

VIRTUAL CONFERENCE
Translational Research Conference

BONE MARROW FAILURE DISORDERS:
From the cell to the cure of the disease

November 13-15, 2020

Chairpersons : Tim Brümmendorf, Inderjeet Dokal, Carlo Dufour, Régis Peffault de Latour,
Marc H. G. P. Raaijmakers, Neal Young

Friday, November 13, 2020

09:30–09:40 Welcome

SESSION I – STEM CELL BIOLOGY: FROM STRESS TO FAILURE

Chairs: Tim Brümmendorf and Markus Manz

09:40-09:45	Introduction	Tim Brümmendorf & Markus Manz
09:45-10:00	Haemopoietic stem cell activation and failure	Andreas Trumpp (Heidelberg)
10:00-10:15	Genotoxic stress and repair in stem cells	Ketan J. Patel (Cambridge)
10:15-10:30	Inflammageing of the haemopoietic system	Markus Manz (Zürich)

SELECTED BRIEF ORAL COMMUNICATION

10:30-10:40 *Immunologic Basis to Poor Graft Function Post Allogeneic Transplantation.* Ashvind Prabahan (Melbourne)

10:40-11:10 *Discussion*

11:10-11:20 Coffee Break

SESSION II – CONSTITUTIONAL MARROW FAILURE 1: FANCONI ANAEMIA (FA)

Chairs: Carlo Dufour and Inderjeet Dokal

11:20-11:25 Introduction Carlo Dufour & Inderjeet Dokal

LIVE Programme

CET Time

11:25-11:40	Fanconi anaemia : diagnostic and natural history in the bone marrow	Jean Soulier (Paris)
11:40-11:55	Classical and new treatment of Fanconi anaemia	Carlo Dufour (Genova)
11:55-12:10	Gene therapy for Fanconi anaemia	Juan Bueren (Madrid)

SELECTED BRIEF ORAL COMMUNICATION

12:10-12:20 *Genetic Rescue of Fanca-/- HSPCs with Evi-1 Overexpression Suggests a Mechanistic Link with DNA Damage Response* Susanne Lux (Heidelberg)

12:20-12:50 *Discussion*

12:50-14:00 Lunch

SESSION III – CONSTITUTIONAL MARROW FAILURE 2: RIBOSOME DISEASES

Chairs: Marc H. G. P. Raaijmakers and Alan Warren

14:00-14:05	Introduction	Marc H.G.P. Raaijmakers & Alan Warren
14:05-14:20	Diamond-Blackfan anaemia (DBA), ribosomes and haemopoiesis	Vijay Sankaran (Boston)
14:20-14:35	Shwachman-Diamond syndrome (SDS) and related ribosomopathies	Alan Warren (Cambridge)
14:35-14:50	Management of ribosomopathies	Yigal Dror (Toronto)

SELECTED BRIEF ORAL COMMUNICATION

14:50-15:00 *A Single Base Change Makes the Cure: Correcting the Most Common SBDS Mutation in Shwachman-Diamond Syndrome* Chi-Yuan Zhang (Boston)

15:00-15:30 *Discussion*

15:30-15:40 Coffee break

SESSION IV – CONSTITUTIONAL MARROW FAILURE 3: TELOMERE BIOLOGY DISEASES

Chairs: Inderjeet Dokal and Tim Brummendorf

15:40-15:45	Introduction	Inderjeet Dokal & Tim Brummendorf
15:45-16:00	Telomeres and telomerase in health and ageing	Alison Bertuch (Houston)
16:00-16:15	Dyskeratosis congenita and related haematological diseases	Inderjeet Dokal (London)
16:15-16:30	Diagnosis and clinical presentation of adult onset telomeropathies	Fabian Beier (Aachen)

LIVE Programme
CET Time

SELECTED BRIEF ORAL COMMUNICATION

- 16:30-16:40 *Natural History and Outcomes Depending on Organ System Involvement in Patients with Clinically-Relevant Short Telomeres* *Abhishek Mangaonkar (Rochester)*
- 16:40-17:10 *Discussion*
- 17:15-18:15 **Poster walks**
Leaders: Tim Brümmendorf and Marc H.G.P. Raaijmakers

Saturday, November 14, 2020

12:00-13:00 3 Simultaneous Meet the Expert sessions

1. Role of Next Generation Sequencing in the diagnosis and follow-up of patients with BMF *Marc H.G.P. Raaijmakers & Jean Soulier*
2. Management of challenging situations in PNH *Britta Höchsmann & Rosario Notaro*
3. How to properly diagnose and treat telomere biology disorders in children and adults *Tim Brümmendorf & Inderjeet Dokal*

13:05-14:05 SATELLITE SYMPOSIUM:

Title: Beyond current endpoints: a patient-centric approach to PNH treatment

Chair: Régis Peffault de Latour

Speakers: Pascale Burmester, Talha Munir and Jens Panse

This symposium is organized by Roche

SESSION V – IMMUNE APLASTIC ANAEMIA AND PAROXYSMAL NOCTURNAL HAEMOGLOBINURIA (PNH) 1. PATHOPHYSIOLOGY AND DIAGNOSIS

Chairs: Neal Young and Régis Peffault de Latour

- 14:10-14:15 Introduction *Neal Young & Régis Peffault de Latour*
- 14:15-14:30 Pathophysiology of immune aplastic anaemia *Neal Young (Bethesda)*
- 14:30-14:45 Pathophysiology of PNH *Rosario Notaro (Florence)*
- 14:45-15:00 Modern diagnosis of aplastic anaemia and PNH *Austin Kulasekararaj (London)*

LIVE Programme
CET Time

SELECTED BRIEF ORAL COMMUNICATION

15:00-15:10 *Single Cell eQTL Mapping Identifies Cell-Type Specific Control of Autoimmune Disease* Kirsten Fairfax (Hobart)

15:10-15:35 *Discussion*

15:40-16:40 SATELLITE SYMPOSIUM:

Title: A journey through the SAA and PNH treatment landscapes

Chair: Régis Peffault de Latour and Antonio Risitano

Speakers: Régis Peffault de Latour and Antonio Risitano

This symposium is organized by Novartis

SESSION VI – IDIOPATHIC APLASTIC ANAEMIA AND PAROXYSMAL NOCTURNAL HAEMOGLOBINURIA (PNH) 2. DIFFERENTIAL DIAGNOSIS AND CLONAL EVOLUTION

Chairs: Neal Young and Jaroslaw P. Maciejewski

16:45-16:50 Introduction Neal Young & Jaroslaw P. Maciejewski

16:50-17:20 Keynote Presentation: Lucio Luzzatto (Dar-es-Salaam)

Bone marrow failure and somatic mutations: the PNH paradigm

17:20-17:30 *Discussion*

17:30-17:45 Innate immunity and clonal evolution in bone marrow failure Alan List (Tampa)

17:45-18:00 Impact of clonal haemopoiesis in aplastic anaemia Jaroslaw P. Maciejewski (Cleveland)

18:00-18:15 Health-related quality of life and its assessment in aplastic anaemia and paroxysmal nocturnal haemoglobinuria Jens Panse (Aachen)

SELECTED BRIEF ORAL COMMUNICATION

18:15-18:25 *The Metabolomic Status of the Differentiating Myeloid Lineage in MDS with Low and High Bone Marrow Blast Counts.* Aikaterini Poulaki (Athens)

18:25-18:55 *Discussion*

19:00-20:00 3 Simultaneous Meet the Expert Sessions

4. Source and sequence of BMT in acquired aplastic anaemia Andrea Bacigalupo & Neal Young
5. When and how to do BMT in inherited bone marrow failure syndromes? Yigal Dror & Carlo Dufour
6. Surveillance of leukaemia predisposition in bone marrow failure: How, and when to act? Lucy Godley & Jaroslaw P. Maciejewski

Sunday, November 15, 2020

13h00-14:00 SATELLITE SYMPOSIUM:

Title: Current unmet need and the future of complement inhibition in PNH

Chair: Peter Hillmen and Antonio Risitano

Speakers: Peter Hillmen and Antonio Risitano

This symposium is organized by Apellis

SESSION VII – TREATMENT OF IMMUNE APLASTIC ANAEMIA (AA)

Chairs: Andrea Bacigalupo and Carlo Dufour

14:05-14:10	Introduction	Andrea Bacigalupo & Carlo Dufour
14:10-14:25	Immunosuppression and TPO agonists	Régis Peffault de Latour (Paris)
14:25-14:40	Transplantation for acquired SAA: are we making progress?	Andrea Bacigalupo (Rome)
14:40-14:55	How to handle moderate aplastic anaemia	Britta Höchsmann (Ulm)

SELECTED BRIEF ORAL COMMUNICATION

14:55-15:05	<i>Blockade of Common Gamma Chain Cytokines with REGN7257, an Interleukin 2 Receptor Gamma (IL2RG) Monoclonal Antibody, Protected Mice from T Cell-Mediated Disease</i>	<i>Audrey Le Floc'h (Tarrytown)</i>
15:05-15:35	<i>Discussion</i>	

SESSION VIII – TREATMENT OF PAROXYSMAL NOCTURNAL HAEMOGLOBINURIA (PNH)

Chairs: Regis Peffault de Latour and Hubert Schrezenmeier

15:35-15:40	Introduction	Regis Peffault de Latour & Hubert Schrezenmeier
15:40-15:55	Classical treatment of PNH	Hubert Schrezenmeier (Ulm)
15:55-16:10	Challenging situations	Peter Hillmen (Leeds)
16:10-16:25	Novel complement inhibitors	Antonio Risitano (Naples)

SELECTED BRIEF ORAL COMMUNICATION

16:25-16:35	<i>Results of the Pegasus Phase 3 Randomized Trial Demonstrating Superiority of the C3 Inhibitor, Pegcetacoplan, Compared to Eculizumab in Patients with Paroxysmal Nocturnal Hemoglobinuria</i>	<i>Regis Peffault de Latour (Paris)</i>
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LIVE Programme
CET Time

16:35-17:05 *Discussion*

17:05-17:15 *Coffee break*

SESSION IX – EMERGING AREAS AND NEW CHALLENGES

Chairs: Marc H. G. P. Raaijmakers and Lucy Godley

17:15-17:20 Introduction

Marc H.G.P. Raaijmakers &
Lucy Godley

17:20-17:35 Genetic editing of haemopoietic stem cells.
Are we getting there?

Matthew Porteus (Stanford)

17:35-17:50 MDS Stem Cell Surveillance, Characterization and
Therapeutic Targeting

Sten Eirik Jacobsen (Stockholm)

17:50-18:05 Genetic predisposition for myeloid malignancies:
clinical management

Lucy Godley (Chicago)

SELECTED BRIEF ORAL COMMUNICATION

18:05-18:15 *Prevalence of Inherited Predisposition Syndromes in Young
Patients with Acute Myeloid Leukemia and Aberrant Karyotypes*

Fabian Beier (Aachen)

18:15-18:45 *Discussion*

18:45-19:15 *Closing Remarks*

MEETING CLOSURE