

# Ninth Annual Diamond Blackfan Anemia International Consensus Conference

Sponsored by:



## SATURDAY MARCH 8<sup>th</sup>

5:15PM Shuttle Service from Ritz Carlton to Tribeca Grill

6:00PM **2008 Kick-Off Dinner – Tribeca Grill**  
Keynote Speaker: **Jason B. Rose**  
Introduction by: **David G. Nathan**

## SUNDAY MARCH 9<sup>th</sup>

8:30 – 9:00 **Breakfast ~ Harbor Vista (Function Level)**

### *Meetings will be held in Salon II & III (Function Level)*

9:00 – 9:30 Welcome Remarks and Introductions  
**Manny and Marie Arturi**, Daniella Maria Arturi Foundation  
**Kathi Vroman and Dawn Baumgartner**, Diamond Blackfan Anemia Foundation

### Animal Models of DBA

Moderators: Stefan Karlsson  
Mohandas Narla

9:30 – 9:40 *RPS19* downregulation in zebrafish as a model for DBA  
**Nadia Danilova**, Kathleen M. Sakamoto and Shuo Lin

9:45 – 9:55 Transgenic mouse models of Diamond Blackfan Anemia associated with the  
RPS19R62W mutation  
Emily E. Devlin and **David M. Bodine**

10:00 – 10:10 Chemically-induced dark skin mouse mutants serve as models for Diamond Blackfan  
anemia  
**Kelly McGowan**, Jun Li, Veronica Beaudry, Holly Tabor, Amit Sabnis, Weibin  
Zhang, Helmut Fuchs, Martin Hrabe De Angelis, Richard Myers, Laura Attardi  
and Greg Barsh

10:15 - 10:30 Coffee Break

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**Ribosomes and Ribosomal Proteins**

Moderators: Monica Bessler  
Jeffrey M. Lipton  
Jonathan Warner

10:30 – 10:50 Ribosomes, Translation and Cell Cycle Regulation in the context of Diamond Blackfan Anemia

**Harvey Lodish**

**Plenary Lecture**

**11:00 – 11:30 Identification of RPS14 as a 5q- syndrome gene by RNA interference screen**

**Benjamin L. Ebert**, Jennifer Pretz, Jocelyn Bosco, Cindy Y. Chang, Pablo Tamayo, Naomi Galili, Azra Raza, David Root, Eyal Attar, Steven R. Ellis, and Todd R. Golub

11:45 – 11:55 Yeast models for DBA and SDS differ in their effect on translation of specific mRNA classes

J.B. Moore IV, J. Farrar, R.J. Arceci, J.M. Liu, and **S.R. Ellis**

12:00 – 12:10 Structural and genetic interaction studies in Diamond Blackfan Anemia

**Til Büender**, Louise M. Tonkin, Stefan M. Freund, Mark Bycroft, and Alan J. Warren

12:15 – 1:15 Lunch ~ Harbor Vista

**Ribosomes and Ribosomal Proteins (continued)**

Moderators: Monica Bessler  
Jeffrey M. Lipton  
Jonathan Warner

1:15 – 1:25 Functional relationships between DBA associated ribosomal proteins

Valérie Choismel, Marlène Faubladièr, Isabelle Léger-Silvestre, Marie-Françoise O'Donohue, Sébastien Fribourg, Hanna Gazda and **Pierre-Emmanuel Gleizes**

1:30 – 1:40 The role of human ribosomal proteins in the maturation of rRNA and ribosome production

Sara Robledo, Rachel Idol, Dan L. Crimmins, David B. Wilson, Jack H. Ladenson, Monica Bessler, and **Philip J. Mason**

1:45 – 1:55 Role of the RPS19-PIM1 interaction in the regulation of cell metabolism

**Fabrizio Loreni**, Valentina Iadevaia, Sara Caldarola, Eleonora Rango, Laura Carosi, Marta Armiraglio, Claudio Santoro, and Irma Dianzani

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**Genetics**

Moderators: Robert Arceci and Hanna Gazda

- 2:00 – 2:10 Mutations of the ribosomal protein genes *l5* and *l11* are common causes of Diamond-Blackfan Anemia; *S15*, *L36*, *S7*, *S27a*, and *S17* are sporadically mutated in DBA  
**Hanna T. Gazda**, Mee Rie Sheen, Valerie Choesmal, Hal Schneider, Natasha Darras, Colin A. Sieff, Sarah E. Ball, Edyta Niewiadomska, Peter E. Newburger, Eva Atsidaftos, Adrianna Vlachos, Jeffrey M. Lipton, Pierre-Emmanuel Gleizes, and Alan H. Beggs
- 2:15 – 2:25 Analysis of genomic variations at the *RPS19* locus (CHR19:47053043-47068080) in a cohort of 89 DBA patients without *RPS19* and *RPS24* mutations  
**Jens Schuster**, Oskar Ericksson, Alvaro Martinez Barrio, Jitendra Badhai, Anne-Sophie Fröjmark, Aleksandra Kaluzny, Erik Bongcam-Rudloff, Sarah Ball and Niklas Dahl
- 2:30 – 2:40 Functional classification of *RPS19* mutations and development of a new database for DBA genes  
**Dianzani I**, Campagnoli MF, Pavesi E, Boria I, Armiraglio M, Aspesi A, Avondo F, Garelli E, Quarello P, Carando A, Fribourg S, Gleizes P-E, Loreni F and Ramenghi U
- 2:45 – 3:00 Coffee Break
- 3:00 – 4:30 Overview and Discussion**  
**Animal Models of DBA**  
– Moderators Karlsson and Narla  
**Ribosomes and Ribosomal Proteins**  
– Moderators Bessler, Lipton and Warner  
**Genetics**  
– Moderators Arceci and Gazda

**Are We Ready for Clinical Trials?**

Moderators: Thierry LeBlanc  
Adrianna Vlachos

- 4:30 – 4:40 Effects of leucine administration in Diamond-Blackfan anemia patients  
**Dagmar Pospisilova**, Jana Cmejlova, Tomas Adam and Radek Cmejla
- 4:45 – 4:55 Proposal for a pilot study of lenalidomide in adult patients with red blood cell transfusion-dependent Diamond-Blackfan Anemia  
**Jason Gotlib**

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**5:00 – 5:30 Overview and Discussion**  
**Are We Ready for Clinical Trials?**  
**– Moderators LeBlanc and Vlachos**

5:30 - 8:30 Cocktail Reception ~ Skyline Vista ~ Function Level

**MONDAY MARCH 10<sup>th</sup>**

8:30 - 9:00 **Breakfast ~ Harbor Vista (Function Level)**

9:00 – 10:30 **Poster Session**

Moderators: George Buchanan and Steven Ellis

1. Laboratory diagnosis of Diamond-Blackfan Anemia  
**Blanche P. Alter**, Neelam Giri, and Bertil Glader
2. *RPS19* and *RPS24* mutations in the National Cancer Institute Diamond-Blackfan anemia cohort  
**Neelam Giri**, Sharon A. Savage, and Blanche P. Alter
3. Diamond Blackfan Anemia in Israel  
Nishri D, Aviner S, Kuperman A, Lotan A, Yacobovich J, Kaplushnik J, Ben Barak A, Vilks-Ravel S, Dvir R, Stepensky P, Zilber R and **Tamary H**
4. The Role of Transplantation in Diamond Blackfan Anemia  
**Adrianna Vlachos**, Eva Atsidaftos, and Jeffrey M. Lipton
5. Severe iron overload in transfusion dependent DBA patients  
**Quarello P**, Roggero S, Vinciguerra T, Campagnoli MF, Longo F, Piga A and Ramenghi U
6. Identification of new *RPS19* deletions using the MLPA technique  
**Quarello P**, Garelli E, Carando A, Campagnoli MF, Brusco A, Pappi P, Barberis M, Dianzani I, and Ramenghi U
7. Analysis of telomeres in peripheral blood mononuclear cells from patients with Diamond-Blackfan Anemia (DBA)  
**Pavesi E**, Aspesi A, Avondo F, Garelli E, Quarello P, Campagnoli MF, Pigullo S, Ramenghi U, and Dianzani I
8. Translational activation mediated by PIM1 kinase  
**Valentina Iadevaia**, Barbara Cerroni, Sara Caldarola, Anna Aspesi, Claudio Santoro, Irma Dianzani, and Fabrizio Loreni
9. Downregulation of *RPS19* expression enhances alternate splicing of *FLVCR1* transcript and dramatically reduces expression of *FLVCR1* protein that is critical for erythroid progenitor cell development  
**Simon P. Duffy**, Michelle A. Rey, Yigal Dror and Chetan Tailor
10. Transgenic mouse models of Diamond Blackfan Anemia associated with the *RPS19R62W* mutation  
**Emily E. Devlin** and David M. Bodine

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10:30 – 11:00 **Overview of Poster Session with Coffee Break  
– Moderators Buchanan and Ellis**

**Pathogenesis & Mechanisms**

Moderators: Blanche P. Alter and Sarah Ball

11:00 – 11:10 Pathogenesis of the erythroid failure in Diamond Blackfan Anemia  
**Colin A. Sieff** and Harvey Lodish

11:15 – 11:25 Heme toxicity: a potential pathophysiology in Diamond-Blackfan Anemia (DBA)  
**Sioban Keel**

11:30 – 11:40 An update on the role of FLVCR1 in Diamond Blackfan Anemia  
**Michelle Rey**, Jennifer Brown, Simon Duffy, James Kennedy, John Dick, Yigal Dror and Chetan Tailor

11:45 – 11:55 Potential molecular mechanism for the therapeutic effect of steroids in Diamond Blackfan Anaemia  
Carine Marius, Yaw Ohene-Abuakwa and **Sarah Ball**

12:00 – 12:10 Retrospective study of 32 DBA patients who underwent complete remission  
**Meerpohl JJ**, Happersberger N, Kartal M, and Niemeyer CM

12:15 - 12:35 **Overview and Discussion**  
**Pathogenesis & Mechanisms**  
– **Moderators Alter and Ball**

12:35 – 1:00 Using human stem cells to model bone marrow failure  
**George Daley**

1:00 – 1:05 Update from the NHLBI  
**Pankaj Qasba**

1:05 – 1:15 Closing Remarks  
**Marie and Manny Arturi**

1:15 Lunch & Departure

