, Diamond Blackfan Anemia Foundation, Inc.

9:00 – 10:00 PLENARY SESSION
Heme-globin dyscoordination and the pathogenesis of macrocytic anemia in DBA
Janis L. Abkowitz

Pathogenic Mechanisms
Moderators: Lionel Blanc / Fabrizio Lorenzi

10:00 – 10:10 Ribosome Levels Selectively Regulate Translation and Lineage Commitment in Human Hematopoiesis
Vijay G. Sankaran

10:15 – 10:25 mRNA translational regulations mediated by upstream open reading frames in normal and pathological erythropoiesis
Wenqian Hu

10:30 – 10:40 How do defects in the production of the small ribosomal subunit lead to the activation of the tumour suppressor p53?
Nicholas J. Watkins
10:45 – 11:00  
*Coffee Break and Poster Viewing*

**DBA Models**  
*Moderators – Jeffrey M. Lipton / Lydie Da Costa*

11:05 – 11:15  
Heterogeneity of Human Erythroid Progenitors: Implications for understanding the erythroblastopenia in Diamond Blackfan Anemia  
*Mohandas Narla*

11:20 – 11:30  
A Pharmacologic Approach Using Pomalidomide and Dexamethasone to Study Stage-Specific Regulation of Erythroid Progenitors  
*Anu Narla*

11:35 – 11:45  
Fetal erythropoiesis is defective in heterozygous Rpl11+/− Diamond Blackfan Anemia mice and increases in severity in young animals.  
*Colin A. Sieff*

11:50 – 12:00  
Bone Development and Cancer Predisposition in Diamond Blackfan Anemia (DBA)  
*Jimmy Hom*

12:05 – 12:15  
Development of clinical gene therapy for RPS19-deficient Diamond-Blackfan anemia.  
*Stefan Karlsson*

12:20 – 1:20  
*Lunch - Grand Ballroom – Salons III and IV (Lower Level)*

**Genetics and Genomics of DBA**  
*Moderators: Irma Dianzani / Hanna Gazda*

1:25 – 1:35  
A small nucleolar RNA involved in Diamond-Blackfan anemia  
*Lydie Da Costa*

1:40 – 1:50  
Revertant Somatic Mosaicism – Natural Gene Therapy in Diamond Blackfan Anemia  
*Hamish S. Scott*

1:55 – 2:05  
Deficiency of Adenosine Deaminase 2 is a Common Cause of Congenital Pure Red Cell Aplasia  
*Marcin W. Wlodarski*

2:10 – 2:20  
The Genetic Landscape of Diamond-Blackfan Anemia  
*Jeffrey M. Verboon*
Whole Genome Sequencing to Detect Small Deletions in Ribosomal Protein Genes in Patients with Diamond Blackfan Anemia
    David Bodine

The Ribosomal Phenotype of Lymphoblastoid Cell Lines From DBA Patients Can Be Rescued by Gene Complementation: A Functional Assay for the Classification of RP Variants of Unknown Significance
    Anna Aspesi

Coffee Break and Poster Viewing

New Treatments for DBA - I
    Moderators:  Blanche Alter / Dagmar Pospisilova

Deferiprox as an alternative chelation in Diamond Blackfan anemia patients?
    Thierry LeBlanc

Leucine for the Treatment of Diamond Blackfan Anemia in Transfusion Dependent Patients
    Adrianna Vlachos

Modulation of autophagy as a therapeutic strategy in DBA
    Sergei Doulatov

Exploiting the competition between GATA1 and GATA2 Transcription Factors in Fetal Erythropoiesis to rescue the erythroid developmental defect of Diamond Blackfan Anemia – a Mouse Model
    Yehudit Birger

Genotype-phenotype correlations in Diamond Blackfan Anemia: findings from the United Kingdom DBA cohort
    Josu de la Fuente

The Italian Diamond Blackfan Anaemia Registry: twenty-three years of activity
    Irma Dianzani

Clinical features and Genotype of Patients with mutated or deleted RPL35A in the NCI Diamond Blackfan anemia cohort
    Neelam Giri

Targeting p53 and nucleolar stress in Diamond-Blackfan anaemia
    Melissa D. Ilsley
Consistent Differences in the Epigenetic Profiles of Cultured Erythroid Cells from Transfusion Dependent, Steroid Responsive and Remission Patients with Diamond Blackfan Anemia

Jessica Kang

Greek Registry of Diamond Blackfan Anemia

Antonis Kattamis

Analysis of the translational efficiencies of mRNAs in a zebrafish model of Diamond-Blackfan anemia

Naoya Kenmochi


Arati Khanna-Gupta

6:00 – 8:30  
*Cocktail Reception ~ The Plaza Ballroom (Second Floor)*

**MONDAY, MARCH 12th**

7:40 – 8:40  
*Breakfast ~ Grand Ballroom – Salons III and IV (Lower Level)*

8:45 – 10:00  
*Short Talks (accompanied by Poster) Session II  
Facilitators: David Bodine / Anu Narla*

Global chromatin occupancy and epigenetic signature analysis reveal new insights into the function of GATA1 N-terminus in erythropoiesis

Te Ling

The anemia and lethal bone marrow failure in a mouse model for Diamond Blackfan anemia is cured by bone marrow transplantation without preconditioning

Yang Liu

Quantitative Analysis of rRNA and rDNA in Cells from DBA Patients

Fabrizio Loreni

New phenotypes linked to new RP gene mutations

Alyson MacInnes

Malignant diseases frequency in patients with Diamond-Blackfan anemia in the Czech National DBA Registry

Dagmar Pospisilova
Use of next generation targeted sequencing for enhanced genotyping of Diamond-Blackfan anemia: An update from the Israeli DBA registry

**Hannah Tamary**

Deletions and Novel Mutations in Genes Coding for Ribosomal Proteins (RPL11, RPL35A, RPS19) in Diamond-Blackfan Anemia (DBA)

**Jana Volejnikova**

**10:05 – 10:25**

*Coffee Break and Hotel Check-Out*

**10:30 – 10:45**

Update from the NHLBI

**Dr. Pankaj Qasba**, Program Director
Blood Diseases Branch
Division of Blood Diseases and Resources
National Heart, Lung, and Blood Institute, NIH

**New Treatments for DBA - II**

*Moderators: Blanche Alter / Dagmar Pospisilova*

**10:50 – 11:00**

Small molecule screens identify CDK8 inhibitors as candidate Diamond Blackfan Anemia drugs

**Johan Flygare**

**11:05 – 11:15**

Nemo-like Kinase is Hyperactive in RPS19-Insufficient Erythroid Progenitors

**Mark Wilkes**

**11:20 – 11:30**

High-throughput screening approaches to identify therapeutic targets for the treatment of Diamond Blackfan Anaemia

**Amee J. George**

**11:35 – 11:45**

High throughput discovery of novel regulators of human ribosome biogenesis reveals 4/12 Diamond-Blackfan Anemia genes

**Susan J. Baserga**

**11:50 – 12:15**

Wrap-Up and Closing Remarks

**Steve Ellis**

**12:15 – 12:30**

Farewell

**DBA Foundation**

**12:30**

*Lunch & Departure*