

## AGENDA

### Thursday – September 27, 2012

3:00            **Registration opens**  
*Imperial Ballroom Foyer, Second Floor*

4:00 - 6:00    **FA 101: An Introduction to the Medicine and Biology of Fanconi Anemia**  
*Mt. Sopris, Lobby Level*

Note: 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:**

**Grover C. Bagby Jr., MD**

Chair, Scientific Advisory Board, Fanconi Anemia Research Fund  
Oregon Health & Science University, Portland, Oregon

**Faculty:**

**Ray Monnat Jr., MD**

Scientific Advisory Board, Fanconi Anemia Research Fund  
University of Washington, Seattle, Washington

**Akiko Shimamura, MD, PhD**

Fred Hutchinson Cancer Research Center, Seattle, Washington

**International Fanconi Anemia Registry (IFAR) Demonstration:**

**Arleen Auerbach, PhD**

The Rockefeller University, New York, New York

**Research Opportunities:**

**Pankaj Qasba, PhD**

National Heart, Lung, and Blood Institute, NIH, Bethesda, Maryland

6:00 - 8:00

**Welcome Reception**

**Poster Viewing**

*Grand Ballroom, Second Floor*

Presenters with an odd-numbered poster will be at their posters 6:00-7:00.

Presenters with an even-numbered poster will be at their posters 7:00-8:00.

**Friday – September 28, 2012**

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7:00 - 8:00 { **Buffet Breakfast**  
*Mt. Sopris, Lobby Level*  
**Poster Viewing**  
*Grand Ballroom, Second Floor*

{ **Plenary Session**  
*Imperial Ballroom, Second Floor*

8:00 - 8:20 **Welcome**  
**David Frohnmayr, JD**  
Co-founder and Advisor, Board of Directors  
Fanconi Anemia Research Fund, Eugene, Oregon

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

8:20 - 8:30 **Fanconi Anemia Research Fund: Research Agenda and Resources**  
**Ray Monnat Jr., MD**  
Scientific Advisory Board, Fanconi Anemia Research Fund  
University of Washington, Seattle, Washington

**Session I: Drug and Small Molecule Therapeutics**

**Chair: Richard Gelinas, PhD**  
**Scientific Advisory Board, Fanconi Anemia Research Fund**  
**Institute for Systems Biology, Seattle, Washington**

8:30 - 8:35 **Session Overview: Richard Gelinas, PhD**

8:35 - 8:45 **Robert Sclafani, PhD**, University of Colorado School of Medicine, Aurora, Colorado: *Potential Therapeutic Use of Resveratrol for Head and Neck Carcinogenesis in Fanconi Anemia*

8:45 - 8:50 Questions and Answers

8:50 - 9:00 **Michael Spiotto, MD, PhD**, The University of Chicago, Chicago, Illinois: *A High Throughput Cell-based Screen Identifies Compounds That Inhibit the Human Papillomavirus Oncoprotein E6 Without Damaging DNA*

9:00 - 9:05 Questions and Answers

9:05 - 9:15 **Qingshuo Zhang, PhD**, Oregon Health & Science University, Portland, Oregon: *Decipher the Magic of Oxymetholone*

9:15 - 9:20 Questions and Answers

- 9:20 - 9:30 **Johanna Svahn, MD**, G. Gaslini Children's Hospital, Genova, Italy: *BIRB796, a p38MAPK Inhibitor, Stimulates in vitro Hematopoietic Progenitor and Stem Cell Colony Growth in FA-A Patients*
- 9:30 - 9:35 Questions and Answers
- 9:35 - 9:40 **Session Wrap-up: Richard Gelinas, PhD**
- 9:40 - 10:00 **Break**

**Session II: Genome Engineering and Stem Cells**

**Chair: Ray Monnat Jr., MD**  
**Scientific Advisory Board, Fanconi Anemia Research Fund**  
**University of Washington, Seattle, Washington**

- 10:00 - 10:05 **Session Overview: Ray Monnat Jr., MD**
- 10:05 - 10:15 **Juan Bueren, PhD**, CIEMAT, Madrid, Spain: *Generation of Disease-free Fanconi Anemia Hematopoietic Progenitors by Gene Targeting and Cell Reprogramming Approaches*
- 10:15 - 10:20 Questions and Answers
- 10:20 - 10:30 **Laura Marquez Loza**, Oregon Health & Science University, Portland, Oregon: *Generation of Isogenic Pairs of Mutant and Complemented Fanconi Anemia Human Induced Pluripotent Stem Cells*
- 10:30 - 10:35 Questions and Answers
- 10:35 - 10:45 **Mark Osborn, PhD**, University of Minnesota, Minneapolis, Minnesota: *Transcription Activator-like Effector Nuclease-mediated Genome Editing for Fanconi Anemia*
- 10:45 - 10:50 Questions and Answers
- 10:50 - 10:55 **Session Wrap-up: Ray Monnat Jr., MD**

**Session III: Bone Marrow Transplant Center Reports**

**Chair: Eva Guinan, MD**  
**Scientific Advisory Board, Fanconi Anemia Research Fund**  
**Harvard Medical School, Dana-Farber Cancer Institute**  
**Boston, Massachusetts**

- 10:55 - 11:00 **Session Overview: Eva Guinan, MD**
- 11:00 - 11:10 **Carmem Bonfim, MD**, Federal University of Parana, Curitiba, Brazil: *Long-term Follow-up After HSCT for Fanconi Anemia: Analysis of 126 Patients Surviving More Than Two Years After Transplant*
- 11:10 - 11:15 Questions and Answers

- 11:15 - 11:25 **Farid Boulad, MD**, Memorial Sloan-Kettering Cancer Center, New York, New York: *Long-term Follow-up of Patients with Fanconi Anemia After Allogeneic Hematopoietic Stem Cell Transplantation*
- 11:25 - 11:30 Questions and Answers
- 11:30 - 11:40 **Margaret MacMillan, MD**, University of Minnesota, Minneapolis, Minnesota: *Twenty Years of Alternative Donor Hematopoietic Cell Transplantation for Fanconi Anemia at the University of Minnesota*
- 11:40 - 11:45 Questions and Answers
- 11:45 - 11:50 **Session Wrap-up: Eva Guinan, MD**
- 11:50 - 12:50 { **Buffet Lunch**  
Mt. Sopris, Lobby Level  
**Poster Viewing**  
Grand Ballroom, Second Floor

<b>Special Session: Bone Marrow Transplantation: Point/Counterpoint in the Landscape of Change</b>
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**Chair: Jakub Tolar, MD, PhD**  
**Scientific Advisory Board, Fanconi Anemia Research Fund**  
**University of Minnesota, Minneapolis, Minnesota**  
*(see page 12 for Special Session participant statements)*

- 12:50 - 12:55 **Session Overview: Jakub Tolar, MD, PhD**

***Should danazol be offered before transplant for bone marrow failure in FA?***

- 12:55 - 1:05 **Helmut Hanenberg, MD**, Indiana University, Indianapolis, Indiana
- 1:05 - 1:15 **Stella Davies, MBBS, PhD, MRCP**, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio
- 1:15 - 1:25 Questions and Answers

***Should radiation be used in the conditioning regimen for FA transplants?***

- 1:25 - 1:35 **Margaret MacMillan, MD**, University of Minnesota, Minneapolis, Minnesota
- 1:35 - 1:45 **Farid Boulad, MD**, Memorial Sloan-Kettering Cancer Center, New York, New York
- 1:45 - 1:55 Questions and Answers

***Does transplant dramatically increase the risk of cancer in FA?***

- 1:55 - 2:05 **Blanche Alter, MD, MPH**, National Cancer Institute, Rockville, Maryland
- 2:05 - 2:15 **John Wagner, MD**, University of Minnesota, Minneapolis, Minnesota
- 2:15 - 2:25 Questions and Answers

- 2:25 - 2:40 **Session Questions and Answers and Wrap-up: Jakub Tolar, MD, PhD**

- 2:40 - 3:00 **Break**

**Session IV: Disease Drivers: Mutations from Metabolism**

**Chair: Alan D'Andrea, MD**

**Dana-Farber Cancer Institute, Boston, Massachusetts**

- 3:00 - 3:05      **Session Overview: Alan D'Andrea, MD**
- 3:05 - 3:15      **Gerry Crossan, PhD**, MRC Laboratory of Molecular Biology, Cambridge, United Kingdom: *A Specific Requirement for Both Aldehyde Catabolism and Fanconi-mediated DNA Repair for Haematopoietic Stem Cell Function*
- 3:15 - 3:20      Questions and Answers
- 3:20 - 3:30      **Juan Garaycochea, PhD Student**, MRC Laboratory of Molecular Biology, Cambridge, United Kingdom: *Endogenously Generated Aldehydes are Genotoxic to FA Pathway Deficient Haematopoietic Stem Cells*
- 3:30 - 3:35      Questions and Answers
- 3:35 - 3:45      **Nina Oberbeck, Graduate Student**, MRC Laboratory of Molecular Biology, Cambridge, United Kingdom: *Maternal Endogenous Aldehydes Lead to in utero DNA Damage and Developmental Defects in FANCA Deficient Embryos*
- 3:45 - 3:50      Questions and Answers
- 3:50 - 4:00      **Asuka Hira, MD, Graduate Student**, Kyoto University, Kyoto, Japan: *Variant ALDH2 is Associated with Accelerated Progression of Bone Marrow Failure in Japanese Fanconi Anemia Patients*
- 4:00 - 4:05      Questions and Answers
- 4:05 - 4:10      **Session Wrap-up: Alan D'Andrea, MD**

**Special Session: The Pursuit of Stem Cell Expansion**

**Chair: Jakub Tolar, MD, PhD**

**Scientific Advisory Board, Fanconi Anemia Research Fund**

**University of Minnesota, Minneapolis, Minnesota**

*(see page 19 for Special Session participant statements)*

- 4:10 - 4:15      **Session Overview: Jakub Tolar, MD, PhD**
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- 4:15 - 4:25      ***Special Session Stem Cell Expansion Panel***
- 4:15 - 4:25      **Hans-Peter Kiem, MD, FACP**, Fred Hutchinson Cancer Research Center, Seattle, Washington
- 4:25 - 4:35      **Colleen Delaney, MD**, Fred Hutchinson Cancer Research Center, Seattle, Washington
- 4:35 - 4:45      **Anthony Boitano, PhD**, Genomics Institute of the Novartis Research Foundation, San Diego, California
- 4:45 - 4:55      **Yosef Refaeli, PhD**, University of Colorado Denver, Aurora, Colorado
- 4:55 - 5:10      Questions and Answers
- 5:10 - 5:15      **Session Wrap-up: Jakub Tolar, MD, PhD**

6:30 - 8:30 { **Joint Meeting and Dinner**  
Board of Directors and Scientific Advisory Board  
Fanconi Anemia Research Fund  
*Maroon Peak, Second Floor*

**Saturday – September 29, 2012**

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7:00 - 8:00 { **Buffet Breakfast**  
*Mt. Sopris, Lobby Level*  
**Poster Viewing**  
*Grand Ballroom, Second Floor*

**Session V: Experimental Hematology**

**Chair: Markus Grompe, MD**

**Oregon Health & Science University, Portland, Oregon**

8:00 - 8:05 **Session Overview: Markus Grompe, MD**

8:05 - 8:15 **Wei Du, MD, PhD**, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio: *Enhanced Notch Signaling Skews Hematopoietic Stem Cell Differentiation in Fanconi Anemia Murine Models*

8:15 - 8:20 Questions and Answers

8:20 - 8:30 **Grover Bagby Jr., MD**, Oregon Health & Science University, Portland, Oregon: *HOXA9 Plays a Critical Role in the Evolution of Myelodysplastic Clones in Fanconi Anemia*

8:30 - 8:35 Questions and Answers

8:35 - 8:45 **Laura Hays, PhD**, Oregon Health & Science University, Portland, Oregon: *ROS-mediated Damage is Necessary, but Not Sufficient, for LPS-induced TNF Overproduction in Fancc-deficient M1 Macrophages*

8:45 - 8:50 Questions and Answers

8:50 - 9:00 **Neelam Giri, MD**, National Cancer Institute, Rockville, Maryland: *TNF-alpha and IFN-gamma Expression in Fanconi Anemia Bone Marrow*

9:00 - 9:05 Questions and Answers

9:05 - 9:15 **Madeleine Carreau, PhD**, Laval University, Quebec, Canada: *FANCC with CtBP1 Act in Transcriptional Regulation of the Wnt Antagonist Dickkopf-1*

9:15 - 9:20 Questions and Answers

9:20 - 9:30 **Michael Rotelli**, Oregon Health & Science University, Portland, Oregon: *Regulation of FANCL by Glycogen Synthase Kinase-3beta Links the FA Pathway to Self-renewal and Survival Signals*

9:30 - 9:35 Questions and Answers

9:35 - 9:40      **Session Wrap-up: Markus Grompe, MD**

9:40 - 10:00    **Break**

**Session VI:    FA Genes and Mutations**

**Chair: Johan de Winter, PhD**

**Vrije Universiteit Medical Center, Amsterdam, Netherlands**

10:00 - 10:05    **Session Overview: Johan de Winter, PhD**

10:05 - 10:15    **Kai Rogers**, University of Washington, Seattle, Washington: *Fanconi Mutations as Novel Therapeutic Opportunities*

10:15 - 10:20    Questions and Answers

10:20 - 10:30    **Elizabeth Flynn, PhD**, National Human Genome Research Institute, Bethesda, Maryland: *Comprehensive Study of Deletion Mutations Found in Fanconi Anemia Genes*

10:30 - 10:35    Questions and Answers

10:35 - 10:45    **Jordi Surrallés, PhD**, Universitat Autònoma de Barcelona, Barcelona, Spain: *Discovery of a Novel Fanconi Anemia Gene by Whole Exome Sequencing*

10:45 - 10:50    Questions and Answers

10:50 - 11:00    **Najim Ameziane, PhD**, Vrije Universiteit Medical Center, Amsterdam, Netherlands: *The First Real FA-M Patient Identified Through Next Generation Sequencing*

11:00 - 11:05    Questions and Answers

11:05 - 11:10    **Session Wrap-up: Johan de Winter, PhD**

**Session VII:    FA Protein Structure and Function I: Replication and Repair**

**Chair: Alexandra Sobeck, PhD**

**University of Minnesota, Minneapolis, Minnesota**

11:10 - 11:15    **Session Overview: Alexandra Sobeck, PhD**

11:15 - 11:25    **George-Lucian Moldovan, PhD**, Dana-Farber Cancer Institute, Boston, Massachusetts: *Inhibition of the Homologous Recombination Regulator PARI Improves Genomic Stability of Fanconi Anemia Patients*

11:25 - 11:30    Questions and Answers

11:30 - 11:40    **Julia Sidorova, PhD**, University of Washington, Seattle, Washington: *FANCD2 and the Response of DNA Replication to Crosslinker Drugs in Human Cells*

11:40 - 11:45    Questions and Answers

11:45 - 11:55 **Suhasini Avvaru, PhD**, National Institute on Aging, Baltimore, Maryland:  
*Fanconi Anemia Group J Helicase and MRE11 Nuclease Interact to Facilitate the DNA Damage Response*

11:55 - 12:00 Questions and Answers

12:00 - 1:30 { **Buffet Lunch**  
*Mt. Sopris, Lobby Level*  
**Poster Viewing**  
*Grand Ballroom, Second Floor*

**Session VII: FA Protein Structure and Function I: Replication and Repair (continued)**

1:30 - 1:40 **Andrew Deans, PhD**, St. Vincent's Institute, Melbourne, Australia: *Structural Analysis of FANCM/FAAP24: Insights into DNA Targeting of the Fanconi Anemia Core Complex*

1:40 - 1:45 Questions and Answers

1:45 - 1:55 **Magdalena Budzowska, PhD**, Harvard Medical School, Boston, Massachusetts:  
*Mechanism of Translesion Synthesis During DNA Interstrand Crosslink Repair*

1:55 - 2:00 Questions and Answers

2:00 - 2:05 **Session Wrap-up: Alexandra Soback, PhD**

**Session VIII: FA Protein Structure and Function II: Chromatin and Signaling**

**Chair: Toshiyasu Taniguchi, MD, PhD**

**Fred Hutchinson Cancer Research Center, Seattle, Washington**

2:05 - 2:10 **Session Overview: Toshiyasu Taniguchi, MD, PhD**

2:10 - 2:20 **Weidong Wang, PhD**, National Institute on Aging, Baltimore, Maryland: *RNF8 and FAAP20 Constitute a Ubiquitin Cascade That Controls Recruitment of Fanconi Anemia Core Complex to Damaged DNA*

2:20 - 2:25 Questions and Answers

2:25 - 2:35 **Angelos Constantinou, PhD**, IGH-Institute of Human Genetics, Montpellier, France: *The FANCD2/FANCI Complex is a General Effector of ATR Signaling That Controls the MCM2-7 Replicative Helicase*

2:35 - 2:40 Questions and Answers

2:40 - 2:50 **Paul Andreassen, PhD**, Cincinnati Children's Research Foundation, Cincinnati, Ohio: *FANCN/PALB2 is Recruited by the MDC1-RNF8-RAP80-Abraxas Ubiquitin-dependent Signaling Network*

2:50 - 2:55 Questions and Answers

- 2:55 - 3:05 **Indrajit Chaudhury, PhD**, University of Minnesota, Minneapolis, Minnesota: *FANCD2 Coordinates the BLM Helicase Complex With the Fanconi-associated Nuclease FAN1 to Promote Replication Restart*
- 3:05 - 3:10 Questions and Answers
- 3:10 - 3:15 **Session Wrap-up: Toshiyasu Taniguchi, MD, PhD**
- 3:15 - 3:35 **Break**

**Session IX: Carcinogenesis and Human Papillomavirus**

**Chair: William N. William Jr., MD**

**Scientific Advisory Board, Fanconi Anemia Research Fund**

**The University of Texas MD Anderson Cancer Center, Houston, Texas**

- 3:35 - 3:40 **Session Overview: William N. William Jr., MD**
- 3:40 - 3:50 **Blachy Davila Saldana, MD**, Oregon Health & Science University, Portland, Oregon: *Altered Immune Response of Fancc<sup>-/-</sup> Mice to HPV Vaccination*
- 3:50 - 3:55 Questions and Answers
- 3:55 - 4:05 **Blanche Alter, MD, MPH**, National Cancer Institute, Rockville, Maryland: *The HPV Status of Patients with Fanconi Anemia*
- 4:05 - 4:10 Questions and Answers
- 4:10 - 4:20 **Susanne Wells, PhD**, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio: *HPV-dependent and -independent FA Activities in Head and Neck Cancer Models*
- 4:20 - 4:25 Questions and Answers
- 4:25 - 4:35 **Eunmi Park, PhD**, Dana-Farber Cancer Institute, Boston, Massachusetts: *The Fanconi Anemia Pathway Suppresses Squamous Cell Carcinogenesis*
- 4:35 - 4:40 Questions and Answers

- 5:30 - 7:30 { **Poster Presentations Reception**  
*Grand Ballroom, Second Floor*  
Presenters with an odd-numbered poster will be at their posters 5:30-6:30.  
Presenters with an even-numbered poster will be at their posters 6:30-7:30.
- 7:30 - 9:30 { **Symposium Dinner**  
*Mt. Sopris, Lobby Level*

**Sunday – September 30, 2012**

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7:00 - 8:00 { **Buffet Breakfast**  
*Mt. Sopris, Lobby Level*  
**Poster Viewing**  
*Grand Ballroom, Second Floor*

**Session IX: Carcinogenesis and Human Papillomavirus (continued)**

8:00 - 8:10 **David Kutler, MD**, Weill Cornell Medical Center, New York, New York:  
*Natural History and Management of Fanconi Anemia Patients with Head and Neck Squamous Cell Carcinomas: A 10-year Follow-up*

8:10 - 8:15 Questions and Answers

8:15 - 8:25 **Ian Mackenzie, DDS, PhD**, Barts and The London Medical School, London, United Kingdom:  
*Enhanced Epithelial-mesenchymal Transition (EMT) and Therapeutic Resistance in Cells Lacking Fanconi Gene Function*

8:25 - 8:30 Questions and Answers

8:30 - 8:35 **Session Wrap-up: William N. William Jr., MD**

**Session X: FA Protein Structure and Function III: Cellular Roles**

**Chair: Stephen Meyn, MD, PhD**  
**Scientific Advisory Board, Fanconi Anemia Research Fund**  
**Hospital for Sick Children, Toronto, Canada**

8:35 - 8:40 **Session Overview: Stephen Meyn, MD, PhD**

8:40 - 8:50 **Markus Grompe, MD**, Oregon Health & Science University, Portland, Oregon:  
*Fancd2 and p21 Work Independently in Hematopoiesis Maintenance*

8:50 - 8:55 Questions and Answers

8:55 - 9:05 **Dong Zhang, PhD**, University of South Dakota, Vermillion, South Dakota:  
*FANCI Regulates Normal Centrosome Cycle and HU-induced Centrosome Over-amplification*

9:05 - 9:10 Questions and Answers

9:10 - 9:20 **Celine Jacquemont, PhD**, Fred Hutchinson Cancer Research Center, Seattle, Washington:  
*Function of USP28 as a Negative Regulator of the Fanconi Anemia Pathway and Homologous Recombination*

9:20 - 9:25 Questions and Answers

9:25 - 9:35 **Meghan Larin, MSc**, University of Toronto, Toronto, Canada: *Interplay Between Fanconi Anemia Signaling and Mus81 in Vivo*

9:35 - 9:40 Questions and Answers

- 9:40 - 9:50 **Nigel Jones, PhD**, University of Liverpool, Liverpool, United Kingdom:  
*FANCG Functions Independently of FANCA in the D1-D2-G-X3-RAD51C Complex: Evidence for Incomplete Epistasis of FANCG/A*
- 9:50 - 9:55 Questions and Answers
- 9:55 - 10:15 **Break**
- 10:15 - 10:25 **Supawat Thongthip, Graduate Student**, The Rockefeller University, New York, New York: *FAN1, a Nuclease in ICL Repair is Essential for Normal Kidney Function and Can Act Independently of the FA Pathway*
- 10:25 - 10:30 Questions and Answers
- 10:30 - 10:40 **Yucai Wang, MD, MS**, University of Texas MD Anderson Cancer Center, Houston, Texas: *FANCM and FAAP24 Maintain Genomic Stability Through Cooperative as Well as Unique Functions*
- 10:40 - 10:45 Questions and Answers
- 10:45 - 10:50 **Session Wrap-up: Stephen Meyn, MD, PhD**
- 10:50 - 12:00 ***Symposium Town Hall: An Interactive Discussion About FA Research***  
**Chair: Grover C. Bagby Jr., MD**  
Chair, Scientific Advisory Board, Fanconi Anemia Research Fund  
Oregon Health & Science University, Portland, Oregon