

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 Loreni

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

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International Consensus Conference**

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

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International Consensus Conference**

- 2:25 – 2:35 Whole Genome Sequencing to Detect Small Deletions in Ribosomal Protein Genes in Patients with Diamond Blackfan Anemia
David Bodine
- 2:40 – 2:50 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
- 2:55 – 3:10 *Coffee Break and Poster Viewing*

New Treatments for DBA - I

Moderators: Blanche Alter / Dagmar Pospisilova

- 3:15 – 3:25 Deferiprox as an alternative chelation in Diamond Blackfan anemia patients?
Thierry LeBlanc
- 3:30 – 3:40 Leucine for the Treatment of Diamond Blackfan Anemia in Transfusion Dependent Patients
Adrianna Vlachos
- 3:45 – 3:55 Modulation of autophagy as a therapeutic strategy in DBA
Sergei Doulatov
- 4:00 – 5:30 **Short Talks (accompanied by Poster) Session I**
Facilitators: David Bodine / Anu Narla

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

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International Consensus Conference**

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

Complete clinical response in a patient diagnosed with Goshal's Hematodiphyseal dysplasia following
supplementation with an L-Leucine-rich diet: a bench to bedside story marrow failure in a mouse model
for RPS19-deficient Diamond-Blackfan anemia.
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

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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

Ame 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*
