

**12th Diamond Blackfan Anemia
International Consensus Conference**

Sponsored by:



DiamondBlackfanAnemia.org

Daniella Maria Arturi Foundation  Research For The Cure

SATURDAY MARCH 17th

6:00PM Shuttle Service Departure from Ritz Carlton to Bobby Van's Steakhouse
6:30PM **2012 Kick-Off Dinner**

Bobby Van's Steakhouse & Grill
25 Broad St (at Exchange Pl.)
New York, NY 10004

Speaker: Jessica & Jeff Bond – Captain Courageous, Australia
Introduction by: **Marie Arturi**

SUNDAY MARCH 18th

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 – 9:40 **Plenary Session:** “Assessment of iron loading in Diamond Blackfan anemia”
Thomas Coates
9:45 – 10:10 **Ellis Neufeld**
10:15 – 10:30 Discussion

Developmental Biology

Moderators: Mitchell Weiss/ George Thomas

10:30 – 10:40 **Lydie DaCosta** - Could DBA be revealed before birth? The first identified mutation in RPS19 gene in a hydrops fetalis
L. Da Costa, G. Chanoz-Poulard, M. Simansour, M. Ffrench, R. Bouvier, F. Prieur, A L Delezoide, T. Leblanc, R. Touraine.

10:45 – 10:55 **Johnson Liu** - Ontogenetic and Mechanistic Differences in Two Murine ES Cell Models of Diamond Blackfan Anemia: Infantile Versus Fetal Anemia
Johnson M. Liu, MD, Tracie A. Goldberg, MD, Sharon Singh, MD, Adrianna Henson, Jonathan Solaimanzadeh, Steven R. Ellis, PhD, Jeffrey Michael Lipton, MD, PhD

11:00 – 11:15 *Coffee Break*

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Genetics

Moderators: Irma Dianzani/ David Bodine

- 11:15 – 11:25 **Introduction** – Mitchell Weiss
- 11:25 – 11:35 **Hanna Gazda** - Exome Sequencing Identifies *GATA1* Mutations Resulting in Diamond-Blackfan Anemia
Vijay G. Sankaran, Roxanne Ghazvinian, Ron Do, Prathapan Thiru, Jo-Anne Vergilio, Alan H. Beggs, Colin A. Sieff, Stuart H. Orkin, David G. Nathan, Eric S. Lander, Hanna T. Gazda
- 11:40 – 11:50 **Jason Farrar** - The Frequent Identification of Ribosomal Protein Gene Deletions in DBA Patients without Detectable RP Gene Mutations
Jason E. Farrar, Adrianna Vlachos, Eva Atsidaftos, Robert J. Arceci, Steven R. Ellis, Jeffrey M. Lipton, David M. Bodine
- 11:55 – 12:05 **Adrianna Vlachos** - 5q- Myelodysplastic Syndrome, In One of 66 Patients Lacking a Known Ribosomal Gene Mutation, Masquerading as Diamond Blackfan Anemia
Adrianna Vlachos, Jason Farrar, Eva Atsidaftos, Ellen Muir, Thomas C. Markello, Sharon Singh, Johnson M. Liu, Steven Ellis, Robert J. Arceci, David M. Bodine, Jeffrey M. Lipton
- 12:10 – 12:20 **Annarita Migliaccio** - Implications of the polymorphism of the human glucocorticoid receptor for the pathobiology of Diamond Blackfan Anemia
Lilian Varricchio, Barbara Ghinassi, Marieke von Lindern, Ugo Ramenghi and Anna Rita Migliaccio
- 12:25 – 12:35 **Marcin Wlodarski** - DBA in Germany: Comprehensive Genomic Characterization, Family Analysis and Genotype-Phenotype Correlation.
Marcin W Wlodarski, M.D., Jessica Moetter, Ph.D., Mutlu Kartal, M.D., Friedrich Kapp, Alexandra Fischer, Charlotte M Niemeyer and Joerg J Meerpohl, M.D. On behalf of the German DBA study group.
- 12:45 – 1:45 *Lunch*

Basic Mechanisms & Translation

Basic Mechanisms

Moderators: Jeffrey Lipton/ Harvey Lodish

- 1:50 – 2:00 **Teng Teng** - Aberrant cell cycle progression caused by mutation in RPL11: Implications for Diamond Blackfan Anemia
Teng Teng, Stefano Fumagalli, Carol. A. Mercer and George Thomas
- 2:05 – 2:15 **Alan Warren** - p53-dependent ribosomal stress signaling in the absence of MDM2
Shengjiang Tan, Li Jin, Alan J. Warren

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- 2:20 – 2:30 **Pierre-Emmanuel Gleizes** - Frameshift mutation in p53 regulator RPL26 is associated with multiple physical abnormalities and a specific pre-rRNA processing defect in Diamond-Blackfan anemia
Milena Preti, Mee-Rie Sheen, Marie-Françoise O'Donohue, Hanna Gazda, Pierre-Emmanuel Gleizes
- 2:35 – 2:45 **Fabrizio Loreni** – Analysis of the signaling pathways activated in response to ribosomal stress
Sara Caldarola, Gaia Lisi, Angelo Gismondi, Giada Juli, Fabrizio Loreni
- 2:50 – 3:00 *Coffee Break*

Translation

Moderators: Stefan Karlsson/H. Leighton Grimes

- 3:05 – 3:15 **Alison Taylor** - Calmodulin inhibition rescues ribosomal protein deficiency in zebrafish and cell models of DBA
Alison M. Taylor, David M. Raiser, Jessica M. Humphries, Benjamin L. Ebert, and Leonard I. Zon
- 3:20 – 3:30 **Lingbo Zhang** - Identification and functional characterization of an RNA binding protein that is essential for glucocorticoid- induced BFU-E self-renewal
Lingbo Zhang, Laboratory of Dr. Harvey F. Lodish, Whitehead Institute, Cambridge, MA
- 3:35 – 3:45 **Jan Abkowitz** - Does heme excess contribute to the pathogenesis of Diamond Blackfan Anemia?
Janis L Abkowitz. University of Washington, Seattle, WA.
- 3:50 – 4:00 **Adrianna Vlachos** - The Incidence of Neoplasia in Diamond Blackfan Anemia: A Report from the Diamond Blackfan Anemia Registry
Adrianna Vlachos, Philip S. Rosenberg, Eva Atsidaftos, Blanche P. Alter, Jeffrey M. Lipton
- 4:05 – 4:50 Wrap-Up:

Developmental Biology

Moderators: Mitchell Weiss/ George Thomas

Genetics

Moderators: Irma Dianzani/ David Bodine

Basic Mechanisms & Translation

Moderators: Jeffrey Lipton/ Harvey Lodish

Moderators: Stefan Karlsson/H. Leighton Grimes

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5:00 – 6:00 **POSTER SESSION**

Moderators: Blanche Alter/ Jan Abkowitz

Nadia Danilova - Beneficial effects of nucleoside treatment in zebrafish models of Diamond Blackfan Anemia deficient in ribosomal proteins L11 or S19

Nadia Danilova, Kathleen M. Sakamoto and Shuo Lin

Francisca Aidoo - A potential role for serine-arginine rich splicing factors in the erythropoietic defect observed in Diamond Blackfan Anemia.

Francisca Aidoo, Abigail Fernandes, Yigal Dror and Chetan Tailor

Wenqian Hu - Long non-coding RNA mediated anti-apoptotic activity in murine erythroid terminal differentiation

Wenqian Hu, Bingbing Yuan, Johan Flygare, and Harvey F. Lodish, Whitehead Institute for Biomedical Research

Jonathan Warner - WHY S19?

Varun Gupta and Jonathan R. Warner, Department of Cell Biology, Albert Einstein College of Medicine

Elena Bibikova - Aberrant miR34a Regulation in RPS19 Insufficient Hematopoietic Stem Cells

Elena Bibikova, Shuo Lin, Ph.D., and Kathleen Sakamoto, M.D, Ph.D.

Chetankumar Tailor - Aberrant splicing of FLVCR1 transcript and disrupted erythropoiesis is specific to Diamond Blackfan Anemia and not Shwachman Diamond Syndrome, and implications that RPS17 may not be responsible for erythroid failure in DBA

Abigail Fernandes, Francisca Aidoo, Yigal Dror and Chetan Tailor

Sarah Bray - The role of Myb in Diamond Blackfan Anaemia

Sarah C. Bray, Michelle Perugini, Amilia Wee, Louise Winteringham, L. Bik To, Heather Tapp, Thomas J. Gonda, S. Peter Klinken, Richard D'Andrea

Pekka Jakko - Bone Marrow Failure in RPS19-deficient Mice Responds to L-Leucine Treatment

Pekka Jaako, Shubhranshu Debnath, Karin Olsson, Johan Flygare, Stefan Karlsson

Akiko Shimamura – RPS19 haploinsufficiency activates p53 without RPL11 upregulation in human hematopoietic cells

Hong-Yan Du, Blanche P Alter, Tarek Elghetany, Angela D. Trobaugh-Lotrario, and Akiko Shimamura

Marieke von Lindern - BAG1 AND CSDE1, Two Proteins Suppressed in DBA, Are Important for Erythropoiesis

Rastislav Horos, Dagmar Pospisilova, Regine Sendtner, Michael Sendtner, Marieke von Lindern

Paola Quarello - High frequency of ribosomal protein gene copy number variations in Italian Diamond Blackfan anemia patients detected by MLPA assay

Paola Quarello, Emanuela Garelli, Alfredo Brusco, Adriana Carando, Cecilia Mancini, Patrizia Pappi, Irma Dianzani, Ugo Ramenghi

Elizabeth Chao - The Ambry Genetics Experience in Molecular Diagnostic Testing for Diamond Blackfan Anemia: Our first 200 families

Elizabeth Chao, Steven Keiles

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POSTER SESSION Cont'd

Irma Dianzani - Dissecting the transcriptional phenotype of ribosomal protein deficiency: implications for Diamond-Blackfan Anemia.

A.Aspesi, E.Pavesi, E.Robotti, P.Roncaglia, R.Crescitelli, F.Avondo, I.Boria, L.Da Costa, H.Moniz, N.Mohandas, U.Ramenghi, A.Ronchi, F.Loreni, A.Follenzi, E.Marengo, S.R.Ellis, S.Gustincich, C.Santoro, I.Dianzani

John Fargo - Adenosine Deaminase: How Good is it for the Diagnosis of Diamond-Blackfan Anemia?

John H. Fargo, Christian P. Kratz, Neelam Giri, Sharon A. Savage, Carolyn Wong, Karen Backer, Blanche P. Alter, and Bertil E. Glader

Thierry LeBlanc - Does Diamond-Blackfan anemia phenotype in adulthood include B-cell immunodeficiency?

Thierry LEBLANC, Monique FORVEILLE, Djaouida BENGOUFA, Isabelle MARIE, Lydie DA COSTA, Anne DURANDY

Surveillance of Patients with Diamond Blackfan Anemia in the United States 2011 – A CDC Project

Rogers ZR, Mouradian K, Boger D, Hulihan M, Grant A, and Glader B

University of Texas Southwestern Dallas TX, Stanford University Palo Alto CA, Centers for Disease Control Atlanta GA.

Lee Grimes - microRNA antagonists block acute myeloid leukemia stem cell activity

H. Leighton Grimes, Chinavenmeni S Velu, Aditya Chaubey, Shane Horman, Balkrishen Bhat, Craig Jordan, James Phelan, Bruce Aronow, Anil Jegga, Monica Guzman, Jianjun Chen, Nancy Zeleznik-Le, Brian Gebelein

Dagmar Pospisilova – Effects of an L-Leucine Administration in Diamond-Blackfan Anemia – An update from the Czech Diamond Blackfan Anemia registry

Dagmar Pospisilova, Jana Cmejlova, Tomas Adam, Radek Cmejla, Jiri Hak

Josu de la Fuente - Iron Load Can Be Severe and Presents Early in DBA Patients Even When Receiving Adequate Chelation Treatment: Need for Integration of Liver Biopsy and MR techniques

Josu de la Fuente, Yvonne Harrington, Sonia Bonanomi

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

MONDAY MARCH 19th

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

9:00 – 10:00 Poster Wrap-Up

Clinical Treatment & Drug Development

Moderators: Monica Bessler/ Lydie DaCosta

- 10:00 – 10:10 **Anu Narla** - L-Leucine improves the anemia and developmental defects associated with Diamond Blackfan Anemia (DBA) and MDS del(5q) by activating the mTOR pathway
Elspeth Payne, MB, ChB, PhD, Maria Virgilio, BS, Anupama Narla, MD, Hong Sun, MD, Michelle Levine, BS, Barry H Paw, MD, Ph.D, A Thomas Look, MD, Benjamin Ebert, MD, DSc, Arati Khanna-Gupta, Ph.D
- 10:15 – 10:25 **Sara Sjögren** - Towards Mechanism-Based Treatment for Diamond-Blackfan Anemia
Sara E Sjögren, Abdul G Alattar, Alexander Mattebo, Pekka Jaako, John G Doench, Stefan Karlsson, David E Root and Johan Flygare.
- 10:30 – 10:40 **Johnson Liu** - Sotatercept in Diamond Blackfan Anemia
Johnson Liu, Adrianna Vlachos, Jeffrey M. Lipton, Ellen Muir, Eva Atsidaftos, Abderrahmane Laadem, Victoria Sung, Robert Peter Gale
- 10:45 – 10:55 **Carol Mercer** - Pifithrin, a small molecule inhibitor of p53, alleviates anemia in an RPS6 mouse model of DBA and 5q- syndrome
Carol A Mercer, Vasily Ivanenkov, Fukun Guo, Yi Zheng, Stefano Fumagalli, George Thomas
- 11:00 – 11:10 *Coffee Break*
- 11:15 - 11:45 Wrap-up:

Clinical Treatment & Drug Development

Moderators: Monica Bessler/ Lydie DaCosta

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- 11:50 – 12:00 Update from the NHLBI
Pankaj Qasba, Program Director
Blood Diseases Branch
Division of Blood Diseases and Resources
National Heart, Lung, and Blood Institute, NIH
- 12:00 – 12:10 Update from Telemedicine & Advanced Technology Research Center (TATRC)
Dr. Charles Peterson
Henry M. Jackson Foundation for the Advancement of Military Medicine
Telemedicine & Advanced Technology Research Center (TATRC)
- 12:15 – 12:30 Closing Remarks
Marie and Manny Arturi

Lunch & Departure

Supported by conference grants from:

