



Inborn Errors Working Party

Bénédicte Neven (chair)

Michael Albert (secretary/ESID chair)

Paris, France

Munich, Germany

STUDIES

1. Outcome of HSCT in patients with Chediak Higashi syndrome
2. Allogeneic HSCT in infantile Osteopetrosis
3. HSCT for Cartilage Hair Hypoplasia: Joint study with CIBMTR
4. HSCT for DADA2: Joint study with CIBMTR
5. The influence of pre-transplant immune dysregulation on transplant outcomes for CID and immune dysregulatory disorders.
6. Setting the stage for newborn screening and gene therapy of FHL2 and FHL3
7. NFKBIA EDA-ID outcome and management
8. HSCT for MSDM due to defects of IL12- and IL23- signalling

If you wish to participate in data collection for these studies, please contact us at iewp@ebmt.org

RECENT PUBLICATION ON BEHALF OF IEWP

Hematopoietic Stem Cell Transplantation for C1q Deficiency: A Study on Behalf of the EBMT Inborn Errors Working Party

Helena Buso, Etai Adam, Peter D Arkwright, Sagar Bhattad, Amir Ali Hamidieh, Maryam Behfar, Alexandre Belot, Sarah Benezech, Alice Y Chan, Yanick J Crow, Christopher C Dvorak, Aisling M Flinn, Urvi Kapoor, Arjan Lankester, Masao Kobayashi, Risa Matsumura, Hadi Mottaghishah, Satoshi Okada, Marie Ouachee, Nima Parvaneh, Stalin Ramprakash, Prakash Satwani, Samin Sharafian, Clément Triaille, Robert F Wynn, Nasim Movahedi, Vahid Ziaee, Eleri Williams, Mary Slatter, Andrew R Gennery

Hematopoietic stem cell transplantation for CTLA-4 insufficiency across Europe: A European Society for Blood and Marrow Transplantation Inborn Errors Working Party study

Christo Tsilifis, Carsten Speckmann, Su Han Lum, Thomas A Fox, Adriana Margarit Soler, Yasmina Mozo, Dolores Corral, Anna-Maria Ewins, Rosie Hague, Christina Oikonomopoulou, Krzysztof Kalwak, Katarzyna Drabko, Robert Wynn, Emma C Morris, Suzanne Elcombe, Venetia Bigley, Vassilios Lougaris, Michele Malagola, Fabian Hauck, Petr Sedlacek, Alexandra Laberko, Jennifer M L Tjon, Emilie P Buddingh, Claudia Wehr, Bodo Grimbacher, Andrew R Gennery, Arjan C Lankester, Michael H Albert, Bénédicte Neven, Mary A Slatter

Allogeneic Hematopoietic Stem Cell Transplantation in Immunodeficiency-Centromeric Instability-Facial Dysmorphism (ICF) Syndrome: an EBMT/ESID Inborn Errors Working Party Study

Dagmar Berghuis, Lubna S Mehyar, Rolla Abu-Arja, Michael H Albert, Jessie L Barnum, Horst von Bernuth, Reem Elfeky, Philippe Lewalle, Alexandra Laberko, Sujal Ghosh, Mary A Slatter, Corry M R Weemaes, Akif Yesilipek, Tiarlan Sirait, Bénédicte Neven, Andrew R Gennery, Arjan C Lankester

Outcomes of HLA-mismatched HSCT with TCR $\alpha\beta$ /CD19 depletion or post-HSCT cyclophosphamide for inborn errors of immunity

Su Han Lum, Michael H Albert, Patrick Gilbert, Tiarlan Sirait, Mattia Algeri, Rafaella Muratori, Benjamin Fournier, Alexandra Laberko, Musa Karakukcu, Elrem Unal, Mouhab Ayas, Satya Prakash Yadav, Tunc Fisgin, Reem Elfeky, Juliana Fernandes, Maura Faraci, Theresa Cole, Ansgar Schulz, Roland Meisel, Marco Zecca, Marianne Ifversen, Alessandra Biffi, Jean-Sebastien Diana, Tanja Vallée, Stefano Giardino, Gizem Zengin Ersoy, Despina Moshous, Andrew R Gennery, Dmitry Balashov, Carmem Bonfim, Franco Locatelli, Arjan Lankester, Bénédicte Neven, Mary Slatter

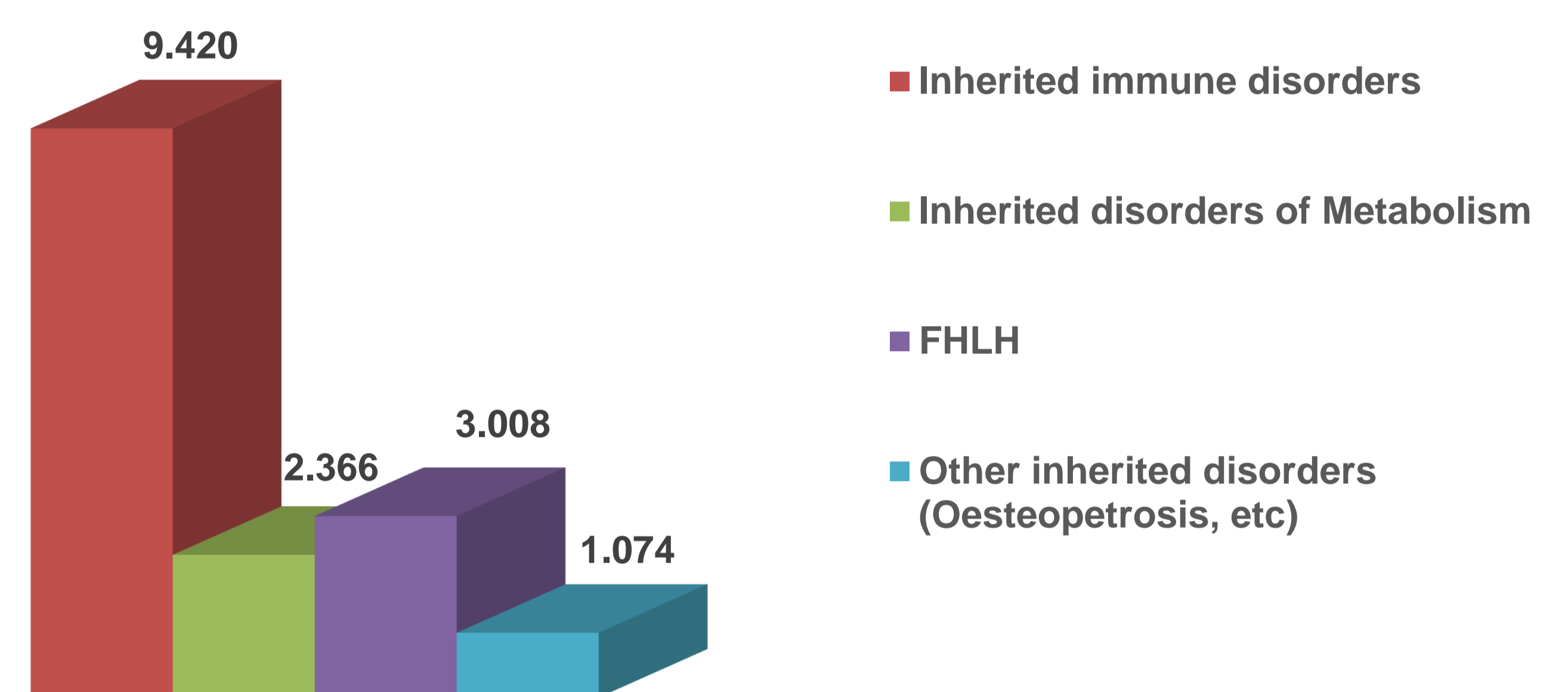
JAK inhibitor treatment for inborn errors of JAK/STAT signaling: An ESID/EBMT-IEWP retrospective study

Marco Fischer, Peter Olbrich, Jérôme Hadjadj, Volker Aumann, Shahrzad Bakhtiar, Vincent Barlogis, Philipp von Bismarck, Markéta Bloomfield, Claire Booth, Emmeline P Buddingh, Deniz Cagdas, Martin Castelle, Alice Y Chan, Shanmuganathan Chandrakasan, Kritika Chetty, Pierre Cougoul, Etienne Crickx, Jasmeen Dara, Angela Deyà-Martinez, Susan Farmand, Renata Formankova, Andrew R Gennery, Luis Ignacio Gonzalez-Granado, David Hagin, Leif Gunnar Hanitsch, Jana Hanzliková, Fabian Hauck, José Ivorra-Cortés, Kai Kisand, Ayca Kiykim, Julia Körholz, Timothy Ronan Leahy, Joris van Montfrans, Zohreh Nademi, Brigitte Nelken, Suhag Parikh, Silvi Plado, Jan Ramakers, Antje Redlich, Frédéric Rieux-Laucat, Jacques G Rivière, Yulia Rodina, Pérsio Roxo Júnior, Sarah Salou, Catharina Schuetz, Anna Shcherbina, Mary A Slatter, Fabien Touzot, Ekrem Unal, Arjan C Lankester, Siobhan Burns, Mikko R J Seppänen, Olaf Neth, Michael H Albert, Stephan Ehl, Bénédicte Neven, Carsten Speckmann

Extended clinical phenotypes and treatment modalities in 32 JAGN1-deficient patients. A multicenter study by EBMT IEWP

Julia Fekadu-Siebold, Emilia Salzmänn-Manrique, Jan Robert Heusel, Andre Willasch, Fabian Hauck, Luis Ignacio Gonzalez-Granado, Zahra Chavoshzadeh, Samin Sharafian, Franziska Cuntz, Safa Baris, Andrea Finocchi, Mattia Algeri, Roya Sherkat, Maja Kludel-Dreszler, Cornelia Zeidler, Christine Bellanné-Chantelot, Gerhard Kindle, Blandine Beaupain, Catherine Paillard, Markus G Seidel, Peter Bader, Michael H Albert, Bénédicte Neven, Jean Donadieu, Shahrzad Bakhtiar

NUMBER OF PATIENTS REGISTERED AT EBMT REGISTRY



IEWP SESSIONS AT EBMT ANNUAL MEETING 2025

Tuesday, 1 April 2025

- 11:00 - 12:15 Inborn Errors Working Party Session**
Room Nazioni Chair: Bénédicte Neven (France)
- **Diagnosis and treatment of SCID in the era of newborn screening**
Manfred Hoening (Ulm, Germany)
 - **Personalised serotherapy: are we there?**
Arjan Lankester (Leiden, the Netherlands)
 - **To transplant or not to transplant – A prospective observational study on combined immunodeficiencies**
Stephan Ehl (Freiburg, Germany)
 - **Discussion**
- 12:45 - 14:00 Inborn Errors Working Party Business Meeting**
Room Dante (S-1) Chair: Bénédicte Neven (France)
- 16:30 - 17:45 Inborn Errors Oral Session**
Room Nazioni Chair: Bénédicte Neven (France)

FUTURE EVENTS

- **IEWP Spring School: 27-30 April 2025 in Seville, Spain**
- **IEWP Annual conference: 26-28 September 2025 in Freiburg, Germany**

IEWP TEAM

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Michael Albert	Secretary	michael.albert@med.uni-muenchen.de
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Jeroen Knippenberg	Data manager	jeroen.knippenberg@ebmt.org
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