

AGENDA

THURSDAY 17

3:00 Symposium Check-in and Registration Opens

Metropolitan Foyer

FA 101 Introductory Presentation

Metropolitan West

Note: This session is intended for those new to Fanconi anemia research and clinical care. However, all meeting participants interested in an overview of unanswered questions and new research and clinical directions in Fanconi anemia are encouraged to attend.

4:00 - 4:10 **Introductions:**

Amy Frohnmayer, MA, Bend, United States

Board of Directors, Fanconi Anemia Research Fund, FA Adult

Christopher Byrd, Esq., Winter Springs, United States

Board of Directors, Fanconi Anemia Research Fund, FA Adult

4:10 - 4:55 **Medicine: Carmen Bonfim, MD, Federal University of Paraná, Curitiba, Brazil**

4:55 - 5:05 Questions and answers

5:05 - 5:50 **Biology: Agata Smogorzewska, MD, PhD, The Rockefeller University, New York, United States**

5:50 - 6:00 Questions and answers

Welcome Reception and Poster Viewing

Metropolitan Centre

6:00 - 8:00 **Presenters in odd-numbered poster sections will be at their posters 6:00 - 7:00.**

Presenters in even-number poster sections will be at their posters 7:00 - 8:00.

FRIDAY 18

7:00 - 8:00 Breakfast Buffet

Metropolitan East

7:00 - 8:00 **Poster viewing available**

Welcome and Introduction

Metropolitan West

8:00 - 8:25 **Lynn Frohnmayer, MSW, Eugene, United States**

Co-founder and Board Vice President, Fanconi Anemia Research Fund, FA Parent

Raymond Monnat Jr., MD, University of Washington, Seattle, Washington

Chair of Scientific Advisory Board, Fanconi Anemia Research Fund

FA Genetics: Mutation to Phenotype

Metropolitan West

Chair: Jordi Surrallés, PhD

Universitat Autònoma de Barcelona, Barcelona, Spain

8:25 - 8:30 **Session Overview: Jordi Surrallés, PhD**

8:30 - 8:40 **Asuka Hira, MD, Radiation Biology Center, Kyoto University, Kyoto, Japan**

Mutations in the Gene Encoding the E2 Conjugating Enzyme UBE2T Cause Fanconi Anemia

8:40 - 8:45 Questions and answers

8:45 - 8:55	Kim Rickman, MD/PhD Student, The Rockefeller University, New York, United States <i>Deficiency of UBE2T, the E2 Ubiquitin Ligase Necessary for D2 and I Ubiquitination, Causes a New Subtype of FA</i>
8:55 - 9:00	Questions and answers
9:00 - 9:10	Helmut Hanenberg, MD, Heinrich Heine University, Düsseldorf, Germany <i>Germline Mutations and Somatic Reversions in FANCT That Cannot be Detected by Genomic Sequencing</i>
9:10 - 9:15	Questions and answers
9:15 - 9:25	Jung-Young Park, PhD, Cincinnati Children's Research Foundation, Cincinnati, United States <i>Protein Binding to PALB2 and Reversion of DNA Interstrand Crosslink Sensitivity Qualifies XRCC2 as a Fanconi Anemia Gene</i>
9:25 - 9:30	Questions and answers
9:30 - 9:40	Josephine Dorsman, PhD, VU University Medical Center, Amsterdam, Netherlands <i>Expression of RAD51^{A293T} Leads to Defective Strand Exchange Activity in a Dominant Manner and is Associated with a New FA Subtype, "FA-R".</i>
9:40 - 9:45	Questions and answers
9:45 - 9:55	Blanche Alter, MD, MPH, National Cancer Institute, Rockville, United States <i>Novel FANCI Mutations in Fanconi Anemia with VACTERL Association</i>
9:55 - 10:00	Questions and answers
10:00 - 10:05	Session Wrap-up: Jordi Surrallés, PhD
Replication: Initiation, Forks and RNA	
<i>Metropolitan West</i>	
Chair: Toshi Taniguchi, MD, PhD	
Fred Hutchison Cancer Research Center, Seattle, United States	
10:05 - 10:10	Session Overview: Toshi Taniguchi, MD, PhD
10:10 - 10:20	Advaitha Madireddy, PhD, Albert Einstein College of Medicine, New York, United States <i>Genomic Influence of FANCD2 in Facilitating DNA Replication</i>
10:20 - 10:25	Questions and answers
10:25 - 10:35	Grant Stewart, PhD, University of Birmingham, Birmingham, England <i>BOD1L Is Required to Suppress Deleterious Resection of Stressed Replication Forks</i>
10:35 - 10:40	Questions and answers
10:40 - 11:00	Break
11:00 - 11:10	Nigel Jones, PhD, University of Liverpool, Liverpool, England <i>Diadenosine Tetraphosphate (Ap4A) Is Synthesized in Response to ICL and Inhibits the Initiation of DNA Replication</i>
11:10 - 11:15	Questions and answers
11:15 - 11:25	Kevin Hiom, PhD, Jacqui Wood Cancer Centre, Dundee, Scotland <i>New Factors Required In the Repair of DNA Breaks, Link Replication and RNA Processing</i>
11:25 - 11:30	Questions and answers
11:30 - 11:35	Session Wrap-up: Toshi Taniguchi, MD, PhD

Recombination: A Tale of Two Pathways*Metropolitan West***Chair: Stephen Meyn, MD, PhD**

The Hospital for Sick Children, Toronto, Canada

11:35 - 11:40 **Session Overview: Stephen Meyn, MD, PhD**11:40 - 11:50 **Brooke Conti, The Rockefeller University, New York, United States***Suppression of Non-Homologous End Joining Does Not Rescue Sensitivity to ICL-Inducing Agents in Fanconi Anemia Cells*

11:50 - 11:55 Questions and answers

11:55 - 12:05 **Zeina Kais, PhD, Dana Farber Cancer Institute, Boston, United States***FANCD2 Functions in Homologous Recombination and Alternative End-Joining*

12:05 - 12:10 Questions and answers

12:10 - 12:20 **Eva Lee, MSc, University of Toronto, Toronto, Canada***Distinctive Roles for FANCI and FANCD2 in the Resection of Radiation-Induced DNA Double-Strand Breaks*

12:20 - 12:25 Questions and answers

12:25 - 12:35 **Weixing Zhao, PhD, Yale University, New Haven, United States***Promotion of BRCA2-dependent Homologous Recombination by DSS1 via RPA Targeting and DNA Mimicry*

12:35 - 12:40 Questions and answers

12:40 - 12:45 **Session Wrap-up: Stephen Meyn, MD, PhD****12:45 - 2:00 Lunch Buffet***Metropolitan East*12:45 - 2:00 **Mentorship Lunch for Early Investigators**12:45 - 2:00 **Poster Viewing Available****Immunity and Inflammation***Metropolitan West***Chair: Eva Guinan, MD**

Dana Farber Cancer Institute, Boston, United States

2:00 - 2:05 **Keynote Introduction: Eva Guinan, MD**2:05 - 2:45 **Glen Barber, PhD, University of Miami School of Medicine, Miami, United States****KEYNOTE ADDRESS:** *Inflammation and DNA Damage*2:45 - 2:50 **Keynote Wrap-up and Session Introduction: Eva Guinan, MD**2:50 - 3:00 **Melinda Butsch Kovacic, MPH, PhD, Cincinnati Children's Hospital Medical Center, Cincinnati, United States***Impaired Immune Function in Individuals with Fanconi Anemia over Time*

3:00 - 3:05 Questions and answers

3:05 - 3:15 **Said Aoufouchi, PhD, Gustave Roussy Institute, Villejuif, France***Fancc Loss-Of-Function Leads To Altered Diversification of Immunoglobulin Genes in Differentiating B Cells*

3:15 - 3:20 Questions and answers

3:20 - 3:40 **Break**3:40 - 3:50 **Flavia R.F. Teles, DDS, MS, DMSc, UNC School of Dentistry, Chapel Hill, United States***Inflammation and Oral Carcinogenesis in Fanconi Anemia*

3:50 - 3:55 Questions and answers

3:55 - 4:05 **Michael Garbati, PhD, Oregon Health & Science University/Portland VA Medical Center, Portland, United States**

Loss of DACH1 Expression in FA Macrophages Leads to TLR Pathway Hyperactivity

4:05 - 4:10 Questions and answers

4:10 - 4:15 **Session Wrap-up: Eva Guinan, MD**

Poster Reception

Metropolitan Centre

5:30 - 7:30 **Presenters in even-numbered poster sections will be at their posters 5:30- 6:30.**

Presenters in odd-number poster sections will be at their posters 6:30 - 7:30.

7:30 - 9:30 Symposium Banquet

Metropolitan East

7:00 - 8:00 Breakfast Buffet

Metropolitan East

7:00 - 8:00 **Poster viewing available**

Squamous Cell Carcinoma: Prevalence, Etiology and Treatment Options

Metropolitan West

Chair: Susanne Wells, PhD

Cincinnati Children's Hospital Medical Center, Cincinnati, United States

8:00 - 8:05 **Session Overview: Susanne Wells, PhD**

8:05 - 8:15 **Gregor Castrillón Oberndorfer, MD, DMD, City Hospital Karlsruhe, Karlsruhe, Germany**

Facing the Challenge: Treatment of Oral Cancer in Young Patients with Fanconi Anemia

8:15 - 8:20 Questions and answers

8:20 - 8:30 **Krupa Patel, The Rockefeller University/Weill Cornell Medical College, New York, United States**

Squamous Cell Carcinoma of the Anogenital Tract in Patients with Fanconi Anemia

8:30 - 8:35 Questions and answers

8:35 - 8:45 **Susanne Wells, PhD, Cincinnati Children's Hospital Medical Center, Cincinnati, United States**

FA Pathway Loss Impairs the Integrity of Normal and Transformed Epidermis

8:45 - 8:50 Questions and answers

8:50 - 9:00 **Michael Epperly, PhD, University of Pittsburgh, Pittsburgh, United States**

Radioprotection of Fancg^{-/-} Mouse Oral Cavity by Mitochondrial Targeted JP4-039

9:00 - 9:05 Questions and answers

9:05 - 9:15 **Quintin Pan, PhD, The Ohio State University Medical Center, Columbus, United States**

Development of a Small Molecule P300 Ligand to Simultaneously Reactivate P53 and Rb in HPV-Associated HNSCC

9:15 - 9:20 Questions and answers

9:20 - 9:30 **Molly Gilligan, Harvard Medical School/Beth Israel Deaconess Medical Center, Boston, United States**

Suppression of Cell Debris-Stimulated FANCC^{-/-} Head & Neck Cancer Progression by Resolvin Mediated Clearance

9:30 - 9:35 Questions and answers

- 9:35 - 9:40 **Session Wrap-up: Susanne Wells, PhD**
- 9:40 - 9:45 **Keynote Introduction: William William, MD, The University of Texas MD Anderson Cancer Center, Houston, United States**
- 9:45 - 10:25 **Michael Jensen, MD, Seattle Children's Research Institute, Seattle, United States**
KEYNOTE ADDRESS: *Enhancing the Synthetic IQ of CAR T Cells*
- 10:25 - 10:30 **Wrap-up: William William, MD**
- 10:30 - 10:50 **Break**

FA Protein Structure and Function

Metropolitan West

Chair: Andrew Deans, PhD

St Vincent's Institute, Fitzroy, Australia

- 10:50 - 10:55 **Session Overview: Andrew Deans, PhD**
- 10:55 - 11:05 **Andrew Deans, PhD, St Vincent's Institute, Fitzroy, Australia**
Recombinant Fanconi Anemia Core Complex: Insights into the Ubiquitination Reaction at the Centre of the FA Pathway
- 11:05 - 11:10 Questions and answers
- 11:10 - 11:20 **Yie Liu, PhD, National Institute on Aging/National Institutes of Health, Baltimore, United States**
The BTB Domain Is Both a Structural and Functional Pivot of the SLX4-Nuclease Complex
- 11:20 - 11:25 Questions and answers
- 11:25 - 11:35 **Robert Brosh, PhD, National Institute on Aging/National Institutes of Health, Baltimore, United States**
Structure-Function Analyses of Patient-Derived Missense Mutations in the Fanconi Anemia Complementation Group J (FANCI)
- 11:35 - 11:40 Questions and answers
- 11:40 - 11:50 **Ronald Cheung, MD, PhD, Fred Hutchinson Cancer Research Center, Seattle, United States**
Regulation of the Fanconi Anemia Pathway by the Multi-Step Phosphorylation of the FANCI SQ Cluster
- 11:50 - 11:55 Questions and answers
- 11:55 - 12:00 **Session Wrap-up: Andrew Deans, PhD**

12:00 - 2:00 Lunch Buffet

Metropolitan East

- 12:00 - 2:00 **Joint Board of Directors and Scientific Advisory Board Meeting**
Queens Quay Conference Room
- 12:00 - 2:00 **Poster Viewing Available**

Hematology: Past, Present and Future

Metropolitan West

Chair: Carlo Dufour, MD

Giannina Gaslini Children's Hospital, Genova, Italy

- 2:00 - 2:05 **Session Overview: Carlo Dufour, MD**
- 2:05 - 2:15 **Carlo Dufour, MD, Giannina Gaslini Children's Hospital, Genova, Italy**
The Effect of Hematopoietic Stem Cell Transplant on Survival Based on Hematological Status at Eligibility to Transplant
- 2:15 - 2:20 Questions and answers
- 2:20 - 2:30 **Nicholas Khan, MSPH, National Cancer Institute, Rockville, United States**
Preemptive Bone Marrow Transplantation and Event-Free Survival in Fanconi Anemia
- 2:30 - 2:35 Questions and answers

- 2:35 - 2:45 **Margaret MacMillan, MD, University of Minnesota, Minneapolis, United States**
Impact of Hematopoietic Cell Transplantation for Biallelic BRCA2
- 2:45 - 2:50 Questions and answers
- 2:50 - 3:00 **Arleen Auerbach, PhD, The Rockefeller University, New York, United States**
Unusual Revertant Mosaicism in Three Siblings Affected with Fanconi Anemia FA-G and Cancer
- 3:00 - 3:05 Questions and answers
- 3:05 - 3:15 **Susana Navarro, PhD, CIEMAT/CIBERER/IIS-FJD, Madrid, Spain**
Lentiviral-Mediated Gene Correction of Hematopoietic Progenitors and Repopulating Cells from FA-A Patients Using a Validated GMP Transduction Protocol
- 3:15 - 3:20 Questions and answers
- 3:20 - 3:30 **Wei Tong, PhD, Children's Hospital of Philadelphia, Philadelphia, United States**
Lnk Deficiency Ameliorates Hematopoietic Stem/Progenitor Defects in Fanconi Anemia Mice
- 3:30 - 3:35 Questions and answers
- 3:35 - 3:40 **Session Wrap-up: Carlo Dufour, MD**
- 3:40 - 4:00 **Break**

Crosslinks: Convergence of Repair Pathways

Metropolitan West

Chair: Nigel Jones, PhD

University of Liverpool, Liverpool, England

- 4:00 - 4:05 **Session Overview: Nigel Jones, PhD**
- 4:05 - 4:15 **Niyo Kato, PhD Student, Columbia University, New York, United States**
Recognition and Processing of DNA Interstrand Cross-Links by Mismatch Repair Proteins (MMR)
- 4:15 - 4:20 Questions and answers
- 4:20 - 4:30 **Elizabeth Vuono, University of Rhode Island, Kingston, United States**
The PTEN Phosphatase Functions Cooperatively with the Fanconi Anemia Proteins in DNA Crosslink Repair (HR)
- 4:30 - 4:35 Questions and answers
- 4:35 - 4:45 **Jieqiong Zhang, PhD, Harvard Medical School, Boston, United States**
Incision-Independent Repair of a Psoralen DNA Interstrand Crosslink (TLS)
- 4:45 - 4:50 Questions and answers
- 4:50 - 4:55 **Session Wrap-up: Nigel Jones, PhD**

Experimental Hematology and Small Molecules

Metropolitan West

Chair: Madeleine Carreau, PhD

Laval University, Quebec, Canada

- 8:00 - 8:05 **Session Overview: Madeleine Carreau, PhD**
- 8:05 - 8:15 **Stefan Meyer, MD, PhD, FRCPCH, University of Manchester, Manchester, England**
Perturbation of Age Dependent Gene Expression in Fanconi Anaemia (FA) - a Novel Aspect of the FA Phenotype
- 8:15 - 8:20 Questions and answers

8:20 - 8:30	Haojian Zhang, PhD, Dana Farber Cancer Institute, Boston, United States <i>TGF-beta Pathway Inhibition Rescues the Function of HSPCs Derived from Patients with Fanconi Anemia</i>
8:30 - 8:35	Questions and answers
8:35 - 8:45	Qingshuo Zhang, PhD, Oregon Health & Science University, Portland, United States <i>Dietary Metformin Administration Delayed Tumor Onset and Alleviated Bone Marrow Defects in Fanconi Anemia Mice</i>
8:45 - 8:50	Questions and answers
8:50 - 8:55	Session Wrap-up: Madeleine Carreau, PhD
8:55 - 9:00	Keynote Introduction: Maureen Hoatlin, PhD, MBA, Oregon Health and Science University, Portland, United States
9:00 - 9:40	Patrick Gray, PhD, Omeros, Seattle, United States KEYNOTE ADDRESS: <i>Drug Discovery and Development for Novel Targets</i>
9:40 - 9:50	Wrap-up and Rare Disease Organizations and Drug Development: Maureen Hoatlin, PhD, MBA
9:50 - 10:10	Break
Potential New Roles for FA Proteins <i>Metropolitan West</i>	
Chair: Richard Gelinias, PhD Institute for Systems Biology, Seattle, United States	
10:10 - 10:15	Session Overview: Richard Gelinias, PhD
10:15 - 10:25	Roberta Bottega, PhD, University of Trieste, IRCCS Burlo Garofolo, Trieste, Italy <i>Any Direct Role of FANCA in Mitochondria?</i>
10:25 - 10:30	Questions and answers
10:30 - 10:40	Alex Lyakhovich, PhD, International Clinical Research Center of St. Anne's University Hospital, Brno, Czech Republic <i>Impaired Mitophagy in Fanconi Anemia</i>
10:40 - 10:45	Questions and answers
10:45 - 10:55	Samuel Sondalle, MD/PhD Student, Yale University School of Medicine, New Haven, United States <i>A Nucleolar Role for the Fanconi Anemia Pathway Protein, FANCI</i>
10:55 - 11:00	Questions and Answers
11:00 - 12:00	Town Hall <i>Metropolitan West</i>