

# 2017 FA Scientific Symposium Agenda

*Thursday, 14*

<b>3:00</b> <i>Prefunction</i>	<b>Symposium Check-in and Registration Opens</b>
<b>4:30 – 6:00</b> <i>Grand Ballroom 2&amp;3</i>	<b>Living With FA: Natural History of Disease &amp; Clinical Perspectives</b> <i>Chair: Mark Quinlan, Executive Director, Fanconi Anemia Research Fund, Eugene, United States</i>
<b>4:30 – 4:35</b>	<b>Introduction:</b> Mark Quinlan, Executive Director, Fanconi Anemia Research Fund
<b>4:35 – 4:55</b> 4:55 – 5:00 Q&A	<b>Early Childhood (0 – 10 yrs)</b> <i>Akiko Shimamura, Dana-Farber/Boston Children's Hospital, United States</i> <i>Lisa Mingo, FA Parent, Vancouver, Canada</i>
<b>5:00 – 5:20</b> 5:20 – 5:25 Q&A	<b>Late Childhood/Young Adults (11 – 20 yrs)</b> <i>Stella Davies, Cincinnati Children's Hospital Medical Center, United States;</i> <i>Board of Directors, Fanconi Anemia Research Fund</i> <i>Stan and Michelle Kalemba, FA Parents, St. John, United States</i>
<b>5:25 – 5:45</b> 5:45 – 5:50 Q&A	<b>Adulthood</b> <i>Farid Boulad, Memorial Sloan Kettering Cancer Center, New York, United States</i> <i>Jason Brannock, Adult with FA, Apex, United States</i>
<b>5:50 – 6:00 Q&amp;A</b>	All panelists
<b>6:00 – 8:00</b> <i>Prefunction and Buckhead Ballroom</i>	<b>Welcome Reception and Poster Viewing</b>  <i>Presenters of odd-numbered posters will be at their posters 6:00 to 7:00</i> <i>Presenters of even-numbered posters will be at their posters 7:00 to 8:00</i>

*Friday, 15*

<b>7:00 – 8:00</b> <i>Grand Ballroom 1</i>	<b>Breakfast</b>
<b>8:00 – 8:10</b> <i>Grand Ballroom 2&amp;3</i>	<b>Welcome and Introduction</b>  <i>Kevin McQueen, President, Board of Directors, Fanconi Anemia Research Fund, Midlothian, United States</i> <i>Ray Monnat, Jr., University of Washington, Seattle, United States; Chair of Scientific Advisory Board, Fanconi Anemia Research Fund</i>
<b>8:10 – 9:50</b> <i>Grand Ballroom 2&amp;3</i>	<b>Cancer in FA</b> <i>Chair: Agata Smogorzewska, The Rockefeller University, New York, United States</i>
<b>8:10 – 8:15</b>	<b>Session Overview: Agata Smogorzewska</b>
<b>8:15 – 8:40</b> 8:40 – 8:45 Q&A	<b>Keynote Address: Epidemiology of Cancer in Fanconi Anemia</b> <i>Blanche P. Alter, National Cancer Institute, Rockville, United States</i> <span style="float: right;">p. 6</span>
<b>8:45 – 8:55</b> 8:55 – 9:00 Q&A	<b>Lipidomic Profiling of Fanconi Anemia Deficient Normal and Transformed Keratinocytes Reveals Perturbed Glycosphingolipid Metabolism</b> <i>Susanne Wells, Cincinnati Children's Hospital Medical Center, Cincinnati, United States</i> <span style="float: right;">p. 8</span>

<b>9:00 – 9:10</b> 9:10 – 9:15 Q&A	<b>Sequencing of Clinically Relevant Somatic Gene Mutations in Brush Biopsies of Oral Potentially Malignant Lesions of FA and non-FA Patients</b> <i>Nadarajah Vigneswaran, University of Texas School of Dentistry at Houston, United States</i>	<i>p. 9</i>
<b>9:15 – 9:25</b> 9:25 – 9:30 Q&A	<b>Quercetin Decreases the Proliferation and Causes Cell Death in Fanconi Anemia Deficient Head and Neck Cancer Cell Lines In Vitro</b> <i>Ruby Khoury, Cincinnati Children's Hospital Medical Center, Cincinnati, United States</i>	<i>p. 10</i>
<b>9:30 – 9:40</b> 9:40 – 9:45 Q&A	<b>Identification of a Therapeutic Strategy Targeting Fanconi Anemia (FA) Pathway Defects in Cancers</b> <i>Ali Suliman, Yale University School of Medicine, New Haven, United States</i>	<i>p. 11</i>
<b>9:45 – 9:50</b>	<b>Session Wrap-up: Agata Smogorzewska</b>	
<b>9:50 – 10:10</b>	<b>Break</b>	
<b>10:10 – 12:00</b> <i>Grand Ballroom 2&amp;3</i>	<b>Cancer Immunotherapy</b> <i>Chair: Premal Patel, Immuno-Oncology Early Development, Pfizer, Redwood City, United States</i>	
<b>10:10 – 10:15</b>	<b>Session Overview: Premal Patel</b>	
<b>10:15 – 10:40</b> 10:40 – 10:45 Q&A	<b>Advances in Immuno-Oncology</b> <i>Premal Patel</i>	<i>p. 12</i>
<b>10:45 – 11:10</b> 11:10 – 11:15 Q&A	<b>Effects of Head and Neck Tumor Heterogeneity on Immune Response</b> <i>John Sunwoo, Stanford University, United States</i>	<i>p. 12</i>
<b>11:15 – 11:40</b> 11:40 – 11:45 Q&A	<b>Keynote Address: Immune Escape by Head and Neck Cancer and Therapeutic Reversal</b> <i>Robert Ferris, University of Pittsburg, United States</i>	<i>p. 7</i>
<b>11:45 – 12:00</b>	<b>Panel Discussion: All Speakers</b>	
<b>12:00 – 2:00</b> <i>Grand Ballroom 1</i>	<b>Lunch Buffet</b>	
<b>12:15 – 2:00</b> <i>Grand Ballroom 1</i>	<b>Mentorship Lunch for Early Investigators</b>	
<b>2:00 – 2:35</b> <i>Grand Ballroom 2&amp;3</i>	<b>Rare Disease Research and Development: The NF Story</b>	
<b>2:00 – 2:05</b>	<b>Introduction: Maureen Hoatlin, Oregon Health &amp; Science University, Portland, United States</b>	
<b>2:05 – 2:30</b> 2:30 – 2:35 Q&A	<b>The Children's Tumor Foundation Business Model: an Emerging Niche in the Rare Disease Ecosystem</b> <i>Salvatore La Rosa, Children's Tumor Foundation, New York, United States</i>	<i>p. 13</i>
<b>2:35 – 5:00</b> <i>Grand Ballroom 2&amp;3</i>	<b>Preclinical Models</b> <i>Chair: Elaine Ostrander, National Human Genome Research Institute, NIH, Bethesda; Scientific Advisory Board, Fanconi Anemia Research Fund, United States</i>	
<b>2:35 – 2:40</b>	<b>Session Overview: Elaine Ostrander</b>	
<b>2:40 – 3:05</b> 3:05 – 3:10 Q&A	<b>Engineering Hematopoietic Stem Cells from Human IPS Cells – Application to Fanconi Anemia</b> <i>Grant Rowe, Boston Children's Hospital, Boston, United States</i>	<i>p. 14</i>
<b>3:10 – 3:20</b> 3:20 – 3:25 Q&A	<b>Porcine Model of FANCA</b> <i>William H. Fleming, Oregon Health &amp; Science University, Portland, United States</i>	<i>p. 15</i>
<b>3:25 – 3:35</b> 3:35 – 3:40 Q&A	<b>Identification and Characterization of a Blood Stem Cell-Enriched Cell Population for Gene Therapy</b> <i>Stefan Radtke, Fred Hutchinson Cancer Research Center, Seattle, United States</i>	<i>p. 16</i>
<b>3:40 – 3:45</b>	<b>Session Wrap-up: Elaine Ostrander</b>	
<b>3:45 – 4:05</b>	<b>Break</b>	

<b>4:05 – 4:15</b>	<b>Fanconi Anemia: The Bone Side of the Disease</b> <i>Melody Mazon, Laval University, Québec, Canada</i>	p. 17
<b>4:15 – 4:25</b>	<b>Fanconi Anemia (FA) Fancd2<sup>-/-</sup> Mouse Bone Marrow Stromal Cells Demonstrate Increased Irradiation Induced Senescence</b> <i>Aranee Sivananthan, University of Pittsburg, United States</i>	p. 18
<b>4:25 – 4:35</b>	<b>DNA Crosslink Repair is Essential for Germ Cell Development</b> <i>Gerry Crossan, MRC Laboratory of Molecular Biology, Cambridge, England</i>	p. 19
<b>4:35 – 4:45</b>	<b>A FANCI Knock-out Mouse Model Supports New Roles for FANCI, Independently of FANCD2</b> <i>Laure Guitton, Centre de Recherche du CHU de Québec, Canada</i>	p. 20
<b>4:45 – 5:00</b>	<b>Panel Discussion and Wrap-up</b>	
<b>5:30 – 7:30</b> <i>Prefunction and Buckhead Ballroom</i>	<b>Poster Reception</b>  <i>Presenters of even-numbered posters will be at their posters from 5:30 to 6:30 Presenters of odd-numbered posters will be at their posters from 6:30 to 7:30</i>	
<b>7:30 – 9:30</b> <i>Grand Ballroom 2&amp;3</i>	<b>Symposium Banquet</b>	

## Saturday, 16

<b>7:00 – 8:30</b> <i>Grand Ballroom 1</i>	<b>Breakfast</b>	
<b>8:30 – 9:55</b> <i>Grand Ballroom 2&amp;3</i>	<b>The FA Pathway: Functions &amp; Interactions – Part 1</b> <i>Chair: Alan D'Andrea, Dana-Farber Cancer Institute, Harvard Medical School, Boston, United States</i>	
<b>8:30 – 8:35</b>	<b>Session Overview: Alan D'Andrea</b>	
<b>8:35 – 8:45</b> 8:45 – 8:50 Q&A	<b>FANCD2 and FANCI Function Independently During the Cellular Replication Stress Response</b> <i>Jung Eun Yeo, Ulsan National Institute of Science and Technology, Ulju-gun, South Korea</i>	p. 21
<b>8:50 – 9:00</b> 9:00 – 9:05 Q&A	<b>FANCB Mediates Dimerization and Subcellular Localization of the Fanconi Anemia Core Complex</b> <i>Sylvie van Twest, St Vincent's Institute of Medical Research, Fitzroy, Australia</i>	p. 22
<b>9:05 – 9:15</b> 9:15 – 9:20 Q&A	<b>Identification of the UHRF2 (RNF107) Protein as an ICL Sensor, Assisting in Recruiting FANCD2 to Chromatin</b> <i>Anna Motnenko, University of Oxford, United Kingdom</i>	p. 23
<b>9:20 – 9:30</b> 9:30 – 9:35 Q&A	<b>The Replisome Promotes Hierarchical Usage of Two Replication-Coupled ICL Repair Pathways</b> <i>Dan Semlow, Harvard University, Boston, United States</i>	p. 24
<b>9:35 – 9:45</b> 9:45 – 9:50 Q&A	<b>Regulation of FANCD2 Monoubiquitination and Function in Replication Fork Maintenance via a Conserved Phosphosite Cluster Proximal to K561</b> <i>David Vierra, University of Rhode Island, Newport, United States</i>	p. 25
<b>9:50 – 9:55</b>	<b>Session Wrap-up: Alan D'Andrea</b>	
<b>9:55 – 10:15</b>	<b>Break</b>	

<b>10:15 – 11:55</b> Grand Ballroom 2&3	<b>The FA Pathway: Functions &amp; Interactions – Part 2</b> Chair: Niall Howlett, University of Rhode Island, Newport, United States	
<b>10:15 – 10:20</b>	<b>Session Overview: Niall Howlett</b>	
<b>10:20 – 10:30</b> 10:30 – 10:35 Q&A	<b>FANCD2 Cooperates with the ATRX/DAXX Chromatin Remodeler to Promote HR-Mediated Replication Fork Recovery</b> Maya Raghunandan, University of Minnesota, Minneapolis, United States	p. 26
<b>10:35 – 10:45</b> 10:45 – 10:50 Q&A	<b>Fanconi Anaemia Deficient Cells Rely on Transcription Coupled Repair for Survival</b> Jamie Langton, University of Oxford, United Kingdom	p. 27
<b>10:50 – 11:00</b> 11:00 – 11:05 Q&A	<b>Acetaldehyde DNA Interstrand Crosslinks are Repaired via the Fanconi Anemia Pathway and a Potentially Novel Pathway</b> Puck Knipscheer, Hubrecht Institute, Utrecht, Netherlands	p. 28
<b>11:05 – 11:15</b> 11:15 – 11:20 Q&A	<b>Evidence of Interferon Stress in Hematopoietic Stem Cells (HSC) in Response to Increased Aldehydic Load</b> Lauren Van Wassenhove, Stanford University, United States	p. 29
<b>11:20 – 11:30</b> 11:30 – 11:35 Q&A	<b>Fatty Aldehyde Dehydrogenase as a Novel Binding Partner Of FANCD2</b> Wataru Sakai, Kobe University, Japan	p. 30
<b>11:35 – 11:45</b> 11:45 – 11:50 Q&A	<b>A Protein Folding Way to Precision Health for Fanconi Anemia</b> Georgios Karras, Whitehead Institute for Biomedical Research, Cambridge, United States	p. 31
<b>11:50 – 11:55</b>	<b>Session Wrap-up: Niall Howlett</b>	
<b>12:00 – 2:00</b> Grand Ballroom 1	<b>Lunch Buffet</b>	
<b>2:00 – 3:20</b> Grand Ballroom 2&3	<b>Advances in Transplantation</b> Chair: John Wagner, University of Minnesota, Minneapolis, United States	
<b>2:00 – 2:05</b>	<b>Session Overview: John Wagner</b>	
<b>2:05 – 2:15</b>	<b>Engraftment and Repopulation Advantage of Gene-Corrected Hematopoietic Precursors in Non-Conditioned Fanconi Anemia Patients</b> Paula Rio, CIEMAT/CIBERER/IIS-Fundación Jimenez Diaz (IIS-FJD, UAM), Madrid, Spain	p. 32
<b>2:15 – 2:25</b>	<b>Preliminary Conclusions of a Clinical Trial for The Mobilization Of Hematopoietic Stem Cells from Fanconi Anemia Patients with Plerixafor and Filgrastim</b> Susana Navarro, CIEMAT/CIBERER/IIS-Fundación Jimenez Diaz (IIS-FJD, UAM), Madrid, Spain	p. 33
<b>2:25 – 2:35</b>	<b>StemRegenin-1 Expanded Umbilical Cord Blood Hematopoietic Stem Cell Transplantation for Fanconi Anemia</b> John Wagner	p. 34
<b>2:35 – 2:45</b>	<b>Excellent Outcome For 91 Fanconi Anemia Patients Undergoing Matched Related Transplants Using Cyclophosphamide 60mg/Kg in Brazil</b> Carmem Bonfim, Federal University of Paraná, Curitiba, Brazil	p. 35
<b>2:45 – 2:55</b>	<b>Allogeneic Stem Cell Transplantation Using HLA Matched Donors in Patients with Fanconi Anaemia – Single Centre Experience from India</b> Biju George, Christian Medical College, Vellore, India	p. 36
<b>2:55 – 3:05</b>	<b>TGF-<math>\beta</math> Pathway Inhibition by AVID200 Rescues Genotoxicity in Hematopoietic Stem and Progenitor Cells from Fanconi Anemia Mice</b> Alfredo De Jesús Rodríguez Gómez, Dana Farber Cancer Institute/Harvard Medical School, Boston, United States	p. 37
<b>3:05 – 3:20</b>	<b>Panel Discussion: All Speakers</b>	

**3:20 – 3:40**      **Break**

**3:40 – 5:05**      **Genetics & Gene Editing**  
*Grand Ballroom 2&3*  
*Chair: Richard Gelinas, Institute for Systems Biology, Seattle, United States; Board of Directors and Scientific Advisory Board, Fanconi Anemia Research Fund*

**3:40 – 3:45**      **Session Overview: Richard Gelinas**

**3:45 – 3:55**      **Biallelic Mutations in the Ubiquitin Ligase RFD3 Cause Fanconi Anemia**      *p. 38*  
3:55 – 4:00 Q&A      *Detlev Schindler, University of Wurzburg, Germany*

**4:00 – 4:10**      **Identification of 8 Patients with Biallelic Mutations in FANCM with Normal Hematology, Early On Set Cancer and Hypertoxicity to Chemotherapy**      *p. 39*  
4:10 – 4:15 Q&A      *Jordi Surrallés, Universitat Autònoma Barcelona/Hospital Sant Pau, Barcelona, Spain*

**4:15 – 4:25**      **FANCN Patient Mutations are Hypomorphic and Rescue Catastrophic Genomic Instability**      *p. 40*  
4:25 – 4:30 Q&A      *Elizabeth Thompson, University of Minnesota, Minneapolis, United States*

**4:30 – 4:40**      **CRISPR-Cas9 Genome Editing in Human Cells Works via the Fanconi Anemia Pathway**      *p. 41*  
4:40 – 4:45 Q&A      *Chris Richardson, UC Berkeley, United States*

**4:45 – 4:55**      **NHEJ-Mediated Gene Editing: A New Approach to Correct Different Mutations in Fanconi Anemia Hematopoietic Stem and Progenitor Cells**      *p. 42*  
4:55 – 5:00 Q&A      *Francisco J. Roman-Rodriguez, CIEMAT/CIBERER/IIS-Fundación Jimenez Diaz (IIS-FJD, UAM), Madrid, Spain*

**5:00 – 5:05**      **Session Wrap-up: Richard Gelinas**

*Sunday, 17*

**7:00 – 8:30**      **Breakfast**  
*Grand Ballroom 1*

**8:30 – 10:25**      **Rare Disease Research & Drug Development**  
*Grand Ballroom 2&3*  
*Chair: Maureen Hoatlin, Oregon Health & Science University, Portland, United States*

**8:30 – 8:35**      **Session Overview: Maureen Hoatlin**

**8:35 – 9:00**      **The Translator Project: Turning Biomedical Data Into Knowledge**      *p. 43*  
9:00 – 9:05 Q&A      *Christine Colvis, Drug Development Partnership Programs, National Institutes of Health, Bethesda, United States*

**9:05 – 9:15**      **Data Translator: an Open Science Data Platform for Mechanistic Disease Discovery**      *p. 43, 105*  
9:15 – 9:20 Q&A      *Melissa Haendel, Oregon Health & Science University, Portland, United States*

**9:20 – 9:30**      **A Data Translator for Fanconi Anemia**      *p. 43, 105*  
9:30 – 9:35 Q&A      *Maureen Hoatlin*

**9:35 – 10:00**      **Challenges and Opportunities in Rare Disease Drug Development**      *p. 44*  
10:00 – 10:05 Q&A      *Jeffrey Siegel, Genentech, San Francisco, United States*

**10:05 – 10:15**      **Enabling Clinical Trials and Translational Research Through a Fanconi Database**      *p. 43*  
10:15 – 10:20 Q&A      *Erica Jonlin, University of Washington, Seattle, United States*

**10:20 – 10:25**      **Session Wrap-up: Maureen Hoatlin**

**10:25 – 10:45**      **Break**

**10:45 – 11:45**      **Living With FA: Symposium Recap & Research Priorities**  
*Co-chairs: Ray Monnat Jr., University of Washington, Seattle, United States; Chair of Scientific Advisory Board, Fanconi Anemia Research Fund*  
*Bradley D. Preston, Scientific Director, Fanconi Anemia Research Fund, Seattle, United States*

