13th Diamond Blackfan Anemia International Consensus Conference

Sponsored by:

DIAMOND BLACKFAN ANEMIA FOUNDATION, INC.

SATURDAY, MARCH 8th

6:30PM  
2014 Kick-Off Dinner ~ The Plaza Ballroom (Second Floor)  
Welcome:  Dawn M. Baumgardner

SUNDAY MARCH 9th

7:30 – 8:30  
Breakfast Buffet ~ 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 – 9:10  
Overview of DBA  
Jeffrey Lipton

Emerging Therapies  
Moderators: Jeffrey Lipton / Johan Flygare

9:20 – 9:30  
Synergism between PPARα and glucocorticoid receptor signaling promotes self-renewal of human erythroid progenitors  
Hsiang-Ying Lee, Xiaofei Gao, Inma Barrasa, Harvey F. Lodish

9:30 – 9:40  
High-throughput screening identifies many novel potential therapies for Diamond-Blackfan Anemia  
Lingbo Zhang, Lina Prak, Sherry Lee, Xiaofei Guo, Shilpa Hattangadi, and Harvey Lodish

9:50 – 10:00  
Calmodulin inhibition rescues the effects of ribosomal protein deficiency in Diamond Blackfan anemia models  
Elizabeth R. Macari, Alison M. Taylor, David M. Raiser, Kavitha Siva, Jessica M. Humphries, Katherine McGrath, Johan Flygare, Benjamin L. Ebert and Leonard I. Zon
The Australian Diamond-Blackfan Anaemia Program: a multi-centered approach to discover treatments for Diamond-Blackfan anaemia and other rare bone marrow failure syndromes

Coffee Break

Model Systems of DBA
Moderators: Mitch Weiss / Stefan Karlsson

iPS cell lines from Diamond Blackfan Anemia (DBA) patients with RPS19 or RPL5 mutation show differences in ribosome biogenesis and TGF beta signaling
Jingping Ge, Marisa Apicella, Shwetha H. Manjunath, Jason A. Mills, Loïc Garçon, Deborah L. French, Mitchell J. Weiss, Monica Bessler, and Philip J. Mason

Gene therapy corrects the anemia and lethal bone marrow failure in mouse model for RPS19-deficient Diamond-Blackfan anemia
Shubhranshu Debnath, Pekka Jaako, Karin Olsson, Ute Modlich, Michael Rothe, Kavitha Siva, Axel Schambach, H. Bobby Gaspar, Johan Flygare and Stefan Karlsson

RAP-011 efficiently rescues erythropoiesis in zebrafish models of Diamond Blackfan Anemia
Jason Ear, Haigen Huang, Zahra Tehrani, Victoria Sung, Thomas O. Daniel, Rajesh Chopra, and Shuo Lin

Pathogenic Mechanisms
Moderators: Robert Arceci / Pierre-Emmanuel Gleizes

Relevance of RPL11-mediated inhibition of Mdm2 in the stabilization of p53 in an RPS6 mouse model of DBA and 5q- syndrome
Carol Mercer, Teng Teng, Stefano Fumagalli, Fukun Guo, Sara C. Kozma, Yi Zheng, George Thomas

Insights into the pathogenesis of Diamond-Blackfan anemia
Janis Abkowitz, Keel SB, Gerds A, Scott B, Shimamura A, and Yang Z
The RP-Mdm2 interaction underlies the p53-mediated component of the erythroid defect in a mouse model for Diamond-Blackfan anemia

Pekka Jaako, Shubhranshu Debnath, Karin Olsson, Mikael S. Lindström and Stefan Karlsson

The 5S RNP Couples p53 Homeostasis to Ribosome Biogenesis and Nucleolar Stress

Katherine Elizabeth Sloan, Loren Macdonald, Zhao Zhao and Nicholas James Watkins

Lunch - Grand Ballroom – Salons III and IV (Lower Level)

Pathogenic Mechanisms and the Ribosome

Moderators: Arnold Levine / Marieke von Lindern

Analysis the p53 pathway in RPL5/RPL11 deficient erythroid progenitors

Teng Teng, Kelly O’Brien, Jiang Wang, Punam Malik, Adrianna Vlachos, Jeffery M. Lipton, Sara C. Kozma, Carol A. Mercer, David M. Bodine, George Thomas

Molecular mechanisms of ITS1 processing in human cells, a pre-ribosomal RNA maturation step often affected in DBA

Marie-Françoise O'Donohue, Milena Preti, Nathalie Montel-Lehry, Marie-Line Cavaillé-Bortolin, Pierre-Emmanuel Gleizes

A ubiquitin-conjugating enzyme critical for terminal erythroid differentiation: possible implications for Diamond-Blackfan anemia

Anthony Nguyen, Miguel A. Prado, Yuan Shi, Verena Dederer, Paul Schmidt, Dean Campagna, Steven P. Gygi, Mark D. Fleming, and Daniel Finley

GATA1 and DBA

Moderators: David Nathan / Irma Dianzani

Stage selective requirement for the N-terminus of GATA1 during erythropoiesis

Monika Stankiewicz, Timothy Chlon and John D. Crispino

Roles for the GATA1 amino terminus in erythroid differentiation revealed through patient-derived pluripotent stem cells

2:30 – 2:40  The Role of GATA1 in Diamond-Blackfan Anemia  

2:45 – 2:55  Update from the NIDDK/NIH  
Dr. Terry Bishop  
Hematology Program Director  
National Institute of Diabetes and Digestive and Kidney Diseases, NIH

2:55 – 3:05  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

3:05 – 3:15  Coffee Break

3:15 – 3:45  Poster Viewing

Poster Session 1  
3:45 – 5:15  Facilitator: David Bodine / Anu Narla

Patient-derived induced pluripotent stem cells recapitulate the pathophysiology and variable penetrance of DBA  

Gene correction and induced pluripotent stem cell production using DBA fibroblasts  
Erik Westin, Chao Li, Tim Townes, Frederick Goldman

Ginger can stimulate blood cell formation in zebrafish embryos  
Karine F. Ferri-Lagneau, Shengmin Sang, TinChung Leung

L-Leucine improves the anemia of DBA via activation of the mTOR pathway in a p53-independent manner  
Nirmalee Abayasekara, Anupama Narla, Slater Hurst, Hong Sun, Elspeth Payne, Benjamin Ebert, A. Thomas Look, Barry H. Paw, Nancy Berliner, Arati Khanna-Gupta

The downstream effects of RP gene mutations  
Alyson MacInnes

Analyzing the zebrafish model of Diamond Blackfan anemia  
Tamayo Uechi, Yukari Nakajima, Gnaneshwar V. Yadav, Yutaka Suzuki, Sumio Sugano, and Naoya Kenmochi
Defective ribosomal RNA processing in Diamond-Blackfan anemia patients: Potential molecular biomarkers for diagnosis?

Anirban Chakraborty, Marie-Françoise O’Donohue, Hanna T. Gazda, and Pierre-Emmanuel Gleizes

A systematic loss-of-function analysis of zebrafish bystin-like (bysl) mutant: Could pre-ribosomal factors be potential DBA-associated genes?

Anirban Chakraborty, Pascale Dufourcq, Patrick Blader, and Pierre-Emmanuel Gleizes

BMI1 regulates ribosome biogenesis in erythroid progenitor cells

Rui Gao, Sisi Chen, Michihiro Kobayashi, Hao Yu, Sara K. Young, Anthony Soltis, Yingchi Zhang, Yang Wan, Sasidhar Vemula, Ernest Fraenkel, Alan Cantor, Yang Xu, Mervin C. Yoder, Reuben Kapur, Ronald C. Wek, Steven R. Ellis, Xiaofan Zhu, Yan Liu

Erythroid and Osteogenic Differentiation Defects in Rps19- and Rpl5-Mutant Embryonic Stem Cells: Evidence Against a Specific Role of p53

Sharon A. Singh, Brian M. Dulmovits, Tracie Goldberg, Jeffrey M. Lipton, Lionel Blanc, Johnson M. Liu

Ribosomal Stress Activates a Complex Response Involving p53, AMPK and PIM1 Kinase

Vinay Sagar, Sara Caldarola, Angelo Gismondi, Giada Juli, Lidia Chellini, Valentina Iadevaia, Christopher G. Proud, Fabrizio Lorenzi

Conditional targeting of murine Rpl11 during embryogenesis results in fetal loss.

Meagan McGuinness, Minh Nguyen, Yuko Fujiwara and Colin Sieff

Increased Tumorigenesis in Ribosomal Proteins L5 and S24 Heterozygous Mice

Shideh Kazeroonian, Pedro Ciarnlini, Roxanne Ghazvinian, Meritxell Alberich-Jorda, Daniel Yuan, Mugdha Joshi, Hong Zhang, Alan H. Beggs and Hanna T. Gazda

Transcriptional divergence and conservation of human and mouse erythropoiesis

Novalia Pishesha, Prathapan Thiru, Jiahai Shi, Jennifer C. Eng, Vijay G. Sankaran and Harvey F. Lodish

A Mouse Model Depicting the Role of Gata1 N-Terminal in Fetal Erythropoiesis – Relevance to Diamond Blackfan Anemia

Yehudit Birger, Liat Goldberg, Timothy M. Chlon, Inna Muler, Ginette Schiby, Jasmin Jacob-Hirsch, Gideon Rechavi, John D. Crispino and Shai Izraeli

Dosage-dependent GATA1 activity in human hematopoiesis and Diamond-Blackfan anemia

Leif S. Ludwig, Jacob Ulirsch, Aoi Wakabayashi, Harvey Lodish, Hanna T. Gazda, and Vijay G. Sankaran

Selective mRNA translation in erythropoiesis

Klaske Thiadens, Eleonora de Klerk, Emile van den Akker, Kat Moore, Peter-Bram ’t Hoen, Marieke von Lindern

Dysferlin and other non-red cell proteins accumulate in the red cell membrane of Diamond-Blackfan Anemia patients

Esther N. Pesciotta, Sira Sriswasdi, Hsin-Yao Tang, David W. Speicher, Philip J. Mason, Monica Bessler

6:00 – 9:00 Cocktails Reception ~ The Plaza Ballroom (Second Floor)
**MONDAY MARCH 10th**

7:30 - 8:30  **Breakfast Buffet − Grand Ballroom − Salons III and IV (Lower Level)**

**Gene Discovery**  
_Moderators: Monica Bessler / Akiko Shimamura_

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| 8:30 – 8:40 | Autosomal recessive Diamond-Blackfan anemia: Identification of mutations in MCM2 and FLNB  
| 8:45 – 8:55 | Novel genes modulating erythroid proliferation in a mouse model for Diamond-Blackfan Anemia  
*Sara E Sjögren*, Pekka Jaako, John G Doench, Stefan Karlsson, David E Root and *Johan Flygare* |

**Clinical Aspects of DBA**  
_Moderators: Monica Bessler / Akiko Shimamura_

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| 9:00 – 9:10 | Patients with Diamond Blackfan Anaemia have Abnormalities of Cellular and Humoral Immunity  
*Deena Iskander*, Yvonne Harrington, Gareth Gerrard, Alex en Foong, Letizia Foroni, Irene Roberts, Anastasios Karadimitris and Josu de la Fuente |
| 9:15 – 9:25 | Loss of GATA-1 Full Length (GATA-1 FL) as a cause of Diamond-Blackfan anemia (DBA) phenotype  
*Sara Parrella*, Anna Aspesi, Paola Quarelllo, Emanuela Garelli, Elisa Pavesi, Adriana Carando, Margherita Nardi, Steven Ellis, Ugo Ramenghi, *Irma Dianzani* |
| 9:30 – 9:40 | Regulation of Iron Metabolism in Diamond-Blackfan Anemia  
*Dagmar Pospisilova*, D. Holub, Z. Zidova, B. Ludikova, L. Sulovska, V. Divoky, M. Horvathova |
| 9:45 – 9:55 | What is “MDS” in Diamond-Blackfan Anemia and Other Inherited Bone Marrow Failure Syndromes  
*Blanche Alter*, Tarek M Elghetany, Neelam Giri, Sharon Savage, Diane Arthur, Irina Marie |
10:00 – 10:10 Elevated Erythrocyte Adenosine Deaminase (eADA) Activity as a Marker for Diamond Blackfan Anemia in Patients with Congenital Heart Disease
**Adrianna Vlachos**, Diana S. Osorio, Evangelia Atsidaftos, Ellen Muir, Howard Seiden, Dorota Gruber, Jason E. Farrar, Lionel Blanc, Robert Arceci, Steven Ellis, Bertil Glader, David M. Bodine, Jeffrey M. Lipton

10:15 – 10:35 *Coffee Break* (Hotel Check-out and Luggage storage)

10:35 – 11:00 Poster Viewing

**Poster Session 2**
11:00 – 12:00 *Facilitators: Bert Glader / Lionel Blanc*

The application of high-throughput targeted genomics for the molecular characterisation of Diamond-Blackfan anaemia: a cohort study
Gareth Gerrard, Hui-En Foong, Deena Iskander, Mikel Valganon, Dalia Kasperaviciute, Michael Muller, Yvonne Harrington, Irene Roberts, Anastasios Karadimitris, Letizia Foroni, *Josu de la Fuente*

A Novel Deletion Involving RPL31 in a Child with Diamond Blackfan Anemia
**Jason E. Farrar**, Ross Fisher, Adrianna Vlachos, Eva Atsidaftos, Ellen Muir, Robert Arceci, Jeffrey M. Lipton, David M. Bodine, Steven Ellis

Genetics of Diamond-Blackfan anemia in the National Cancer Institute IBMFS Cohort: new and old ribosomal genes
**Lisa Mirabello**, Neelam Giri, Meredith Yeager, Joseph F. Boland, Ji He, Belynda D. Hicks, Laurie Burdett, Sharon A. Savage, Blanche P. Alter

Audiologic and Otologic Manifestations in Diamond-Blackfan Anemia
**Neelam Giri**, Adedoyin O. Kalejaiye, Charleen Adams, Carmen C. Brewer, Christopher K. Zalewski, Kelly King, H. Jeffrey Kim, Blanche P. Alter

Multiplexed genetic evaluation of inherited bone marrow failure and myelodysplastic syndromes
Michael Y. Zhang, Siobán B. Keel, Tom Walsh, Ming K. Lee, Colin C. Pritchard, Amanda C. Watts, Michael R. Jeng, Inga Hofmann, David A. Williams, Mark D. Fleming, Janis L. Abkowitz, Mary-Claire King, and **Akiko Shimamura**

Burden of Pearson marrow pancreas syndrome in a cohort of Diamond Blackfan anemia patients
Katelyn E. Gagne, Roxanne Ghazvinian, Daniel Yuan, Rebecca L. Zon, Kelsie Storm, Magdalena Mazur-Popinska, Laura Andolina, Halina Bubala, Sydonia Golebiowska, Meghan A. Higman, Krzysztof Kalwak, Peter Kurre, Michal Matysiak, Edyta Niewiadowska, Salley Pels, Mary Jane Petruzzi, Aneta Pobudejska-Pientazek, Tomasz Szczepanski, Mark D. Fleming, Hanna T. Gazda and **Suneet Agarwal**

Chelation therapy in Diamond-Blackfan anemia patients: a French survey.
**Thierry LeBlanc**, Isabelle Marie, Lydie Da Costa
Hematopoietic Stem Cell Transplantation as a Feasible Curative Option in Diamond Blackfan Anemia? - An analysis from the German and Italian registries

**Ina Hainmann**, MD, Marcin Wlodarski, MD, Brigite Strahm, MD, Friedrich Kapp, MD, Ugo Ramenghi, MD, Joerg Meerpohl, MD, Franco Locatelli, MD, Charlotte Niemeyer, MD. On behalf of the German and Italian DBA Registries.

Haematopoietic stem cell transplantation for Diamond Blackfan anaemia: a report from the Italian Association of Paediatric Haematology and Oncology Registry

Franca Fagioli, **Paola Quarello**, Marco Zecca, Edoardo Lanino, Paola Corti, Claudio Favre, Mimmo Ripaldi, Ugo Ramenghi, Franco Locatelli, Arcangelo Prete

Diamond Blackfan anemia diagnosis in Italian patients: data from the Italian registry

**Ugo Ramenghi**, Quarello P, Garelli E, Carando A, Botto C, Brusco A, Dianzani I

Genotyping Strategies for Diamond Blackfan Anemia Patients in Canada

Bozana Zlateska B, Hongbing Li, Ibrahim Ghemlas, Robert Klaassen, Conrad V Fernandez, Rochelle Yanofsky, John Wu, Yves Pastore, Mariana Silva, Jeff H Lipton, Jossee Brossard, Bruno Michon, Sharon Abish, McGregor Steele, Roona Sinha, Mark Belletrutti, Vicky Breakey, Lawrence Jardine, Lisa Goodyear, Amanda Wagner, Michaela Cada, **Yigal Dror**

Phenotypic and genotypic characteristics of Diamond Blackfan Anemia patients in Israel

Tracie Goldberg, Joanne Yacobovich, Orly Dgany, Tanya Krasnov, Shoshana Ravel-Vilk, Chaim Kaplinsky, Ayelet Ben Barak, Ronit Elhasid, Joseph Kapelusnik, Ami Ballin8, Hagit Miskin, Amir Kuperman, **Hannah Tamary**

12:00 – 12:15  Closing Remarks  
**Steve Ellis**

12:15 – 12:30  Farewell  
**Diamond Blackfan Anemia Foundation**

12:30  **Lunch & Departure**