



## Breadcrumb



# Entity Print



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## Major achievements

2025 has been the first year for Claire Booth and myself as Co-Chair and Chair, respectively, of the IEWP of the EBMT. It has been characterised by key studies, collaborative achievements and educational events in the field of HCT and gene therapy for Inborn Errors of Immunity and Metabolism.

Several key studies have been published. The retrospective study on **haploidentical HSCT in chronic granulomatous disease** (*Blood*) has provided valuable insights into transplantation strategies in this complex Inborn Error of Immunity (IEI). Another notable retrospective study focused on **HCT outcomes in purine nucleoside phosphorylase deficiency** (*Blood*) covering both immunological and neurological aspects of the disease and highlighting how early diagnosis is critical to improve outcomes.

Other studies investigated HCT outcomes for **STAT1 gain-of-function disease following JAK inhibition bridging** (*J Hum Immun*) and **late effects after HCT in patients with HLH,** the latter in collaboration with the Histiocyte Society, PDWP, and TCWP (*JACI*). Another relevant publication focused on **clinical phenotypes and treatment modalities in 32 JAGN1-deficient patients**, in collaboration with ESID (*Blood Advances*).

Moreover, pivotal studies are ongoing that will impact the landscape of HCT for inborn errors, including research on patients with **interferon gamma receptor 1/ 2 deficiencies, cartilage hair hypoplasia, FHL2 and FHL3**, and **long-term outcomes in Chediak-Higashi syndrome**.

In terms of educational events, the **25$^{th}$ IEWP Annual Conference** was held in Freiburg, Germany in September 2025, bringing together over 180 attendees, including 12 nurses, to share knowledge and advance the field on HCT and cellular therapies, including ex-vivo gene therapies, in these disorders. Moreover, the **third edition of IEWP Spring School** on HCT in Inborn Errors of Immunity, took place extremely successfully in Seville, Spain in April 2025 and gave the chance to over 30 international experts and junior attendees to deepen knowledge and share expertise on cellular therapies in these disorders.  Indeed, many of the attendees from previous Spring Schools now sit on the IEWP Juniors Committee, helping to shape the Annual Meeting and support the community.

The IEWP continues to advance the frontiers of scientific research and clinical practice by promoting collaboration between Europe and the United States, with the goal of enhancing patient outcomes and optimising transplant protocols.

# Principal research studies

HSCT experience in patients with Interferon gamma receptor deficiencies
Study type

Retrospective Studies
Diseases
Inherited disorders
Group
Inborn Errors Working Party (IEWP)
Type of treatment
Allogeneic
Principal investigator
Peter Olbrich
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
Allogeneic hematopoietic stem cell transplantation in cartillage hair hypoplasia (CHH); a joint EBMT-CIBMTR study
Study type
Retrospective Studies
Diseases
Inherited disorders
Group
Inborn Errors Working Party (IEWP)
Type of treatment
Allogeneic
Principal investigator
Heljä Lång
Outcome of haematopoietic stem cell transplant (HSCT) in patients with Chediak Higashi syndrome
Study type
Retrospective Studies
Diseases

Inherited disorders
Group
Inborn Errors Working Party (IEWP)
Type of treatment
Allogeneic
Principal investigator
Reem Elfeky
A prospective outcome study on patients with profound combined immune deficiency (p-CID)
Study type
Retrospective Studies
Diseases

Group
Inborn Errors Working Party (IEWP)
Type of treatment

Principal investigator
Stephan Ehl

## Key publications

2025
Improved outcome of HSCT in STAT1 gain-of-function disease following JAK inhibition bridging
Group
Inborn Errors Working Party (IEWP)
Other society
1st listed author
Emilie Pauline Buddingh
Journal
J Hum Immun.
2025
Late effects after hematopoietic stem cell transplantation in patients with HLH: A Histiocyte Society, PDWP, IEWP, and TCWP EBMT Study
Group
Inborn Errors Working Party (IEWP)
Complications Working Party (CWP)

Paediatric Diseases Working Party (PDWP)
1st listed author
Kim Ramme
Journal
J Allergy Clin Immunol.
2025
Hematopoietic stem cell transplantation for purine nucleoside phosphorylase deficiency: an EBMT-IEWP retrospective study
Group
Inborn Errors Working Party (IEWP)
1st listed author
Uli S Herrmann
Journal
Blood.
2025
HLA-haploidentical stem cell transplantation for chronic granulomatous disease: an EBMT-IEWP retrospective study
Group
Inborn Errors Working Party (IEWP)
1st listed author
Quentin Riller
Journal
Blood.
2025
Extended clinical phenotypes and treatment modalities in 32 JAGN1-deficient patients: a multicenter study by ESID and EBMT IEWP
Group
Inborn Errors Working Party (IEWP)
Other society
1st listed author
Julia Fekadu-Siebald
Journal
Blood Adv.
See the full list of the IEWP 2025 publications



# Indicators

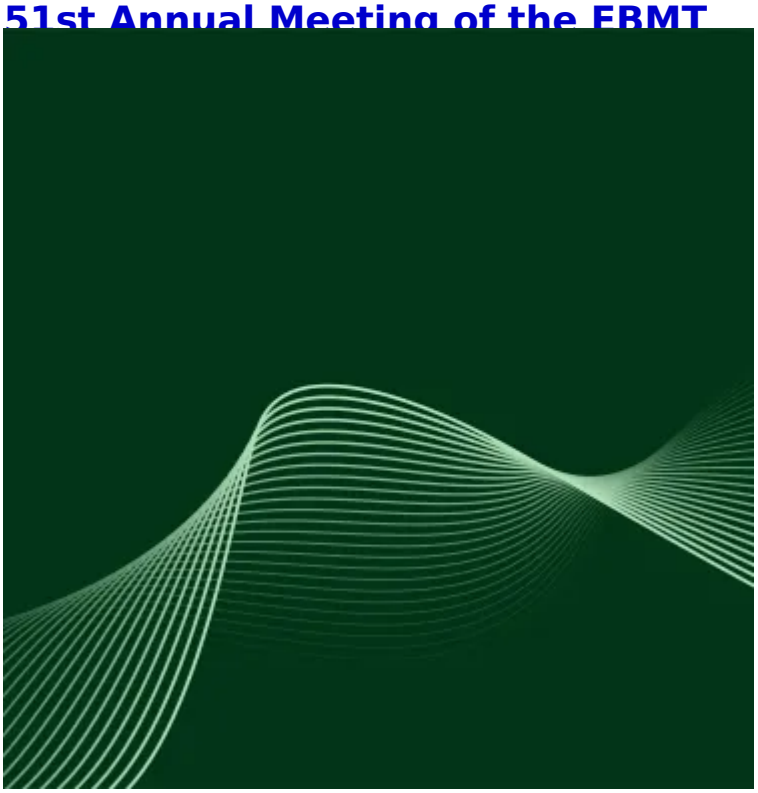
| | 2023 | 2024 | 2025 |
|---|---|---|---|
| **Impact factor** | 53.80 | 62.43 | **64.80** |
| **Oral presentations** | 4 | 4 | **2** |
| **Poster presentations** | 1 | 3 | **1** |
| **Educational events** | 4 | 1 | **1** |

# Major educational events



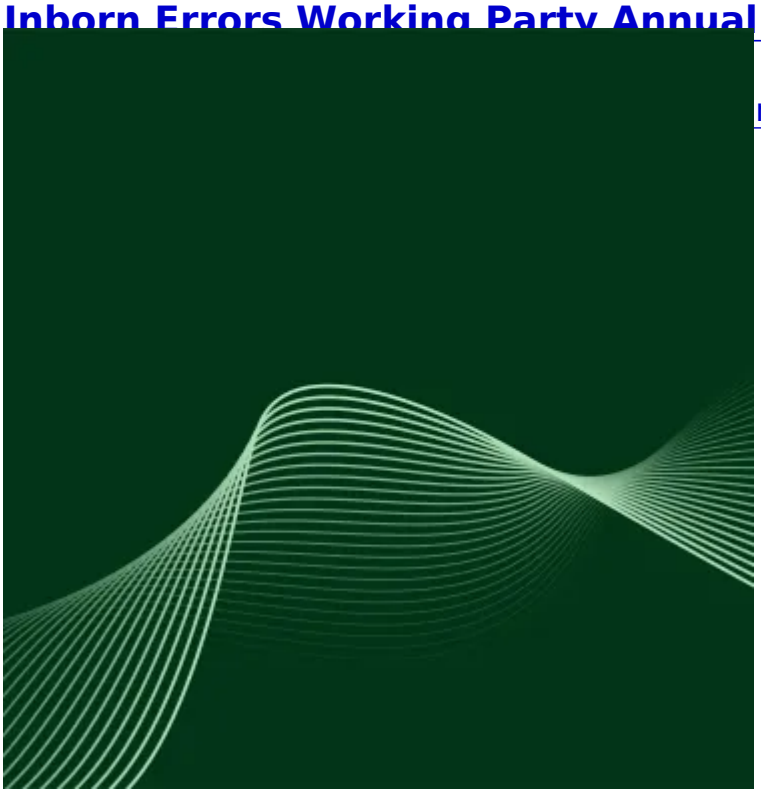
Event

## 51st Annual Meeting of the EBMT



Event

## Inborn Errors Working Party Annual Conference



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Event

## Inborn Errors Working Party Spring School

Apr 27, 2025 - Apr 30, 2025 / Gerena, Seville, Spain
Discover more
VISIT THE IEWP WEBPAGE