

# SYMPOSIUM AGENDA

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**Registration** will take place in Grand Ballroom Foyer  
**General session** (all talks) will take place in Grand Ballroom  
**Poster receptions** will take place in Grand Ballroom & Grand Ballroom Foyer  
**Breakfasts & lunches** will take place in Explore  
**Banquet dinner** will take place in Grand Ballroom

Note: regular talks are 15 minutes: 10 for the presenter, 5 for Q & A

## THURSDAY 19TH

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- 4:00 pm Registration
- 5:00 - 5:10 Welcome  
*Mark Quinlan, Fanconi Anemia Research Fund, United States*
- 5:10 - 6:30 Opening Session: "It is not in the stars to hold our destiny but in ourselves."**  
*Chair: Stella Davies, Cincinnati Children's Hospital, United States*
- 5:10 - 5:30 *Jack Timperley, Adult with FA, Illinois, United States*
- 5:30 - 5:50 *Lindsey Romick-Rosendale, Cincinnati Children's Hospital, United States*
- 5:50 - 6:10 *Rutger Boerema, FA Parent, Pennsylvania, United States*
- 6:10 - 6:25 *Margaret MacMillan, University of Minnesota, United States*
- 6:25 - 6:30 *Stella Davies, Cincinnati Children's Hospital, United States*
- 6:30 - 8:30 Poster Reception  
*Presenters of odd-numbered posters will be at their posters 6:30 - 7:30 pm*  
*Presenters of even-numbered posters will be at their posters 7:30 - 8:30 pm*

## FRIDAY 20TH

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- 7:00 - 8:00 Breakfast
- 8:00 - 9:55 The FA Pathway: Structure and Function**
- 8:00 - 8:05 Session Overview  
*Raymond Monnat, University of Washington, United States*
- 8:05 - 8:35 Keynote Address: TGF $\beta$  Inhibitors Rescue Bone Marrow Failure in Fanconi Anemia p. 1  
*Alan D'Andrea, Dana-Farber Cancer Institute, United States*
- 8:35 - 8:50 Functional crosstalk between the Fanconi anemia and ATRX/DAXX histone chaperone pathways promotes replication for recovery p. 4  
*Alexandra Sobeck, University of Minnesota, United States*

8:50 - 9:05	WRNIP1 is recruited to DNA interstrand crosslinks and promotes repair <i>Di Yang, Oxford University, United Kingdom</i>	p. 5
9:05 - 9:20	FANCG and FANCA have distinct opposing functions in double strand break end resection <i>Niraj Joshi, Dana-Farber Cancer Institute, United States</i>	p. 6
9:20 - 9:25	Keynote Introduction <i>Raymond Monnat, University of Washington, United States</i>	
9:25 - 9:55	Keynote Address: A NextGen Approach for Quantifying FA Pathway Activity <i>Amanda Paulovich, Fred Hutchinson Cancer Center, University of Washington, United States</i>	p. 2
9:55 - 10:15	Break	
<b>10:15 - 12:25</b>	<b>The FA Pathway, continued</b>	
10:15 - 10:30	Defining and rescuing FA mutant biological defects <i>John Tainer, University of Texas MD Anderson Cancer Center, United States</i>	p. 8
10:30 - 10:45	UAF1 DNA binding activity is critical for RAD51-mediated homologous DNA Pairing <i>Fengshen Liang, Yale University School of Medicine, United States</i>	p. 9
10:45 - 11:00	CDK1 regulates the mitotic and DNA damage repair functions of FANCC <i>Elizabeth Sierra Potchanant, Indiana University, United States</i>	p. 11
11:00 - 11:15	FANCD2 tunes the unfolded protein response preventing mitochondrial stress-induced common fragile site gene expression and instability <i>Valeria Naim, University Paris-Sud, Gustave Roussy, France</i>	p. 12
11:15 - 11:30	Mitochondrial DNA replication fork protection by the Fanconi anemia pathway suppresses inflammation and disease <i>Jessica Luzwick, University of Texas MD Anderson Cancer Center, United States</i>	p. 13
11:30 - 11:45	Dissecting Gene by Environment Relationships in Fanconi Anemia <i>Melody Mazón, University of Texas MD Anderson Cancer Center, United States</i>	p. 14
11:45 - 12:20	Special Panel: Targeting the FA Pathway <i>Alan D'Andrea, Dana-Farber Cancer Institute, United States</i> <i>Amanda Paulovich, Fred Hutchinson Cancer Center, University of Washington, United States</i> <i>John Trainer, University of Texas MD Anderson Cancer Center, United States</i> <i>Joel Greenberger, University of Pittsburgh, United States</i>	
12:20 - 12:25	Session Wrap-up <i>Raymond Monnat, University of Washington, United States</i>	
12:25 - 2:00	Lunch Mentorship Lunch	

<b>2:00 - 2:35</b>	<b>Central Nervous System Abnormalities in Fanconi Anemia</b>	
2:00 - 2:05	Session Overview <i>Stella Davies, Cincinnati Children's Hospital, United States</i>	
2:05 - 2:20	Neuroinflammation in Fanconi Anemia: The Role of Polyoma Virus <i>Stella Davies, Cincinnati Children's Hospital, United States</i>	p. 15
2:20 - 2:35	Cranial MRI Findings of Patients with Fanconi Anemia <i>Tekin Aksu, Hacettepe University, Ankara, Turkey</i>	p. 17
<b>2:35 - 3:10</b>	<b>FARF Tank 2018 Winners</b>	
2:35 - 2:50	Addition of mild hyperthermia to improve treatment of FA/BRCA pathway deficient head and neck cancer in FA patients <i>Lianne Vriend, Amsterdam UMC, Netherlands</i>	p. 19
2:50 - 3:05	The FINished Product: A FARF Tank Update on Serotonin as a Novel Target in Fanconi Anemia <i>Allison Bartlett, Cincinnati Children's Hospital, United States</i>	p. 20
3:05 - 3:10	Session Wrap-up <i>Stella Davies, Cincinnati Children's Hospital, United States</i>	
3:10 - 3:30	Break	
<b>3:30 - 5:00</b>	<b>FARF Tank</b>	
5:00 - 5:15	FARF Tank Voting	
5:30 - 7:30	Poster Reception <i>Presenters of even-numbered posters will be at their posters 5:30 - 6:30 pm</i> <i>Presenters of odd-numbered posters will be at their posters 6:30 - 7:30 pm</i>	
7:30 - 9:30	Symposium Banquet	

## SATURDAY 21ST

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7:00 - 8:30	Breakfast	
<b>8:30 - 10:35</b>	<b>Cancer in Fanconi Anemia</b>	
8:30 - 8:35	Session Overview <i>Agata Smogorzewska, The Rockefeller University, United States</i>	
8:35 - 9:05	Keynote Address: Chemoprevention of Head and Neck Cancer with Natural Products or NSAIDs <i>Daniel E. Johnson, University of California San Francisco, United States</i>	p. 3

9:05 - 9:20	Diagnostic accuracy of oral brush cytology and analysis of DNA ploidy in visible oral lesions of patients with Fanconi Anemia <i>Eunike Velleuer, University of Duesseldorf; Children's Hospital Moenchengladbach Neuwerk, Germany</i>	p. 22
9:20 - 9:35	Functional genomics to identify vulnerabilities in Fanconi Anaemia head and neck cancers <i>M. Govind Pai, Amsterdam UMC, Netherlands</i>	p. 23
9:35 - 9:50	Genomic and transcriptomic components of Fanconi anemia head and neck squamous cell carcinoma-derived cell models <i>Khashayar Roohollahi, Amsterdam UMC, Netherlands</i>	p. 24
9:50 - 10:05	Fanconi Anemia Tumorigenesis is Characterized by Complex Somatic Structural Variation <i>Agata Smogorzewska, The Rockefeller University, United States</i>	p. 26
10:05 - 10:20	Genetic variation in the functionally interacting human Fanconi, alcohol and aldehyde dehydrogenase gene families <i>Nyasha Chambwe, Institute for Systems Biology, United States</i>	p. 28
10:20 - 10:35	Developing a chemical tool kit to identify and measure intracellular aldehydes <i>Hyun Shin Park, Stanford University, United States</i>	p. 29
10:35 - 10:55	Break	
<b>10:55 - 12:15</b>	<b>Cancer in Fanconi Anemia continued</b>	
10:55 - 11:10	Safety and pharmacokinetics of FP-045, an ALDH2 activator for Fanconi anemia treatment, in healthy human subjects <i>David Lau, Foresee Pharmaceuticals Co., Ltd, United States</i>	p. 31
11:10 - 11:25	Quercetin chemoprevention for SCC in FA patients <i>Parinda Mehta, Cincinnati Children's Hospital, United States</i>	p. 32
11:25 - 11:40	Characterization of the FA pathway in triple negative breast cancer (TNBC) reveals vulnerability to PARP inhibitors and a role for FANCD2 in mitochondria <i>Nigel Jones, University of Liverpool, United Kingdom</i>	p. 34
11:40 - 11:55	The repair of radiation-induced DNA double strand breaks in Fanconi anemia fibroblasts in G1- and G2-phase <i>Sebastian Zahnreich, University Medical Center, Mainz, Germany</i>	p. 36
11:55 - 12:10	Porcine models for Fanconi anemia <i>William Fleming, Oregon Health Sciences University, United States</i>	p. 38
12:10 - 12:15	Session Wrap-up <i>Agata Smogorzewska, The Rockefeller University, United States</i>	
12:15 - 1:30	Lunch	

## 1:30 - 3:55 **Gene Therapy and Editing in Fanconi Anemia**

*Note: questions usually following each presentation will be held at the end of each section*

- 1:30 - 1:35 **Session Overview**  
*Juan Bueren, Division of Hematopoietic Innovative Therapies, CIEMAT/CIBERER and Advanced Therapies Unit, Spain*
- 1:35 - 1:45 **Conclusions of the FANCOSTEM-I Clinical Trial for the Collection of Filgrastim and Plerixafor Mobilized Hematopoietic Stem Cells from Fanconi Anemia Patients** p. 39  
*Julián Sevilla, Hospital Infantil Universitario Niño Jesús, Spain*
- 1:45 - 1:55 **Gene therapy for Fanconi Anemia Patients: Results of the Clinical Trial After a Follow-Up of 2-3 Years** p. 40  
*Juan Bueren, Division of Hematopoietic Innovative Therapies, CIEMAT/CIBERER and Advanced Therapies Unit, Spain*
- 1:55 - 2:05 **A Phase I Study of Lentiviral-mediated Ex-vivo Gene Therapy for Pediatric Patients with Fanconi Anemia, Complementation Group-A: Initial Patient Experience** p. 42  
*Agnieszka Czechowicz, Stanford University, United States*
- 2:05 - 2:15 **Reverse Mosaicism is associated with improved outcomes in Fanconi anemia** p. 44  
*Jordi Surrallés, Universitat Autònoma de Barcelona / CIBERER, Madrid / Institute of Biomedical Research and Department of Genetics, Sant Pau Hospital, Barcelona, Spain*
- 2:15 - 2:25 **Mosaicism and Fanconi Anemia: Review of Clinical Outcomes in Published Cases** p. 45  
*Jonathan Schwartz, Rocket Pharma, United States*
- 2:25 - 2:55 **Panel discussion with Q & A**
- 2:55 - 3:05 **Hematopoietic Stem Cell Expansion** p. 47  
*John Wagner, University of Minnesota, United States*
- 3:05 - 3:15 **Relevance of Innate Immunity and Vector Signaling for Fanconi Anemia Hematopoietic Stem Cell Gene Therapy** p. 48  
*Anna Kajaste, San Rafael Telethon Institute for Gene Therapy (SR-Tiget), Italy*
- 3:15 - 3:25 **A novel nanoparticle that can deliver CRISPR gene edits to blood stem and progenitor cells in one, passive step** p. 49  
*Jennifer Adair, University of Washington, United States*
- 3:25 - 3:55 **Panel discussion with Q & A**
- 3:55 - 4:15 **Break**
- 4:15 - 5:20 **Gene Therapy and Editing in Fanconi Anemia continued**
- Note: questions usually following each presentation will be held at the end of each section*
- 4:15 - 4:25 **Fanconi Anemia Gene Editing via Programmable DNA Modifying Enzymes** p. 51  
*Mark Osborn, University of Minnesota, United States*

4:25 - 4:35	In-vivo gene therapy for Fanconi anemia <i>Hans-Peter Kiem, Fred Hutchinson Cancer Center, University of Washington, United States</i>	p. 52
4:35 - 4:45	Improved Efficiency of Therapeutic NHEJ-Mediated Gene Editing in a Variety of Fanconi Anemia Complementation Groups <i>Paula Río, Division of Hematopoietic Innovative Therapies, CIEMAT/CIBERER and Advanced Therapies Unit, Spain</i>	p. 53
4:45 - 5:15	<b>Panel discussion with Q &amp; A</b>	
5:15 - 5:20	Session Wrap-up <i>Juan Bueren, Division of Hematopoietic Innovative Therapies, CIEMAT/CIBERER and Advanced Therapies Unit, Spain</i>	

## SUNDAY 22ND

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7:00 - 8:00	Breakfast	
<b>8:00 - 9:40</b>	<b>Bone Marrow Transplantation: Novel Treatment Paradigms</b>	
8:00 - 8:05	Session Overview <i>John Wagner, University of Minnesota, United States</i>	
8:05 - 8:20	Pilot Study of Metformin in Fanconi Anemia <i>Akiko Shimamura, Dana-Farber/Boston Children's Cancer and Blood Disorders Center, United States</i>	p. 5
8:20 - 8:35	Treatment of optimized dose querectin leads to sustained reduction in oxidative stress in persons with Fanconi anemia <i>Parinda Mehta, Cincinnati Children's Hospital Medical Center, United States</i>	p. 57
8:35 - 8:50	Misdiagnosis and eltrombopag treatment in a Fanconi anemia patient <i>Josune Zubicaray, Fundación Investigación Biomédica, Hospital Infantil Universitario Niño Jesús, Spain</i>	p. 59
8:50 - 9:05	Viral specific T cells to treat viral infections in FA patients post-HSCT <i>Adam Nelson, Cincinnati Children's Hospital Medical Center, United States</i>	p. 61
9:05 - 9:20	Haploidentical transplantation with post-transplant cyclophosphamide (Haplo-PTCy) for 53 patients with Fanconi anemia: The experience from Curitiba Brazil <i>Carmem Bonfim, Federal University of Paraná, Brazil</i>	p. 62
9:20 - 9:35	Antibody Conditioning <i>Agnieszka Czechowicz, Stanford University, United States</i>	p. 63
9:35 - 9:40	Session Wrap-up <i>John Wagner, University of Minnesota, United States</i>	
9:40 - 10:00	Break	

**10:00 - 11:10 Bone Marrow Failure: From Models to Mechanisms**

10:00 - 10:05 Session Overview

*Akiko Shimamura, Dana-Farber/Boston Children's Cancer and Blood Disorders Center, United States*

10:05 - 10:20 An induced pluripotent stem cell model of bone marrow failure in Fanconi anemia p. 64

*Grant Rowe, Boston Children's Hospital, United States*

10:20 - 10:35 Apoptosis pathways initiated during erythroid replicative phase of emergency granulopoiesis trigger bone marrow failure in Fanconi Anemia p. 65

*Shirin Hasan, Northwestern University, United States*

10:35 - 10:50 Single cell RNA sequencing reveals MYC as a novel player in the pathogenesis of bone marrow failure in Fanconi anemia p. 66

*Alfredo Rodríguez, Dana-Farber Cancer Institute, Harvard University, United States*

10:50 - 11:05 Protection of hematopoietic stem cells by pharmacological activation of aldehyde dehydrogenase 2 (ALDH2) in murine models of Fanconi anemia p. 67

*Jennifer Tsai, Stanford University, United States*

11:05 - 11:10 Session Wrap-up

*Akiko Shimamura, Dana-Farber/Boston Children's Cancer and Blood Disorders Center, United States*

**11:10 - 12:00 Symposium Town Hall: an Interactive Discussion about FA Research**