

AGENDA

Thursday – October 21, 2010

3:00 **Registration opens**

4:00 - 6:00

FA 101: An Introduction to the Medicine and Biology of Fanconi Anemia
Ballrooms 1 & 2

This session is intended especially for those new to Fanconi anemia research and clinical care. However, all attendees interested in an overview of unanswered questions, new research directions and resources to support research on Fanconi anemia are encouraged to attend.

Introductions:

Hans Joenje, PhD

Scientific Advisory Board, Fanconi Anemia Research Fund
Vrije Universiteit Medical Center, Amsterdam, The Netherlands

Faculty:

Blanche Alter, MD, MPH

National Cancer Institute, Rockville, Maryland

Stephen Meyn, MD, PhD

Scientific Advisory Board, Fanconi Anemia Research Fund
The Hospital for Sick Children, Toronto, Canada

6:00 - 8:00

Welcome Reception
Poster Viewing
Ballroom 3

Friday – October 22, 2010

7:00 - 8:00

Buffet Breakfast
Ballroom 4
Poster Viewing
Ballroom 3

Plenary Session
Ballrooms 1 and 2

- 8:00 - 8:15 **Welcome**
David Frohnmayer
Co-founder and Vice President, Board of Directors,
Fanconi Anemia Research Fund
Eugene, Oregon
- 8:15 - 8:30 **Overview**
Grover C. Bagby, Jr., MD
Chair, Scientific Advisory Board, Fanconi Anemia Research Fund
Oregon Health & Science University
Portland, Oregon
- 8:30 - 9:00 **Special Session A: Dyskeratosis Congenita**
Inderjeet Dokal, MD (*for more information, see page 10*)
Blizzard Institute of Cell and Molecular Science
Barts and The London School of Medicine and Dentistry
London, United Kingdom
- 9:00 - 9:15 Questions and Answers
- 9:15 - 9:30 **Meeting Report: Squamous Cell Carcinoma in Fanconi Anemia Patients**
Ruud Brakenhoff, PhD
Vrije Universiteit Medical Center, Amsterdam, The Netherlands
- Session I: Carcinogenesis**
Chair: Ruud Brakenhoff, PhD
Vrije Universiteit Medical Center
Amsterdam, The Netherlands
- 9:30 - 9:35 **Session Overview: Ruud Brakenhoff, PhD**
- 9:35 - 9:45 **Elizabeth Hoskins, MS**, Cincinnati Children's Hospital Medical Center,
Cincinnati, Ohio: *Oropharyngeal HPV Prevalence in Individuals with FA*
- 9:45 - 9:50 Questions and Answers
- 9:50 - 10:00 **Eunike Velleuer, MD**, Heinrich Heine University of Duesseldorf, Duesseldorf,
Germany: *Lessons from the Field: Towards Stringent Oral Cancer Screening in FA Patients*
- 10:00 - 10:05 Questions and Answers
- 10:05 - 10:20 **Break**
- 10:20 - 10:30 **Holger Tönnies, PhD**, Robert Koch-Institut, Berlin, Germany: *Detailed
Characterization of FA and Non-FA HNSCC Using Molecular Cytogenetics, Array CGH
and Methylation Assays*
- 10:30 - 10:35 Questions and Answers

22nd Annual Fanconi Anemia Research Fund Scientific Symposium

- 10:35 - 10:45 **Ian Mackenzie, DDS, PhD**, Blizard Institute of Cell and Molecular Science, Queen Mary University of London, London, United Kingdom: *Properties of Stem Cells in Cell Lines Derived from Head and Neck Cancers of Fanconi Anemia Patients*
- 10:45 - 10:50 Questions and Answers
- 10:50 - 10:55 **Session Wrap-up: Ruud Brakenhoff, PhD**

Session II: New Genes

Chair: Hans Joenje, PhD
Vrije Universiteit Medical Center
Amsterdam, The Netherlands

- 10:55 - 11:00 **Session Overview: Hans Joenje, PhD**
- 11:00 - 11:10 **Zhijiang Yan, PhD**, National Institute on Aging, Baltimore, Maryland: *FAAP20 is Required for Normal Activation of the Fanconi Anemia-associated DNA Damage Response Pathway*
- 11:10 - 11:15 Questions and Answers
- 11:15 - 11:25 **Abdullah Ali, PhD**, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio: *FAAP20: A Novel FA Core Complex Binding Protein is Required for the Functional Integrity of FANC-BRCA Pathway*
- 11:25 - 11:30 Questions and Answers
- 11:30 - 11:40 **Helmut Hanenberg, MD**, Riley Hospital for Children, Indianapolis, Indiana: *Germ-line Mutations in Breast and Ovarian Cancer Pedigrees Establish RAD51C as a Human Cancer Susceptibility Gene*
- 11:40 - 11:45 Questions and Answers
- 11:45 - 11:55 **Christopher Mathew, PhD**, King's College London Medical School, London, United Kingdom: *Mutation of the RAD51C Gene in a Fanconi Anemia-like Disorder*
- 11:55 - 12:00 Questions and Answers

12:00 - 1:00

Hosted Lunch
Ballroom 4
Poster Viewing
Ballroom 3

- 1:00 - 1:10 **Benjamin Chen, PhD**, University of Texas Southwestern Medical Center, Dallas, Texas: *DNA-PKcs-3A Mutant Mice, an Authentic or Imitated Mouse Model of Fanconi Anemia?*
- 1:10 - 1:15 Questions and Answers
- 1:15 - 1:25 **Johan de Winter, PhD**, Vrije Universiteit Medical Center, Amsterdam, The Netherlands: *Warsaw Breakage Syndrome, a Fanconi Anemia-like Disorder Associated with Mutations in the FANCI-like Helicase DDX11*
- 1:25 - 1:30 Questions and Answers

- 1:30 - 1:40 **Chantal Stoepker, MSc**, Vrije Universiteit Medical Center, Amsterdam, The Netherlands: *SLX4, a Coordinator of Structure-specific Endonucleases, is Mutated in a New Fanconi Anemia Subtype*
- 1:40 - 1:45 Questions and Answers
- 1:45 - 1:55 **Yonghwan Kim, PhD**, The Rockefeller University, New York, New York: *Identification of SLX4 as a Fanconi Anemia Gene*
- 1:55 - 2:00 Questions and Answers
- 2:00 - 2:05 **Session Wrap-up: Hans Joenje, PhD**

Session III: Nuclear Crosslink Metabolism
Chair: Stephen Meyn, MD, PhD
Scientific Advisory Board, Fanconi Anemia Research Fund
The Hospital for Sick Children
Toronto, Canada

- 2:05 - 2:10 **Session Overview: Stephen Meyn, MD, PhD**
- 2:10 - 2:20 **Agata Smogorzewska, MD, PhD**, The Rockefeller University, New York, New York: *A Genetic Screen Identifies FAN1, a Fanconi Anemia Associated Nuclease Necessary for DNA Interstrand Crosslink Repair*
- 2:20 - 2:25 Questions and Answers
- 2:25 - 2:35 **Philip Connell, MD**, University of Chicago, Chicago, Illinois: *Chemical Stimulation of Homologous Recombinational DNA Repair in Fanconi Anemia*
- 2:35 - 2:40 Questions and Answers
- 2:40 - 2:50 **Jen-Wei Huang, Graduate Student**, Fred Hutchinson Cancer Research Center, Seattle, Washington: *The MicroRNAs miR-103 and miR-107 Regulate Homologous Recombination and Cellular Sensitivity to DNA Damaging Agents*
- 2:50 - 2:55 Questions and Answers
- 2:55 - 3:10 **Break**
- 3:10 - 3:20 **Barbara Godthelp, PhD**, Leiden University Medical Center, Leiden, The Netherlands: *Spatio-temporal Organization of DNA Interstrand Crosslink Repair at Sites of ICL in G1 and S-phase Fibroblasts*
- 3:20 - 3:25 Questions and Answers
- 3:25 - 3:35 **Katharina Schlacher, PhD**, Memorial Sloan-Kettering Cancer Center, New York, New York: *BRCA2/FANCD1 Protects Stalled Forks against 3'-5' Degradation by MRE11 via Conserved Cter RAD51 Domain Independent of H*
- 3:35 - 3:40 Questions and Answers
- 3:40 - 3:50 **Johannes Walter, PhD**, Harvard Medical School, Boston, Massachusetts: *A Comprehensive Model for Replication-coupled DNA Interstrand Crosslink Repair*
- 3:50 - 3:55 Questions and Answers

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- 3:55 - 4:05 **Jean Gautier, PhD**, Columbia University Medical School, New York, New York:
Fanconi Anemia Proteins and the Repair of Interstrand Crosslinks
- 4:05 - 4:10 Questions and Answers
- 4:10 - 4:20 **Michelle Duquette, PhD**, University of California, San Diego, La Jolla, California:
CtIP is Required for FANCD2 Recruitment to Interstrand Cross-links
- 4:20 - 4:25 Questions and Answers
- 4:25 - 4:35 **Nicole Persky**, Memorial Sloan-Kettering Cancer Center, New York, New York:
Insights into the Fanconi Anemia DNA Repair Pathway from the Structure of the FANCI-FANCD2 Complex
- 4:35 - 4:40 Questions and Answers
- 4:40 - 4:50 **Michael Seidman, PhD**, National Institute on Aging, Baltimore, Maryland:
Temporal Distinctions in the Recruitment of FancD2 to Double Strand Breaks and Interstrand Crosslinks
- 4:50 - 4:55 Questions and Answers
- 4:55 - 5:00 **Session Wrap-up: Stephen Meyn, MD, PhD**

Saturday – October 23, 2010

7:00 - 8:00

Buffet Breakfast
Ballroom 4
Poster Viewing
Ballroom 3

8:00 - 8:30

The following session is sponsored by the NIAID Radiation Countermeasures Program

Special Session B: Radiation Mitigation and Hematopoiesis

Joel Greenberger, MD (for more information, see page 11)

University of Pittsburgh Cancer Institute

Pittsburgh, Pennsylvania

8:30 - 8:45

Questions and Answers

Session IV:

Transplantation

Chair: John Wagner, MD

University of Minnesota

Minneapolis, Minnesota

8:45 - 8:50

Session Overview: John Wagner, MD

8:50 - 9:00

Elisabeth Korthof, MD, Leiden University Medical Center, Leiden, The Netherlands: *Mismatched Family Donor Stem Cell Transplantation as First Transplantation for Fanconi Anemia: A Survey of the EBMT Work*

9:00 - 9:05

Questions and Answers

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- 9:05 - 9:15 **Margaret MacMillan, MD**, University of Minnesota, Minneapolis, Minnesota:
Alternative Donor Hematopoietic Cell Transplantation in 94 Patients with Fanconi Anemia at the University of Minnesota
- 9:15 - 9:20 Questions and Answers
- 9:20 - 9:25 **Abstract Wrap-up and Introduction to Panel: John Wagner, MD**
- 9:25 - 9:40 **Break**
- Transplantation Center Reports*
- 9:40 - 9:50 **John Wagner, MD**, University of Minnesota, Minneapolis, Minnesota
- 9:50 - 10:00 **Farid Boulad, MD**, Memorial Sloan-Kettering Cancer Center, New York, New York
- 10:00 - 10:10 **Wolfram Ebell, MD**, Charite-Universitaets Medizin Berlin, Berlin, Germany
- 10:10 - 10:20 **Carlo Dufour, MD**, G. Gaslini Children's Hospital, Genova, Italy
- 10:20 - 10:30 **Carmem Bonfim, MD**, Federal University of Parana, Curitiba, Brazil
- 10:30 - 10:50 Questions and Answers
- 10:55 - 11:25 *Special Session C: Gene Rx and Homologous Recombination*
J. Keith Joung, MD, PhD (*for more information, see page 12*)
Massachusetts General Hospital
Charlestown, Massachusetts
- 11:25 - 11:40 Questions and Answers
- 11:40 - 1:40 **Poster Viewing**
Ballroom 3
- 11:40 - 1:40 **Joint Meeting and Lunch**
Board of Directors and Scientific Advisory Board
Fanconi Anemia Research Fund
Deer Lake/Elk Lake Rooms
- 12:00 - 1:00 **Buffet Lunch**
Ballroom 4

Session V: Stem Cells, Germ Cells and Signaling
Chair: Madeleine Carreau, PhD
Laval University
Québec, Canada

- 1:40 - 1:45 **Session Overview: Madeleine Carreau, PhD**
- 1:45 - 1:55 **Jakub Tolar, MD, PhD**, University of Minnesota, Minneapolis, Minnesota: *Gene Editing of Induced Pluripotent Stem Cells from Patients with Fanconi Anemia*
- 1:55 - 2:00 Questions and Answers
- 2:00 - 2:10 **Jennifer Adair, PhD**, Fred Hutchinson Cancer Research Center, Seattle, Washington: *Optimizing Engraftment of FANCA Gene-modified Bone Marrow Cells in a Mouse Model of Fanconi Anemia*
- 2:10 - 2:15 Questions and Answers
- 2:15 - 2:25 **Neelam Giri, MD**, National Cancer Institute, Rockville, Maryland: *Studies of Immune Function in Patients with Fanconi Anemia and Other Inherited Bone Marrow Failure Syndromes*
- 2:25 - 2:30 Questions and Answers
- 2:30 - 2:40 **John Postlethwait, PhD**, University of Oregon, Eugene, Oregon: *Fancd1 Mutant Zebrafish: Ovarian Invasive Tumors and Disturbed Oocyte Polarity*
- 2:40 - 2:45 Questions and Answers
- 2:45 - 3:00 **Break**
- 3:00 - 3:10 **Jie Li, PhD**, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio: *Loss of FA Function Impairs Insulin Signaling*
- 3:10 - 3:15 Questions and Answers
- 3:15 - 3:25 **Laura Hays, PhD**, Oregon Health & Science University, Portland, Oregon: *Human FANCC is Hypomorphic in Murine Fance-deficient Cells*
- 3:25 - 3:30 Questions and Answers
- 3:30 - 3:35 **Session Wrap-up: Madeleine Carreau, PhD**

Session VI: Molecular Diagnostics
Chair: Anna Savoia, PhD
University of Trieste
Trieste, Italy

- 3:35 - 3:40 **Session Overview: Anna Savoia, PhD**
- 3:40 - 3:50 **Anna Savoia, PhD**, University of Trieste, Trieste, Italy: *Molecular Genetic Testing of Fanconi Anemia: Experience of the Italian Research Group on Fanconi Anemia*
- 3:50 - 3:55 Questions and Answers
- 3:55 - 4:05 **Jordi Surrallés, PhD**, Universitat Autònoma de Barcelona, Barcelona, Spain: *Origin, Functional Role and Clinical Impact of Fanconi Anemia FANCA Mutations*
- 4:05 - 4:10 Questions and Answers

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- 4:10 - 4:20 **Elizabeth Flynn, PhD**, National Human Genome Research Institute, Bethesda, Maryland: *Comparative Genomic Hybridization (CGH) using a High-density Oligo Array for Discovering Deletions in all the FA Genes*
- 4:20 - 4:25 Questions and Answers
- 4:25 - 4:35 **Settara Chandrasekharappa, PhD**, National Human Genome Research Institute, Bethesda, Maryland: *Towards Development of a Next-generation Sequencing Strategy for Identification of Disease-causing Mutations in FA Genes*
- 4:35 - 4:40 Questions and Answers
- 4:40 - 4:50 **Arleen Auerbach, PhD**, The Rockefeller University, New York, New York: *An Enhanced International Fanconi Anemia Registry (IFAR)*
- 4:50 - 4:55 Questions and Answers
- 4:55 - 5:00 **Session Wrap-up: Anna Savoia, PhD**

5:00 - 7:00

Poster Presentations Reception

Wine and Hors d'Oeuvres

Ballroom 3

7:00 - 9:00

Symposium Dinner

All registrants are invited

Ballroom 4

Sunday – October 24, 2010

7:00 - 8:00

Buffet Breakfast

Ballroom 4

Poster Viewing

Ballroom 3

8:00 - 8:30

Special Session D: Induced Pluripotent Stem Cells and RNA Interference

M. William Lensch, PhD (for more information, see page 13)

Children's Hospital Boston

Boston, Massachusetts

8:30 - 8:45

Questions and Answers

Session VII: Repair

Chair: K.J. Patel, MD, PhD
MRC Laboratory of Molecular Biology
Cambridge, United Kingdom

8:45 - 8:50 **Session Overview: K.J.Patel, MD, PhD**

8:50 - 9:00 **Yuliang Wu, PhD**, National Institute on Aging, Baltimore, Maryland: *The Q Motif of FANCD1 Helicase Regulates its Dimerization and DNA Repair Function*

9:00 - 9:05 Questions and Answers

9:05 - 9:15 **Nigel Jones, PhD**, University of Liverpool, Liverpool, United Kingdom: *FANCD1 Functions Independently of the FA Core Complex in Homologous Recombination Repair of Phleomycin-induced DNA Damage*

9:15 - 9:20 Questions and Answers

9:20 - 9:30 **Allison Green, MPhil**, Yale University, New Haven, Connecticut: *A Role for RNA and Splicing Factors in the FA Pathway*

9:30 - 9:35 Questions and Answers

9:35 - 9:45 **Jean-Yves Masson, PhD**, Laval University, Québec, Canada: *Synergistic Function of PALB2 and BRCA2 in Stimulating Homologous Recombination is Dependent on PALB2 Dimerization*

9:45 - 9:50 Questions and Answers

9:50 - 10:00 **Jun Wang, PhD**, National Cancer Institute, Frederick, Maryland: *C/EBPdelta-Mediated Nuclear Import of FANCD2 by IPO4 Augments Cellular Response to DNA Damage*

10:00 - 10:05 Questions and Answers

10:05 - 10:10 **Session Wrap-up: K.J.Patel, MD, PhD**

10:10 - 10:30 **Break**

10:30 - 12:00 **Closing Forum: Planning for the Future**

Chair: Grover C. Bagby, Jr., MD
Chair, Scientific Advisory Board, Fanconi Anemia Research Fund
Oregon Health & Science University
Portland, Oregon