

## AGENDA

### Thursday – October 20, 2011

- 15:00            **Registration opens**
- 16:00 - 18:00    ***FA 101: An Introduction to the Medicine and Biology of Fanconi Anemia***  
*Tarragona-Girona Rooms*  
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
- Research Opportunities:**  
**Pankaj Qasba, PhD**  
Program Director, Blood Diseases Branch  
National Heart, Lung, and Blood Institute, NIH, Bethesda, Maryland
- 18:00 - 20:00    { **Welcome Reception**  
                          { **Poster Viewing**  
                          { *Leida Room*

### Friday – October 21, 2011

- 06:30 - 08:20    { **Buffet Breakfast**  
                          { *Batló-Güell Rooms, Mezzanine Level*  
                          { **Poster Viewing**  
                          { *Leida Room*
- 08:30            { **Plenary Session**  
                          { *Tarragona-Girona Rooms*

08:30 - 08:45 **Welcome**  
**David Frohnmayer, JD**  
Co-founder and Advisor, Board of Directors  
Fanconi Anemia Research Fund  
Eugene, Oregon

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

**Session I: FA Proteins and Crosslink Repair**  
**Chair: Ray Monnat, Jr., MD**  
**Scientific Advisory Board, Fanconi Anemia Research Fund**  
**University of Washington, Seattle, Washington**

08:45 - 08:50 **Session Overview: Ray Monnat, Jr., MD**

08:50 - 09:00 **Jean Gautier, PhD**, Columbia University Medical Center, New York, New York:  
*Mechanism of Replication-independent DNA Interstrand Crosslink Repair*

09:00 - 09:05 Questions and Answers

09:05 - 09:15 **Blanka Sengerova, PhD**, Weatherall Institute of Molecular Medicine, University of Oxford, Oxford, United Kingdom: *Human SNM1A and XPF-ERCC1 Collaborate to Initiate DNA Interstrand Cross-link Repair*

09:15 - 09:20 Questions and Answers

09:20 - 09:30 **David Thomas Long, PhD**, Harvard Medical School, Boston, Massachusetts:  
*RAD51 Promotes Repair of DNA Double-strand Breaks Created by the Fanconi Pathway During Interstrand Cross-link Repair*

09:30 - 09:35 Questions and Answers

09:35 - 09:45 **Indrajit Chaudhury, PhD**, University of Minnesota, Minneapolis, Minnesota:  
*FANCD2 Regulates Functions of the BLM DNA Repair Pathway Independently of FANCI*

09:45 - 09:50 Questions and Answers

09:50 - 10:00 **Marina Bellani, PhD**, National Institute on Aging, Baltimore, Maryland: *Rapid Multiphasic Recruitment of FA Associated Nuclease 1 (FAN1) to Sites of Crosslinks in Live Cells*

10:00 - 10:05 Questions and Answers

10:05 - 10:10 **Session Wrap-up: Ray Monnat, Jr., MD**

**Special Session: Gene Therapy/Embryonic Stem Cells & Induced Pluripotent Stem Cells**  
**Chair: Jakub Tolar, MD, PhD**  
**Scientific Advisory Board, Fanconi Anemia Research Fund**  
**University of Minnesota, Minneapolis, Minnesota**  
*(for more information, see page 11)*

- 10:10 - 10:20 **Meeting Report: *International FA Gene Therapy Work Group***  
Jakub Tolar, MD, PhD
- 10:20 - 10:25 Questions and Answers
- 10:25 - 10:45 **Break**
- 10:45 - 10:50 **Session Overview and Keynote Introduction: Jakub Tolar, MD, PhD**

<b>Keynote Address:      Modeling Bone Marrow Failure Syndromes Using Induced Pluripotent Stem Cells</b>
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- 10:50 - 11:10 **Suneet Agarwal, MD, PhD**, Children's Hospital Boston, Boston, Massachusetts  
11:10 - 11:15 Questions and Answers

- 11:15 - 11:25 **Montserrat Barragán, PhD**, Centro de Medicina Regenerativa de Barcelona, Barcelona, Spain: *Targeted Gene Correction of Patient-specific Fibroblasts for Cell-therapy of Spanish Gypsy Fanconi Anaemia*

- 11:25 - 11:30 Questions and Answers

- 11:30 - 11:40 **Susana Navarro, PhD**, CIEMAT, Madrid, Spain: *Generation and Hematopoietic Differentiation of Disease-free Induced Pluripotent Stem Cells from FA-D1 Mouse Fibroblasts*

- 11:40 - 11:45 Questions and Answers

*iPSC Panel – Moderated by Jakub Tolar, MD, PhD*

- 11:45 - 11:55 **Thomas Graf, PhD**, Center for Genomic Regulation, Barcelona, Spain  
11:55 - 12:05 **Angel Raya, MD, PhD**, Institute for Bioengineering of Catalonia, Barcelona, Spain  
12:05 - 12:15 **Toni Cathomen, PhD**, Hannover Medical School, Hannover, Germany  
12:15 - 12:25 **Juan Bueren, PhD**, CIEMAT, Madrid, Spain  
12:25 - 12:40 Questions and Answers

- 12:40 - 13:55 { **Hosted Lunch**  
*Batlló-Güell Rooms, Mezzanine Level*  
**Poster Viewing**  
*Lleida Room*

**Session II: Stem Cells, Hematopoiesis, and Transplantation**

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

- 13:55 - 14:00 **Session Overview: Eva Guinan, MD**

- 14:00 - 14:10 **Margaret MacMillan, MD**, University of Minnesota, Minneapolis, Minnesota:  
*Hematopoietic Cell Transplantation for FA Patients with Acute Leukemia or Advanced MDS*

- 14:10 - 14:15 Questions and Answers

- 14:15 - 14:25 **Ashley Kamimae-Lanning**, Oregon Health & Science University, Portland, Oregon: *Prenatal Origins of Hematopoietic Failure in a Mouse Model of Fanconi Anemia*

- 14:25 - 14:30 Questions and Answers

- 14:30 - 14:40 **Kathrin Scheckenbach, MD**, Heinrich-Heine University, Duesseldorf, Germany: *Successful Treatment of the Bone Marrow Failure in Fanconi Anemia Patients with Danazol*
- 14:40 - 14:45 Questions and Answers
- 14:45 - 14:55 **Kalindi Parmar, PhD**, Dana-Farber Cancer Institute, Boston, Massachusetts: *Hypersensitivity of Fanconi Anemia Pathway Deficient Hematopoietic Cells to Cytokinesis Inhibitors*
- 14:55 - 15:00 Questions and Answers
- 15:00 - 15:10 **Laura Hays, PhD**, Oregon Health & Science University, Portland, Oregon: *Fancc Suppresses TLR-dependent TNF $\alpha$  Gene Expression in M1-, but not M2-, Polarized Macrophages*
- 15:10 - 15:15 Questions and Answers
- 15:15 - 15:25 **Paula Río, PhD**, CIEMAT, Madrid, Spain: *Engraftment of Immunodeficient Mice with FA-like Hematopoietic Repopulating Cells: A New Model to Study Human FA-HSCs*
- 15:25 - 15:30 Questions and Answers
- 15:30 - 15:35 **Session Wrap-up: Eva Guinan, MD**
- 15:35 - 15:55 **Break**

**Session III: Fanconi Anemia Pathway**

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

- 15:55 - 16:00 **Session Overview: Stephen Meyn, MD, PhD**
- 16:00 - 16:10 **Kailin Yang**, Dana-Farber Cancer Institute, Boston, Massachusetts: *Regulation of the Fanconi Anemia Pathway by a SUMO-like Delivery Network*
- 16:10 - 16:15 Questions and Answers
- 16:15 - 16:25 **Agata Smogorzewska, MD, PhD**, The Rockefeller University, New York, New York: *Function of FANCP/SLX4 in the Fanconi Anemia Pathway*
- 16:25 - 16:30 Questions and Answers
- 16:30 - 16:40 **Kim-Hien Dao**, Oregon Health & Science University, Portland, Oregon: *FANCL Ubiquitinates Beta-catenin and Enhances Its Nuclear Function*
- 16:40 - 16:45 Questions and Answers
- 16:45 - 16:50 **Session Wrap-up: Stephen Meyn, MD, PhD**

**Session IV: FA Proteins, Forks, and Branches**

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

- 16:50 - 16:55 **Session Overview: Stephen Meyn, MD, PhD**

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- 16:55 - 17:05 **Maria Jasin, PhD**, Memorial Sloan-Kettering Cancer Center, New York, New York: *Unified Model for Fanconi Anemia and Breast Cancer Gene-network in Preventing Genome Instability at Stalled DNA Forks*
- 17:05 - 17:10 Questions and Answers
- 17:10 - 17:20 **Weidong Wang, PhD**, National Institute on Aging, Baltimore, Maryland: *Crystal Structures Reveal that FANCM Remodels the MHF Tetramer in Favor of Binding Branched DNA*
- 17:20 - 17:25 Questions and Answers
- 17:25 - 17:35 **Tomoko Shigechi, MD**, Kyoto University, Kyoto, Japan: *An Absolute Requirement of ATRIP-ATR Kinase in Replication Stress-induced Triggering of the FA Pathway Activation*
- 17:35 - 17:40 Questions and Answers
- 17:40 - 17:50 **Filippo Rosselli, PhD**, Institut Gustave Roussy, Villejuif, France: *FANCC Pathway Limits 53BP1 Accumulation to DSBs Associated with the Collapse of Stalled Replication Forks*
- 17:50 - 17:55 Questions and Answers
- 17:55 - 18:00 **Session Wrap-up: Stephen Meyn, MD, PhD**

### **Saturday – October 22, 2011**

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- 06:30 - 08:20 { **Buffet Breakfast**  
*Batló-Güell Rooms, Mezzanine Level*  
**Poster Viewing**  
*Leida Room*

#### **Session V: Carcinogenesis I**

**Chair: Erich Sturgis, MD, MPH**  
**Scientific Advisory Board, Fanconi Anemia Research Fund**  
**The University of Texas MD Anderson Cancer Center**  
**Houston, Texas**

- 08:30 - 08:35 **Session Overview and Keynote Introduction: Erich Sturgis, MD, MPH**

**Keynote Address: Human Papillomavirus and Head and Neck Cancer**

- 08:35 - 09:00 **Maura Gillison, MD, PhD**, Ohio State University Comprehensive Cancer Center, Columbus, Ohio  
*(for more information, see page 12)*
- 09:00 - 09:15 Questions and Answers
- 09:15 - 09:25 **Susanne Wells, PhD**, Cincinnati Children's Hospital, Cincinnati, Ohio: *The Fanconi Anemia Pathway Restricts the Human Papillomavirus Replicative Cycle*
- 09:25 - 09:30 Questions and Answers

- 09:30 - 09:40 **Jason Taylor, MD, PhD**, Oregon Health & Science University, Portland, Oregon: *B-cell Defects and Quantification of Antibody Response to HPV Vaccines in FancC-/- Mice*
- 09:40 - 09:45 Questions and Answers
- 09:45 - 09:55 **Gerry Crossan**, MRC Laboratory of Molecular Biology, Cambridge, United Kingdom: *Development of Hematological, Anogenital, and Hepatocellular Cancers in Mature FANCP Deficient Mice*
- 09:55 - 10:00 Questions and Answers
- 10:00 - 10:05 **Session Wrap-up: Erich Sturgis, MD, MPH**

**Session VI: Function of the FA Core Complex**

**Chair: Christopher Mathew, PhD**  
**Scientific Advisory Board, Fanconi Anemia Research Fund**  
**King's College London, London, United Kingdom**

- 10:05 - 10:10 **Session Overview: Christopher Mathew, PhD**
- 10:10 - 10:20 **Lei Li, PhD**, MD Anderson Cancer Center, Houston, Texas: *FANCM and FAAP24 Play Distinct Roles in Maintaining Genomic Stability*
- 10:20 - 10:25 Questions and Answers
- 10:25 - 10:35 **Charlotte Hodson**, London Research Institute, London, United Kingdom: *Structural Analysis of Human FANCL*
- 10:35 - 10:40 Questions and Answers
- 10:40 - 11:00 **Break**
- 11:00 - 11:10 **Jordi Surrallés, PhD**, Universitat Autònoma de Barcelona, Barcelona, Spain: *Is FAN1 a Novel Fanconi Anemia Gene?*
- 11:10 - 11:15 Questions and Answers
- 11:15 - 11:25 **Paul Andreassen, PhD**, Cincinnati Children's Research Foundation, Cincinnati, Ohio: *The Critical Function of BRCA1 in Homologous Recombination is to Localize FANCN/PALB2 to Sites of DNA Damage*
- 11:25 - 11:30 Questions and Answers
- 11:30 - 11:40 **Maria Castella, PhD**, Fred Hutchinson Cancer Research Center, Seattle, Washington: *Identification of New Kinases Involved in the Regulation of the Fanconi Anemia-BRCA Pathway*
- 11:40 - 11:45 Questions and Answers
- 11:45 - 11:55 **Nigel Jones, PhD**, University of Liverpool, Liverpool, United Kingdom: *Inhibition of NHEJ Reveals Distinct Roles for FANCG in the Repair of Interstrand Crosslinks and DNA Strand Breaks*
- 11:55 - 12:00 Questions and Answers

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- 12:00 - 12:10 **Carrie Adelman, PhD**, Cancer Research UK, South Mimms, United Kingdom: *HELQ Helicase: A New FA Associated Protein that Phenocopies FancD2-/- When Mutated in Mice*
- 12:10 - 12:15 Questions and Answers
- 12:15 - 12:25 **Caroline Huard, MSc**, Laval University, Québec City, Canada: *Identification of CtBP1 as a New FA Core Complex Interacting Protein*
- 12:25 - 12:30 Questions and Answers
- 12:30 - 12:40 **Jean-Yves Masson, PhD**, Laval University Cancer Research Center, Québec City, Canada: *Functions of PALB2 and BRCA2 in Stimulating the Invasion Step of Homologous Recombination*
- 12:40 - 12:45 Questions and Answers
- 12:45 - 12:50 **Session Wrap-up: Christopher Mathew, PhD**
- 12:50 - 14:45 { **Hosted Lunch**  
*Batló-Güell Rooms, Mezzanine Level*  
**Poster Viewing**  
*Leida Room*

**Note: Mini-session available at the end of lunch period for interested participants**

**Mini-session: Next-generation Sequencing**

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

- 13:45 - 13:50 **Session Overview: Richard Gelinas, PhD**
- 13:50 - 13:58 **Josephine C. Dorsman, PhD**, Vrije Universiteit Medical Center, Amsterdam, The Netherlands: *A Next-generation Sequencing Prioritization Tool Tailored for Uncovering Novel FA Genes*
- 13:58 - 14:06 **Najim Ameziane, MD**, Vrije Universiteit Medical Center, Amsterdam, The Netherlands: *Novel Comprehensive Mutation Screening Approach for FA by Multiplexed Targeted Next-generation Sequencing*
- 14:06 - 14:14 **Settara Chandrasekharappa, PhD**, National Human Genome Research Institute, Bethesda, Maryland: *Application of Next-gen Sequencing Technologies for Screening of Fanconi Anemia Gene Mutations*
- 14:14 - 14:24 Questions and Answers
- 14:24 - 14:30 **Session Wrap-up: Richard Gelinas, PhD**

14:45 - 14:50 **Keynote Introduction: Grover C. Bagby, Jr., MD**

**Keynote Address: Fanconi Anemia and Novel Drug Targets**

14:50 - 15:15 **Alan D'Andrea, MD**, Dana-Farber Cancer Institute, Boston, Massachusetts  
*(for more information, see page 13)*

15:15 - 15:30 Questions and Answers

**Session VII-A: Experimental Therapies**

**Chair: K. J. Patel, MD, PhD**

**MRC Laboratory of Molecular Biology, Cambridge, United Kingdom**

15:30 - 15:35 **Session Overview: K.J. Patel, MD, PhD**

15:35 - 15:45 **Michael Garbati, PhD**, Oregon Health & Science University, Portland, Oregon:  
*p38 MAPK Inhibition Suppresses the TLR-hypersensitive Phenotype in FANCC-deficient Macrophages*

15:45 - 15:50 Questions and Answers

15:50 - 16:00 **Jordi Villa**, Universitat Autònoma de Barcelona, Barcelona, Spain: *Downregulation of N-Ethylmaleimide Sensitive Factor Protects Fanconi Anemia Cells from DNA Damage*

16:00 - 16:05 Questions and Answers

16:05 - 16:15 **Céline Jacquemont, PhD**, Fred Hutchinson Cancer Research Center, Seattle, Washington: *USP28 is a Modulator of the Fanconi Anemia Pathway and DNA Double-strand Break Repair*

16:15 - 16:20 Questions and Answers

16:20 - 16:30 **Johanna Svahn, MD**, Gaslini Childrens Hospital, Genova, Italy: *Kinase Inhibitors Reduce TNF-alpha Overproduction in Monocytes from Fanconi Anemia Group A Patients*

16:30 - 16:35 Questions and Answers

16:35 - 16:55 **Break**

**Session VII-B: Experimental Therapies**

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

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

16:55 - 17:05 **Ana Filipa Brinco de Oliveira Ponte**, University of Porto, Porto, Portugal:  
*α-Lipoic Acid and N-acetylcysteine as an Effective Cocktail for Decreasing Spontaneous Chromosome Instability in FA*

17:05 - 17:10 Questions and Answers

17:10 - 17:20 **Markus Grompe, MD**, Oregon Stem Cell Center, Portland, Oregon: *Oral Administration of a Direct SIRT1 Activating Compound Enhances Hematopoiesis in Fancd2-/- Mice*

17:20 - 17:25 Questions and Answers



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- 17:25 - 17:35 **Frédéric Langevin**, MRC Laboratory of Molecular Biology, Cambridge, United Kingdom: *An Essential Requirement for the Joint Action of Acetaldehyde Catabolism and Fancd2 in Embryonic Development*
- 17:35 - 17:40 Questions and Answers
- 17:40 - 17:50 **Daria Mochly-Rosen, PhD**, Stanford University School of Medicine, Stanford, California: *Allosteric Activator of Aldehyde Dehydrogenase 2, Alda-1, Reduces Aldehydic Load*
- 17:50 - 17:55 Questions and Answers
- 17:55 - 18:00 **Session Wrap-up: Toshiyasu Taniguchi, MD, PhD**

- 19:00 - 20:00 { **Poster Presentations Reception**  
Wine and Hors d'Oeuvres  
*Lleida Room*
- 20:00 - 22:00 { **Symposium Dinner**  
All registrants are invited  
*Batlló-Güell Rooms, Mezzanine Level*

## Sunday – October 23, 2011

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- 06:30 - 08:20 { **Buffet Breakfast**  
*Batlló-Güell Rooms, Mezzanine Level*  
**Poster Viewing**  
*Lleida Room*
- 07:00 - 08:20 { **Joint Meeting and Breakfast**  
Board of Directors and Scientific Advisory Board  
Fanconi Anemia Research Fund  
*Roses Room*
- 08:30 - 08:35 **Keynote Introduction: Eva Guinan, MD**

**Keynote Address: DNA Damage Signals and Space Radiation Risk**

- 08:35 - 09:00 **Francis Cucinotta, PhD**, NASA Space Radiation Program, Houston, Texas  
*(for more information, see page 14)*
- 09:00 - 09:15 Questions and Answers

**Session VIII: Carcinogenesis II**

**Chair: Markus Grompe, MD**

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

- 09:15 - 09:20 **Session Overview: Markus Grompe, MD**
- 09:20 - 09:30 **Hein te Riele, PhD**, The Netherlands Cancer Institute, Amsterdam, The Netherlands: *Deficiency for FANCM, but not FANCF, Accelerates Tumorigenesis in a Mouse Model for Intestinal Cancer*
- 09:30 - 09:35 Questions and Answers
- 09:35 - 09:45 **Thomas Ludwig, PhD**, Institute for Cancer Genetics, New York, New York: *The Tumor-suppressor Activity of the FancJ/Brip1 Helicase is not Dependent on the Interaction of FancJ/Brip1 with Brca1*
- 09:45 - 09:50 Questions and Answers
- 09:50 - 10:00 **Spencer Luebben**, University of Minnesota, Minneapolis, Minnesota: *A Concomitant Loss of the Fanconi Anemia Pathway and Dormant Replication Origins Accelerates Tumorigenesis*
- 10:00 - 10:05 Questions and Answers
- 10:05 - 10:15 **Ian Mackenzie, DDS, PhD**, Barts and The London Medical School, London, United Kingdom: *Epithelial-mesenchymal Transition (EMT) in Cancer Cell Lines Lacking Fanconi Gene Function*
- 10:15 - 10:20 Questions and Answers
- 10:20 - 10:25 **Session Wrap-up: Markus Grompe, MD**
- 10:25 - 10:45 **Break**
- 10:45 - 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