Eleventh Annual Diamond Blackfan Anemia International Consensus Conference

Sponsored by:

SATURDAY MARCH 13th

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-4th District

SUNDAY MARCH 14th

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

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### Translational Research in DBA

**Moderators:** Monica Bessler  
David Bodine

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<th>Time</th>
<th>Title</th>
<th>Presenters</th>
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<td>10:00 – 10:10</td>
<td>Suppression of p53 upregulation by inhibition of mTOR in cells depleted of ribosomal proteins</td>
<td>Stefano Fumagalli and George Thomas</td>
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<tr>
<td>10:15 – 10:25</td>
<td>Mechanism of prednisone treatment in DBA directs search for novel therapeutic compounds</td>
<td>Johan Flygare, Violeta Rayon Estrada, Colin Sieff, Anupama Narla, Benjamin L Ebert, Harvey F Lodish</td>
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<td>10:45 – 10:55</td>
<td><strong>Coffee Break</strong></td>
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<td>11:00 – 11:10</td>
<td>Cancer Risk in DBA may be lower than in other Inherited Bone Marrow Failure Syndromes</td>
<td>Blanche P Alter, Neelam Giri, Philip S Rosenberg</td>
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<tr>
<td>11:15 – 11:25</td>
<td>Confirmed pathogenic mutations are found in 11 of 80 ribosomal protein genes in Diamond Blackfan anemia</td>
<td>Hanna T Gazda, Mee Rie Sheen, Leana Doherty, Adrianna Vlachos, Valerie Choesmel, 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</td>
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<td>11:30 – 11:40</td>
<td>Sequence abnormalities in genes encoding ribosomal proteins in patients with Diamond Blackfan anemia: A preliminary report from the resequencing project</td>
<td>Jason E Farrar, Robert J Arceci, Eva Atsidaftos, Hanna Gazda, Alan H Beggs, Steven R Ellis, Jeffrey M Lipton, and Adrianna Vlachos</td>
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Eleventh Annual Diamond Blackfan Anemia
International Consensus Conference

11:45 – 11:50
Leucine Update
Dagmar Pospisilova

11:50 – 11:55
Leucine Trial Update
Adrianna Vlachos
Lenalidomide Trial Update
Bert Glader

12:00 – 12:10
Treatment of Diamond Blackfan Anemia: is it time to modify the consensus document?
Sarah Ball

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, Philip J Mason

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, Shenia Patterson, Gene Elliott and David M Bodine

12:45 – 12:55
Characterization of RPS6 heterozygous mice as a model of Diamond-Blackfan Anemia
Sioban Keel, Akiko Shimamura, and Janis Abkowitz

1:00 – 1:10
A Novel Mouse Model for RPS19-deficient Diamond-Blackfan Anemia Locates the Erythroid Defect at CFU-E / Proerythroblast Transition
Pekka Jaako, Johan Flygare, Karin Olsson, Axel Schambach, Christopher Baum, Jonas Larsson, David Bryder, Stefan Karlsson

1:15 – 2:15
Lunch
2:20 – 2:40  
Modeling congenital bone marrow failure syndromes using induced pluripotent stem cells  
Suneet Agarwal and Yuin-Han Loh

2:45 – 2:55  
Hematopoietic Differentiation Defects in Rps19- and Rpl5- Deficient Murine Embryonic Stem (ES) Cell Models of Diamond-Blackfan Anemia  
Tracie A Goldberg, Sharon Singh, Adrianna Henson, Abdallah Nihrane, Jeffrey M Lipton, Steven R Ellis, and Johnson M Liu

3:00 – 3:10  
Deficiency of RPS19 alters RPL11 translation in human primary hematopoietic cells  
Hong Yan Du, Stefano Fumagalli, Sara Kozma, George Thomas, Akiko Shimamura

3:15 – 3:25  
RPS29 is required for blood stem cell production in the Zebrafish Embryo  
Alison M Taylor, Caroline E Burns, and Leonard I Zon

3:30 – 3:40  
Ribosomal protein L11 mutation in zebrafish leads to hematopoietic and metabolic defects  
Nadia Danilova, Kathleen M Sakamoto and Shuo Lin

3:45 – 4:30  
Wrap-Up:  Translational Research in DBA  
Moderators: Monica Bessler  
David Bodine

Model Systems of DBA  
Moderators: Niklas Dahl  
Kelly McGowan

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4:30 – 5:15  **Poster Session (with Coffee Break)**  
**Moderators:** Kathleen Sakamoto  
Jeffrey M Lipton  

**Update of the Diamond Blackfan Anemia Mutation Database**  

**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** 1, Roggero S 2, Vinciguerra T 2, Longo F 2, Dianzani I 3, Piga A 2, Ramenghi U 1.  

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

6:00 – 9:00  **Cocktail Reception ~ Rise Bar ~ 14th Floor**
MONDAY MARCH 15th

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 Lorenzi**

10:15 – 10:30  **Coffee Break**
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

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