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## Entity Print



Bénédicte Neven

IEWP Chair

France

## Major achievements

2023 has once again given us great opportunities to push back the frontiers of our knowledge in the fields of transplantation and cell therapy for immune deficiencies and non-malignant genetic diseases.

Several important studies conducted within the IEWP have been published, such as the results of allograft transplantation in patients with hypomorphic RAG deficiency (in collaboration with the PIDTC), the PREPAD study (Predicting Patient Death after Allogeneic Stem Cell Transplantation for Inborn Errors Using Machine learning), the multicentric retrospective experience of JAK inhibitor treatment for inborn errors of JAK/STAT signalling (in collaboration with ESID). The retrospective study on outcome of HLA-mismatched HSCT in patients with PID following in vitro T-cell depletion with CD3+TCR $\alpha\beta$ /CD19 depleted PBSC or in vivo T-cell depleted HSCT with post-transplant cyclophosphamide is under revision.

Important studies are on-going (on FHL2 and 3, haplo-transplant in CGD) or about to start (osteopetrosis, long term outcome in Chediak-Higashi syndrome...).

In June 2023, we met in Brescia for the midterm meeting co-organised with the PDWP and the AIWP. We all enjoyed our annual working party that took place in the nice city of Graz. The 140 participants enjoyed an outstanding scientific program.

Education remained also our top priority. The very first international IEWP spring school on HSCT in IEIs took place in April at Frauenchiemsee in Germany. 22 attendees and 7 faculty members met for 4 days to cover the main specificities of cellular therapies in this field. This event was very much appreciated by attendees. The second edition will take place in Montserrat, close to Barcelona in June 2024.

## Principal research studies

[Setting the stage for newborn screening and gene therapy: a disease-specific retrospective survey of FHL2 and 3](#)

[Study type](#)

[Retrospective Studies](#)

[Diseases](#)

[Inherited disorders](#)

[Group](#)

[Inborn Errors Working Party \(IEWP\)](#)

[Type of treatment](#)

[Allogeneic](#)

[Principal investigator](#)

[Stephan Ehl, Despina Moshous, Claire Booth, Sujal Ghosh](#)

[Outcome of haploidentical transplant in chronic granulomatous disease](#)

Study type

Retrospective Studies

Diseases

Inherited disorders

Group

Inborn Errors Working Party (IEWP)

Type of treatment

Allogeneic - Haploidentical

Principal investigator

Benedicte Neven

The outcome of second and third allogeneic conditioned HSCT in patients with non-SCID primary immune deficiency (PID) following primary or secondary graft failure

Study type

Retrospective Studies

Diseases

-

Group

Inborn Errors Working Party (IEWP)

Type of treatment

Allogeneic

Principal investigator

Zohreh Nademi

Late effects after hematopoietic stem cell transplantation in patients with HLH

Study type

Retrospective Studies

Diseases

Other non-malignant disorders

Group

Paediatric Diseases Working Party (PDWP)

Inborn Errors Working Party (IEWP)

Transplant Complications Working Party (TCWP)

Type of treatment

-

Principal investigator

AnnaCarin Horne

## **Key publications**

2023

Predicting Patient Death after Allogeneic Stem Cell Transplantation for Inborn Errors Using Machine Learning (PREPAD): A European Society for Blood and Marrow Transplantation Inborn Errors Working Party Study

Group

Inborn Errors Working Party (IEWP)

1st listed author

Erik G J von Asmuth

Journal

Transplant Cell Ther.

2023

Hypomorphic RAG deficiency: impact of disease burden on survival and thymic recovery argues for early diagnosis and HSCT

Group

Inborn Errors Working Party (IEWP)

1st listed author

C Schuetz

Journal

Blood.

2023

JAK inhibitor treatment for inborn errors of JAK/STAT signaling: An ESID/EBMT-IEWP retrospective study

Group

Inborn Errors Working Party (IEWP)

Other society

1st listed author

Marco Fischer

Journal

J Allergy Clin Immunol.

2023

Busulfan-fludarabine- or treosulfan-fludarabine-based conditioning before allogeneic HSCT from matched sibling donors in paediatric patients with sickle cell disease: A study on behalf of the EBMT Paediatric Diseases and Inborn Errors Working Parties

Group

Inborn Errors Working Party (IEWP)

Paediatric Diseases Working Party (PDWP)

1st listed author

Annamária Cseh

Journal

Br J Haematol.

2023

Outcome of haematopoietic cell transplantation in children with lysosomal acid lipase deficiency: a study on behalf of the EBMT Inborn Errors Working Party Group

Inborn Errors Working Party (IEWP)

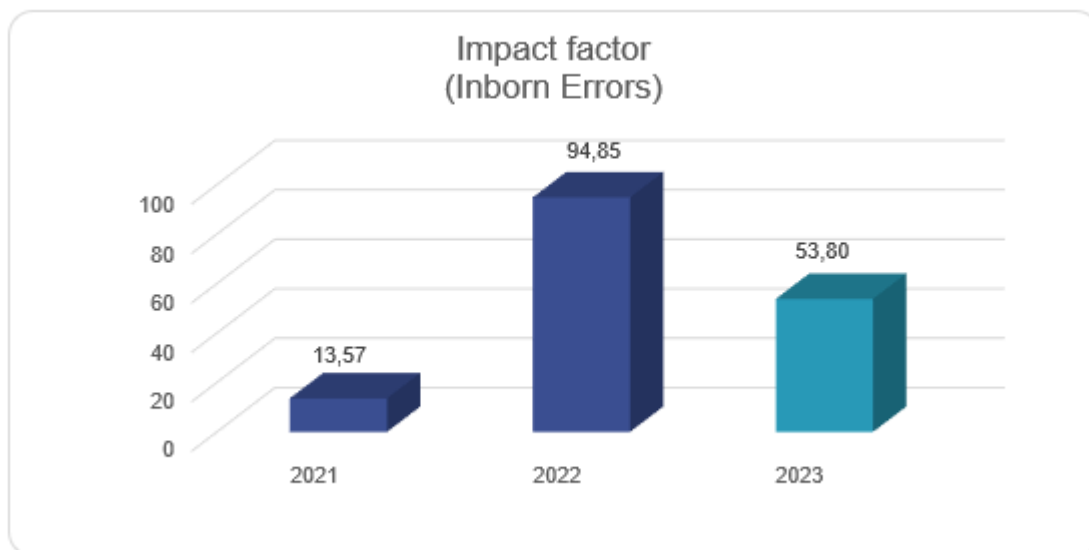
1st listed author

Su Han Lum

Journal

Bone Marrow Transplant.

See the full list of the IEWP 2023 publications



**2021 2022 2023**

<b>Oral presentations</b>	3	7	<b>4</b>
<b>Poster presentations</b>	1	0	<b>1</b>
<b>Educational events</b>	3	3	<b>4</b>



[Event](#)

## **49th Annual Meeting of the EBMT**

[Apr 23, 2023 - Apr 26, 2023 / Paris, France](#)

[Discover more](#)



[Event](#)

**[Inborn Errors Working Party Spring School on Cellular Therapy  
for Inborn Errors of Immunity and Hematopoiesis 2023](#)**

[Apr 02, 2023 - Apr 05, 2023 / Frauenchiemsee, Germany](#)

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