



# Inborn Errors Working Party Annual Conference

Helsinki, Finland  
27-29 September 2024

## WELCOME

Dear Colleagues,

This year, the annual conference of the Inborn Errors Working Party (IEWP) will be organised in Helsinki, Finland, on 27-29 September 2024. 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, as well as new indications, but also the pathophysiological pathways behind these diseases.

This year's special sessions will focus on the pathophysiology of IEI, on inborn errors in the Finnish population, on viral infections, on gene therapy, etc. Two keynote presentations will be given by Dusan Bogunovic and Don Kohn.

We look forward to welcoming you to Helsinki.

**Bénédicte Neven, *IEWP Chair***  
**Michael Albert, *IEWP Secretary***  
**Heljä Lång, *Local Organiser***  
**Samppa Ryhänen, *Local Organiser***

## Friday, 27 September 2024

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

**13:45 – 14:00**      **Welcome**

**Chairs:** *Bénédicte Neven (FR) & Heljä Lång (FI)*

**14:00 – 15:30**      **Session I: Stem cell therapy in metabolic diseases (I)**

**Chairs:** *María Ester Bernardo (IT) & Robert Wynn (UK)*

12 + 3 min Q&A      Update on HSPC-GT for MLD  
10 + 3 min Q&A      Update on HSPC-GT for MPSIH  
10 + 3 min Q&A      Update on HSPC-GT for MPSII  
10 + 3 min Q&A      Update on HSPC-GT for MPSIII  
10 + 3 min Q&A      Platform approach for LSD with skeletal involvement (MPSIVA, MPSIVB, alphaMAN)  
5 + 5 min Q&A      *Case Presentation*

Valeria Calbi (IT)  
María Ester Bernardo (IT)  
Claire Horgan (UK)  
Robert Wynn (UK)  
Alessandro Aiuti (IT)  
Nathaniel Lucas (NZ)

**15:30 – 16:00**      **Coffee Break**

**16:00 – 16:30**      **Session I: Stem cell therapy in metabolic diseases (II)**

**Chairs:** *María Ester Bernardo (IT) & Robert Wynn (UK)*

12 + 3 min Q&A      Retrospective allo-HSCT data on Osteopetrosis  
12 + 3 min Q&A      Lentiviral-based HSPC-GT for Osteopetrosis

Robert Chiesa (UK)  
Francesca Tucci (IT)

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

**Chairs:** *Mikko Seppänen (FI) & Kate Orf (UK)*

20 + 5 min Q&A      New molecular causes of SCID  
15 + 5 min Q&A      STAT1 and STAT3 GOFs- JAKi, HSCT, or both?  
10 + 5 min Q&A      In vitro assays for the assessment of thymic stromal cell defects

Sophie Hambleton (UK)  
Carsten Speckmann (DE)  
Alexandra Kreins (UK)

## Saturday, 28 September 2024

**09:00 – 10:15**      **Session III: Infections (focus on viruses)**

**Chairs:** *Krzysztof Kalwak (PL) & Olaf Neth (SP)*

7 + 3 min Q&A      Letermovir PK, safety and efficacy data in children  
5 + 5 min Q&A      Letermovir EBMT study  
12 + 3 min Q&A      Metagenomic NGS (London/Paris experience)  
12 + 3 min Q&A      HPV infection in SCID and beyond  
12 + 3 min Q&A      Enteric virus infection in the HSCT setting

Krzysztof Kalwak (PL)  
Krzysztof Kalwak (PL)  
Austen Worth (UK)  
Corentin Le Floch (FR)  
Bénédicte Neven (FR)

**10:15 – 10:45**      **Coffee Break**

**10:45 – 11:15**      **Keynote Lecture**

Gene therapy for IEI

Donald Kohn (USA)

## Saturday, 28 September 2024

11:15 – 12:45

### Session IV: Gene therapy

**Chairs:** *Sung-Yun Pai (US) & Claire Booth (UK)*

15 + 5 min Q&A

Lentiviral gene therapy for XLA

7 + 3 min Q&A

Clinical trial of gene edited T cells for CD40L

7 + 3 min Q&A

Gene editing for RAG1-SCID

7 + 3 min Q&A

Gene therapy in RAG1 deficiency

7 + 3 min Q&A

Clinical trial of T cell gene therapy for XLP

7 + 3 min Q&A

Update on clinical trial for p47-CGD

7 + 3 min Q&A

Artemis SCID ongoing trial update

David Rawlings (USA)

Daniele Canarutto (IT)

Anna Villa (IT)

Arjan Lankester (NL)

Claire Booth (UK)

Claire Booth (UK)

Marina Cavazzana (FR)

12:45 – 14:00

### Lunch Break

14:00 – 14:45

### Keynote Lecture

Genetics of Incomplete Penetrance

Dusan Bogunovic (US)

14:45 – 16:00

### Session V: Challenging indications and novel approaches

**Chairs:** *Despina Moshous (FR) & Manfred Hönig (DE)*

10 + 3 min Q&A

The role of somatic variants in IEI

5 + 2 min Q&A

Case Presentation

10 + 3 min Q&A

Immunological reconstitution post-secondary HSCT for SCID:

a box of chocolate (*or: you never know what you're gonna get*)

10 + 3 min Q&A

Shifting Perspective on scReening for Inborn errors

of immunity with Neonatal Genetics (SPRING)

Despina Moshous (FR)

María Trabazo (SP)

Manfred Hönig (DE)

Mirjam van der Burg (NL)

16:00 – 16:30

### Coffee Break

16:30 – 17:30

### Session VI: Characteristic diseases of the Finnish population

**Chairs:** *Samppa Ryhänen (FI) & Heljä Lång (FI)*

12 + 3 min Q&A

Clinical and immunological aspects of CHH

12 + 3 min Q&A

HSCT in CH

15 + 5 min Q&A

Mechanism-based therapeutic interventions for autoimmunity in APECED patients

Svetlana Vakkilainen (FI)

Heljä Lång (FI)

Saila Laakso (FI)

17:30 – 18:35

### Session VII: IEWP Studies and proposals

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

10 + 5 min Q&A

9/10 permissive HLA mismatch

10 + 5 min Q&A

Clinical practice guideline: Long-term follow-up in patients transplanted for IEI

10 + 5 min Q&A

TTC7A

5 + 5 min Q&A

DADA2 proposal

3 + 3 min Q&A

IL2RA and IL2RB proposal

Arjan Lankester (NL)

Reem Elfeky (UK)

Giulia Prunotto (IT)

Etai Adam (IL)

## Sunday, 29 September 2024

09:00 – 10:15

### Session VIII: HSCT for IEI in AYAs (focus on CVID)

**Chair:** Emma Morris (UK) & Klaus Warnatz (DE)

5 + 3 min Q&A  
15 + 5 min Q&A  
15 + 5 min Q&A  
5 + 5 min Q&A  
12 + 3 min Q&A

Case presentation

Challenges with diagnosis of CVID and selection of patients for Allo SCT

GI & Liver disease in CVID and the role of liver transplantation

Case presentation

Recent CVID Allo SCT outcomes and proposed COCOA clinical study

Raffaella Muratori (BR)  
Klaus Warnatz (DE)  
Neil Halliday (UK)  
Muriel Schmutz (FR)  
Emma Morris (UK)

10:15 – 11:15

### Session IX – Haplo HSCT for IEI

**Chair:** Andrew Gennery (UK) & Arjan Lankester (NL)

10 + 10 min Q&A

PTCY or TCR $\alpha\beta$ : takeaways from the IEWP study - a discussion

10 + 5 min Q&A  
7 + 3 min Q&A  
10 + 5 min Q&A

How to best use T-cell directed serotherapy in haplo HSCT

Real-time treosulfan PK

Adoptive cellular therapy with CD45RO+ memory T-cell addback?

Michael Albert (DE)  
Su Han Lum (UK)

Madeleine Powys (AU)  
Su Han Lum (UK)

11:15 – 11:45

### Coffee Break

11:45 – 12:45

### Session X: Bridging strategies to HSCT

**Chairs:** Reem Elfeky (UK) & Carsten Speckmann (DE)

7 + 3 min Q&A  
7 + 3 min Q&A  
7 + 3 min Q&A  
7 + 3 min Q&A

CTLA4 fusion proteins as bridge to HSCT in LRBA deficiency

CTLA4 fusion proteins as bridge to HSCT in CTLA4 deficiency

Leniolisib in APDS1/2 patients under named patient program in Europe

Emapalumab as a bridge to HSCT

Safa Baris (TR)  
Christo Tsilifis (UK)  
Reem Elfeky (UK)  
Pietro Merli (IT)

12:45 – 13:00

### Closing remarks

Bénédicte Neven (FR), Michael Albert (DE) & Heljä Lång (FI)

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