



# Inborn Errors Working Party Annual Conference

Graz, Austria  
22-24 September 2023

Dear Colleagues,

This year, the annual conference of the Inborn Errors Working Party (IEWP) will be organised in Graz, Austria, on 22-24 September 2023. It will be a great pleasure to welcome you on behalf of EBMT

As always, the meeting will feature comprehensive sessions on stem cell transplantation and gene therapy in inborn errors of immunity, inborn errors of metabolism, hemoglobinopathies, as well as new indications.

This year's special sessions will focus on the pathophysiology of IEI, on hematopoietic defects (telomeropathies, rasopathies, bone marrow failures), the use of serotherapy, and how to make better use of existing registries. A keynote presentation will be given by Elie Haddad (Montreal, CA).

We look forward to welcoming you in Graz.

Bénédicte Neven, IEWP Chair  
Michael Albert, IEWP Secretary  
Markus Seidel, Local Organiser

## Friday, 22 September 2023

**13:00 – 13:45**    **Registration & Welcome Coffee**

**13:45 – 14:00**    **Welcome**

*Chairs: Bénédicte Neven (FR), Markus Seidel (AT)*

**14:00– 15:45**    **Session I - Stem cell therapy in metabolic diseases**

*Chairs: María Ester Bernardo (IT), Rob Wynn(UK)*

14:00 – 14:12	Safety and tolerability of myeloablative busulfan-based conditioning regimens in young children below the age of 18 months affected by LSD: combined experience of two transplant centers	Valeria Calbi (IT)
14:12 – 14:24	Allo-HSCT for LSDs in the NBS era: combined experience in US and Europe	Jaap Jan Boelens (US)
14:24 – 14:34	Update on the phase 1/2 Clinical Trial of Intravenous FBX-101 (AAVrh10.hGALC) Administered after UCBT for the treatment of Krabbe Disease	Maria Escolar (US)
14:34 – 14:44	Libmeldy gene therapy in the real world	Valeria Calbi (IT)
14:44 – 14:54	Update on HSPC-GT for MPSIIIA	Rob Wynn (UK)
14:54 – 15:04	Update on HSPC-GT for MPSIH	Francesca Tucci
15:04- 15:20	Update on HCTGT in GD3 and MPSII	Rob Wynn (UK)
15:20 – 15:45	Discussion, Q&A	

**15:45 – 16:15**    **Coffee Break**

**16:15 – 17:30**    **Session II - Pathophysiology of IEI and its role in cellular therapy**

*Chairs: Romain Levy (FR), Fabian Hauck (DE)*

16:15 – 16:35	Treg-opathies	Kaan Boztug (AT)
16:35 – 16:55	A novel Inborn Error of Pattern-triggered Immunity	Fabian Hauck (DE)
16:55 – 17:15	Dupilumab for primary atopic diseases	Romain Levy (FR)
17:15 – 17:30	Discussion, Q&A	

**17:30 – 18:00**    **IEWP study committee meeting (invitation only)**

**19:30**            **Networking Event**

## Saturday, 23 September 2023

### 09:00 – 10:00 **Session III - Hematopoietic defects (Telomeropathies)**

*Chairs: Carsten Speckmann (DE), Carmem Bonfim (BR)*

9:00 – 9:30	Radiation- and Alkylator-Free BMT for Dyskeratosis Congenita	Suneet Agarval (US)
9:30 – 9:45	HSCT for telomeropathies in Latin America	Carmem Bonfim (BR)
9:55 – 10:00	Discussion, Q&A	

### 10:00 – 10:30 **Coffee Break**

### 10:30 – 12:00 **Session IV - Conditioning/PK**

*Chairs: Arjan Lankester (NL), Despina Moshous (FR)*

10:30 – 10:50	Busulfan exposure in PID patients: what is the sweet spot?	Jaap Jan Boelens (US)
10:50 – 11:00	Fludarabine PK/PD: Model-Based Dosing for Everyone?	Christopher Dvorak (US)
11:00 – 11:10	ARTIC study final results or combined Trep PK data	Arjan Lankester (NL)
11:10 – 11:20	Treo/Bu conditioning and fertility	Arjan Lankester (NL)
11:20 – 11:40	Toxicities of PTCY in IELs	Despina Moshous (FR)
11:40 – 12:00	Discussion, Q&A	

### 12:00 – 12:45 **Keynote Lecture**

The Journey of HSCT in SCID

Elie Haddad (CA)

### 12:45 – 14:00 **Lunch Break**

### 14:00 – 15:15 **Session V - Challenging indications and novel approaches**

*Chairs: Mary Slatter (UK), Tomas Fox (UK)*

14:00 – 14:25	HSCT for JIA/Still	Juliana Silva (UK)
14:25 – 14:35	HSCT for complex CVID: don` t do it!	Claudia Wehr (DE)
14:35 – 14:45	HSCT for complex CVID: do!	Emma Morris (UK)
14:45 – 15:00	Solid organ tx and HSCT	Paul Szabolcs (US)
15:00 – 15:15	Discussion, Q&A	

**15:15 – 16:15 Session VI - HLH and related diseases**

*Chairs: Claire Booth (UK)*

15:15 – 15:30	EBV control	Benjamin Fournier (FR)
15:30 – 15:40	DLI after SCT for pHLH	Svea Böhm (DE)
15:40 – 15:50	Polymorphism or risk allele? PRF1 A91V in trans with a “severe” PRF1 mutation	Oliver Wegehaupt (DE)
15:50 – 16:00	Gene editing for XIAP	Annelotte Mudde (UK)
16:00 – 16:15	Discussion, Q&A	

**16:15 – 16:45 Coffee Break**

**16:45 - 17:10 Session VII – Panel Discussion**

Conceptual approach to gene therapy for HLH – a panel discussion

Stephan Ehl (DE),  
Claire Booth (UK),  
Despina Moshous (FR)

**17:15 – 18:30 Session VIII - IEWP Studies and proposals**

*Chairs: Bénédicte Neven (FR), Michael Albert (DE)*

17:15 – 17:25	HSCT for CTLA-4 Haploinsufficiency	Christos Tsilifis (UK)
17:25 – 17:35	Update on HSCT for STAT1GOF	Catharina Schutz (DE)
17:35 – 17:45	Outcome of second allogeneic transplant in non-SCID IEI	Zohreh Nademi (UK)
17:45 – 17:55	FHLH2/3	Stephan Ehl (DE)
17:55 – 18:05	HSCT for CIQ deficiency	Helena Buso (IT)
18:05 – 18:15	Haplo HSCT for CGD	Bénédicte Neven (FR)
18:15 – 18:25	AGORA update	Claire Booth (UK)
18:25 – 18:30	Discussion, Q&A	

**20.00 Networking Dinner**

## Sunday, 24th of September 2023

### 09:15 – 10:00 **Session IX - How can we use registries in a more effective manner?**

*Chairs: Markus Seidel (AT), Christopher Dvorak (US)*

9:15 – 9:25	Landscape of 'Registry' Options in Europe and the US/Canada	Christopher Dvorak (US) Markus Seidel (AT)
9:25 – 9:40	"Lost in registration"/ The path of a PID patient into the registry maze	Stephan Ehl (DE)
9:40 – 9:55	"Speaking different languages" / How to get registries to talk to each other and share common data elements to enable transatlantic studies of rare diseases	Larisa Broglie (US)
9:55 – 10:00	Discussion, Q&A	

### 10:00 – 10:45 **Session X - Long-term follow-up and late complications**

*Chairs: Reem Elfeky (UK), Emma Morris (UK)*

10:00 – 10:10	US SCID long-term outcome post-HSCT	<i>Christopher Dvorak (US)</i>
10:10 – 10:20	Non-osteopenic bone pathology as a late effect post-HSCT in patients with IEI	Reem Elfeky (UK)
10:20 – 10:30	Long-term follow-up post-gene therapy for patients with IEI	Emma Morris (UK)
10:30 – 10:45	Discussion, Q&A	

### 10:45 – 11:15 **Coffee Break**

### 11:15 – 12:30 **Session XI - "Classic" indications (SCID, non-SCID...)**

*Chairs: Manfred Hönig (DE), Andrew Gennery (UK)*

11:15 – 11:35	Conditioning SCID Infants Diagnosed Early randomized busulfan study	Sung-Yun Pai (US)
11:35 – 11:50	GPOH SCID registry: infection, therapy, outcome	Manfred Hönig (DE)
11:50 – 12:05	RAG1/2 gene therapy (RECOMB) results	Arjan Lankester (NL)
12:05 – 12:15	LV-GT for X-SCID	Sung-Yun Pai (US)
12:15 – 12:30	Discussion, Q&A	

### 12:30 – 12:45 **Final Remarks**

*Michael Albert (DE), Markus Seidel (AT)*

# Thanks to our sponsors!

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