

# Eleventh Annual Diamond Blackfan Anemia International Consensus Conference

*Sponsored by:*



## **SATURDAY MARCH 13<sup>th</sup>**

6:00PM

Shuttle Service from Ritz Carlton to Tribeca Grill

6:30PM

**2010 Kick-Off Dinner**

Speaker: **Janet Pereira and Jack**

Introduction by: **Marie Arturi**

Keynote Speaker: **Congresswoman Carolyn McCarthy, NY-4<sup>th</sup> District**

## **SUNDAY MARCH 14<sup>th</sup>**

8:30 – 9:00

**Breakfast ~ Harbor Vista (Function Level)**

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

9:00 – 9:15

Welcome Remarks and Introductions  
**Marie Arturi**, Daniella Maria Arturi Foundation

9:15 – 9:45

What's Next? Can we hit targets once we know what they are?  
**Susan Shurin, MD**  
Acting Director  
National Heart Lung Blood Institute

9:45 – 9:55

Thoughts from a young investigator  
**Anupama Narla**

---

*Supported by a conference grant from:*



*Supported by:*



## Translational Research in DBA

Moderators: Monica Bessler  
David Bodine

- 10:00 – 10:10      Suppression of p53 upregulation by inhibition of mTOR in cells depleted of ribosomal proteins  
**Stefano Fumagalli** and George Thomas
- 10:15 – 10:25      Mechanism of prednisone treatment in DBA directs search for novel therapeutic compounds  
**Johan Flygare**, Violeta Rayon Estrada, Colin Sieff, Anupama Narla, Benjamin L Ebert, Harvey F Lodish
- 10:30 – 10:40      A functional test for Diamond-Blackfan Anemia diagnosis?  
Results of the ARNAB study  
**Thierry Leblanc**, Marie-Françoise O’Donohue, Valérie Choemmel, Maud Simansour, Lydie Da Costa, Pierre-Emmanuel Gleizes for the French DBA Group
- 10:45 – 10:55      *Coffee Break*
- 11:00 – 11:10      Cancer Risk in DBA may be lower than in other Inherited Bone Marrow Failure Syndromes  
**Blanche P Alter**, Neelam Giri, Philip S Rosenberg
- 11:15 – 11:25      Confirmed pathogenic mutations are found in 11 of 80 ribosomal protein genes in Diamond Blackfan anemia  
**Hanna T Gazda**, Mee Rie Sheen, Leana Doherty, Adrianna Vlachos, Valerie Choemmel, Marie-Françoise O’Donohue, Hal E Schneider, Catherine Clinton, Colin A Sieff, Peter E Newburger, Sarah E Ball, Edyta Niewiadomska, Michal Matysiak, Bertil Glader, Eva Atsidaftos, Jeffrey M Lipton, Pierre-Emmanuel Gleizes, and Alan H Beggs
- 11:30 – 11:40      Sequence abnormalities in genes encoding ribosomal proteins in patients with Diamond Blackfan anemia: A preliminary report from the resequencing project  
**Jason E Farrar**, Robert J Arceci, Eva Atsidaftos, Hanna Gazda, Alan H Beggs, Steven R Ellis, Jeffrey M Lipton, and Adrianna Vlachos

---

*Supported by a conference grant from:*



*Supported by:*



11:45 – 11:50	Leucine Update	<b>Dagmar Pospisilova</b>
11:50 – 11:55	Leucine Trial Update	<b>Adrianna Vlachos</b>
	Lenalidomide Trial Update	<b>Bert Glader</b>
12:00 – 12:10	Treatment of Diamond Blackfan Anemia: is it time to modify the consensus document?	<b>Sarah Ball</b>

### Model Systems of DBA

Moderators: Niklas Dahl  
Kelly McGowan

12:15 – 12:25	Modeling Diamond Blackfan Anemia in the Mouse	Rachel A Idol, Sinisa Volarevic, Monica Bessler, <b>Philip J Mason</b>
12:30 – 12:40	A Mouse Model of Diamond-Blackfan Anemia Caused by Dominant Negative Effects of a Point Mutation in the RPS19 Gene	Emily E Devlin, Lydie Da Costa, Mohandas Narla, Shenja Patterson, Gene Elliott and <b>David M Bodine</b>
12:45 – 12:55	Characterization of RPS6 heterozygous mice as a model of Diamond-Blackfan Anemia	Sioban Keel, Akiko Shimamura, and <b>Janis Abkowitz</b>
1:00 – 1:10	A Novel Mouse Model for RPS19-deficient Diamond-Blackfan Anemia Locates the Erythroid Defect at CFU-E / Proerythroblast Transition	<b>Pekka Jaako</b> , Johan Flygare, Karin Olsson, Axel Schambach, Christopher Baum, Jonas Larsson, David Bryder, Stefan Karlsson
1:15 – 2:15	<i>Lunch</i>	

*Supported by a conference grant from:*



*Supported by:*





4:30 – 5:15

**Poster Session (with Coffee Break)**

Moderators: Kathleen Sakamoto  
Jeffrey M Lipton

Update of the Diamond Blackfan Anemia Mutation Database

I Boria, E Garelli, A Aspesi, H Gazda, P Quarello, JJ Meerpohl, L Da Costa, A Proust, T Leblanc, M Simansour, N Dahl, A-S Fröjmark, D Pospisilova, R Cmejla, AH Beggs, MR Sheen, M Landowski, C Buros, C Clinton, L Dobson, A Vlachos, E Atsidaftos, JM Lipton, U Ramenghi, **I Dianzani**

Improved Outcomes in Diamond Blackfan Anemia Treated Via Stem Cell  
Transplantation since the Year 2000

**Adrianna Vlachos**, Jahan Aghalar, Eva Atsidaftos, Jeffrey M Lipton

Neoplasia in Diamond Blackfan Anemia (DBA) as reported to the Diamond Blackfan  
Anemia Registry (DBAR)

**Adrianna Vlachos**, Eva Atsidaftos, Jeffrey M Lipton

Genotype and phenotype of hereditary DBA in German patients

**M Kartal**, JJ Meerpohl, A Fischer, L daCosta, M Wlodarski, CM Niemeyer

Clinical and genetic heterogeneity of German DBA patients

**JJ Meerpohl**, L daCosta, M Kartal, A Fischer, M Wlodarski, CM Niemeyer

Ribosomal protein S19 5'UTR variants: Effects on translation and  
implications for Diamond-Blackfan anemia

Jitendra Badhai, Anne-Sophie Fröjmark, Olof Gidlöf, Jens Schuster, **Niklas Dahl**

Genetic variants in the non-coding region of RPS19 gene in Diamond-Blackfan anemia:  
Potential implications for phenotypic heterogeneity

Aurore Crétien, Alexis Proust, Jean Delaunay, Patricia Rincé, Thierry Leblanc, Rolande Ducrocq, Maud Simansour, Isabelle Marie, Hannah Tamary, Jörg Meerpohl, Charlotte Niemeyer, Hanna Gazda, Colin Sieff, Sarah Ball, Gil Tchernia, Narla Mohandas, and **Lydie Da Costa**

Severe iron overload in Diamond Blackfan Anemia.

**Quarello P**<sup>1</sup>, Roggero S<sup>2</sup>, Vinciguerra T<sup>2</sup>, Longo F<sup>2</sup>, Dianzani I<sup>3</sup>, Piga A<sup>2</sup>, Ramenghi U<sup>1</sup>.

5:15 – 5:30 Wrap-Up: **Poster Session**

**6:00 – 9:00 Cocktail Reception ~ Rise Bar ~ 14<sup>th</sup> Floor**

---

*Supported by a conference grant from:*



*Supported by:*



**MONDAY MARCH 15<sup>th</sup>**

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

**Biochemistry, Cell Biology and Mechanisms**

Moderators: Steven Ellis  
Ben Ebert

- 9:00 – 9:10 Distinct Differences in in vitro Erythroid Proliferation and Differentiation, p53 and Apoptosis Pathways in Diamond-Blackfan Anemia in Conjunction with Depletion of RPS19, RPL5 and RPL11 mRNA  
**Hélène Moniz**, Corinne Hurtaud, Thierry Leblanc, Gil Tchernia, Narla Mohandas, and Lydie Da Costa
- 9:15 – 9:25 Differential down-regulation of RPL35a in human bone marrow progenitors demonstrates a p53 independent mechanism mediating the Diamond Blackfan Anemia phenotype  
**Emi Hojo Caywood**, Jason E Farrar, Jeffrey M Lipton, and Robert J Arceci
- 9:30 – 9:40 Ribosomal deficiencies cause translational deregulation of genes crucial for erythropoiesis  
**Rastislav Horos**, Hanna I Jspeert, Fane Mensah, Erdogan Taskesen, Ivo Touw, Dagmar Pospisilova, Marieke von Lindern
- 9:45 – 9:55 Role of DBA-associated ribosomal proteins in regulation of p53 and c-Myc  
Mu-Shui Dai, Xiao-Xin Sun, Shelya X Zeng, Jun-ming Liao, Guifeng He, Jun-ho Lee, Dorothy Lo, Xiang Zhou, **Hua Lu**
- 10:00 – 10:10 Regulatory mechanisms in the synthesis of ribosomal components in response to ribosomal stress  
Angelo Gismondi, Sara Caldarola, **Fabrizio Loreni**
- 10:15 – 10:30 *Coffee Break*

---

*Supported by a conference grant from:*



*Supported by:*



- 10:30 - 10:40      Loss of function of ribosomal proteins linked to DBA alters reformation of nucleoli at the end of mitosis  
Carron C, Choesmal V, Faubladier M, O'Donohue MF, **Gleizes PE**
- 10:45 – 10:55      Structural, biochemical and genetic dissection of the RPS19-18S rRNA interface  
Til Buender, Mark Bycroft, Stefan M Freund, **Alan J Warren**
- 11:00 - 11:30      **Wrap-up: Biochemistry, Cell Biology and Mechanisms**  
Moderators:    Steven Ellis  
                         Ben Ebert
- 11:30 – 11:45      Update from the NHLBI  
**Pankaj Qasba, Ph.D.**  
Program Director  
Blood Diseases Branch  
Division of Blood Diseases and Resources  
National Heart, Lung, and Blood Institute, NIH
- 11:45 – 12:00      Update from the NIDDK  
**Terry Rogers Bishop, Ph.D.**  
Hematology Program Director  
DKUHD/NIDDK/NIH
- 12:00 – 12:15      Closing Remarks  
**Marie and Manny Arturi**
- 12:30                *Lunch & Departure*

---

*Supported by a conference grant from:*



*Supported by:*

