



Annual Review of the Inborn Errors Working Party

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ONGOING STUDIES

1. Allogeneic HSCT in infantile osteopetrosis*
2. Setting the stage for newborn screening and gene therapy of FHL2 and FHL3*
3. Outcome of HSCT in patients with Chédiak-Higashi Syndrome*
4. HSCT for cartilage hair hypoplasia*
5. Outcome of haploidentical HSCT in CGD patients
6. Second and third allo-HSCT in patients with (non-SCID) PID
7. TCR $\alpha\beta$ or PTCY for HLA-mismatched HSCT in PID
8. Graft failure and immune cytopenia in cord blood transplantation (CBT) for inherited metabolic diseases (IMD)
9. HSCT in patients with IFNGR1/2 deficiency

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

RECENT PUBLICATIONS

JAK inhibitor treatment for inborn errors of JAK/STAT signaling: An ESID/EBMT-IEWP retrospective study. *J Allergy Clin Immunol*, 2024

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-Martínez, 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 Slatte, Fabien Touzot, Ekrem Unal, Arjan C Lankester, Siobhan Burns, Mikko R J Seppänen, Olaf Neth, Michael H Albert, Stephan Ehl, Bénédicte Neven, Carsten Speckmann.

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. *Br J Haematol*, 2024

Annamária Cseh, Jacques-Emmanuel Galimard, Josu de la Fuente, Antonella Isgro, Marco Zecca, Birgit Garwer, Alexandra Biffi, Mahmoud Aljurf, Mikael Sundin, Cristina Belendez, Franco Locatelli, Adriana Balduzzi, Sarah Lawson, Henrik Sengeloev, Marianne Ifversen, Riccardo Saccardi, Robert Wynn, Arjan C Lankester, Selim Corbacioglu, Christina Peters.

Hypomorphic RAG deficiency: impact of disease burden on survival and thymic recovery argues for early diagnosis and HSCT. *Blood*, 2023

C. Schuetz, J Gerke, M Ege, J Walter, M Kusters, A Worth, J A Kanakry, D Dimitrova, B Wolska-Kuśnierz, K Chen, E Unal, M Karakucuk, O Pashchenko, J Leiding, T Kawai, P J Amrolia, D Berghuis, J Buechner, D Buchbinder, M J Cowan, A R Gennery, T Gungör, J Heimall, M Miano, I Meyts, E C Morris, J Rivière, S O Sharapova, P J Shaw, M Slatter, M Honig, P Veys, A Fischer, M Cavazzana, D Moshous, A Schulz, M H Albert, J M Puck, A C Lankester, L D Notarangelo, B Neven.

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. *Transplant Cell Ther*, 2023

Erik G J von Asmuth, Bénédicte Neven, Michael H Albert, Alexander B Mohseny, Marco W Schilham, Harald Binder, Hein Putter, Arjan C Lankester.

Decision making on HSCT in patients with hemoglobinopathies; an EBMT Pediatric Diseases Working Party and Inborn Errors Working Party scenario-based survey on physicians' perspectives. *Bone Marrow Transplant.*, 2023

Hilda Mekelenkamp, Martine de Vries, Benny Markovitch, Tiarlan Sirait, Arwen H Pieterse, Joëll Bense, Katharina Kleinschmidt, Michael H Albert, Bénédicte Neven, Selim Corbacioglu, Arjan Lankester, Frans Smiers.

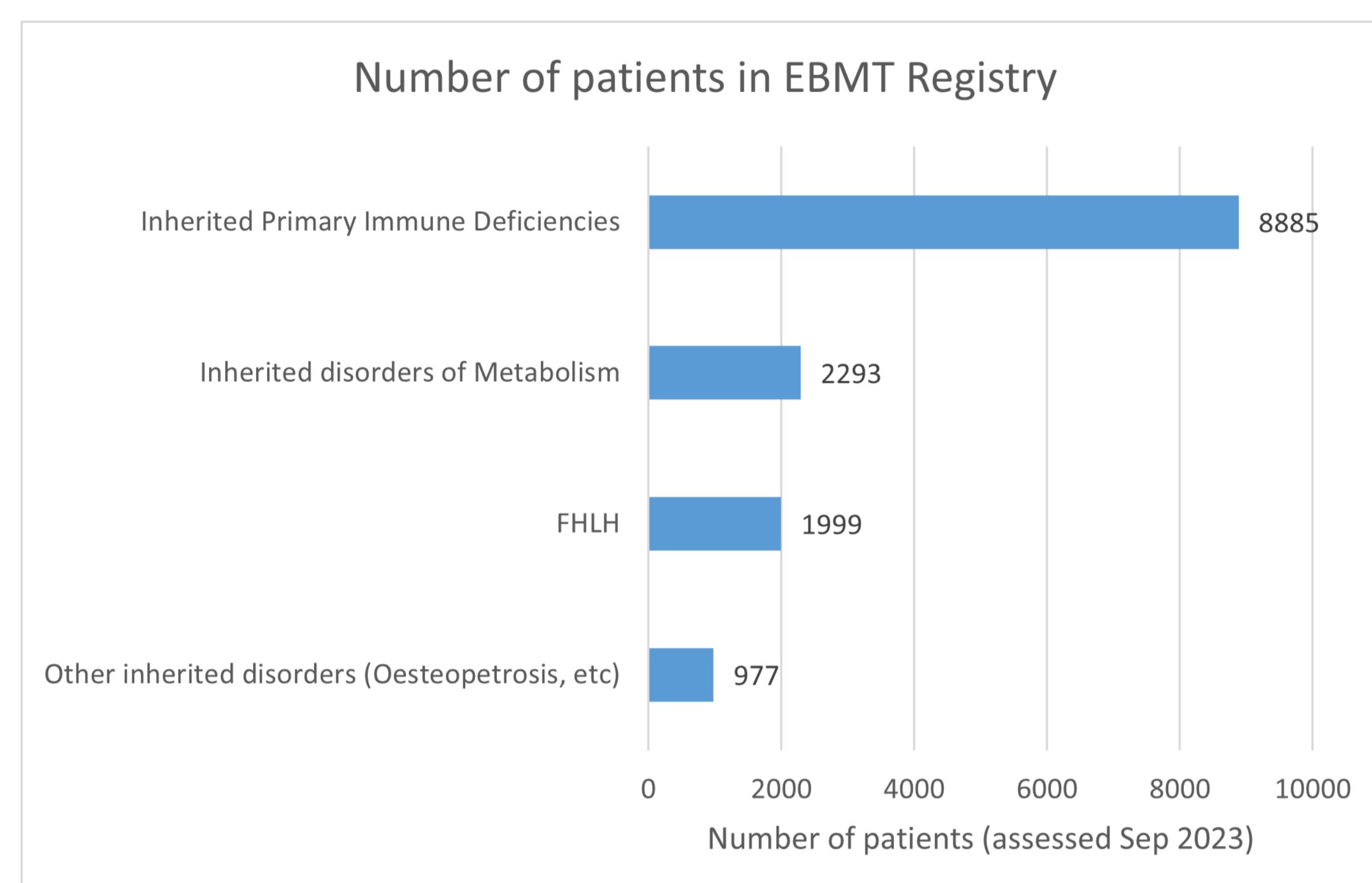
Outcome of haematopoietic cell transplantation in children with lysosomal acid lipase deficiency: a study on behalf of the EBMT Inborn Errors Working Party. *Bone Marrow Transplant.*, 2023

Su Han Lum, Milen Minkov, Simon A Jones, Sheree Hazelaar, Tiarlan Sirait, Jane E Potter, Polina Stepensky, Frederic Garban, Herbert Pichler, Jerry Stein, Zuhre Kaya, Ansgar Schulz, Karin Mellgren, Cristina Diaz de Heredia, Cecile Pochon, Susana Riesco, Miguel Angel Diaz, Gérard Michel, Caroline Lindemans, Bernd Gruhn, Michael H Albert, Arjan C Lankester, Bénédicte Neven, Robert Wynn.

Use of eculizumab in children with allogeneic haematopoietic stem cell transplantation associated thrombotic microangiopathy - a multicentre retrospective PDWP and IEWP EBMT study. *Bone Marrow Transplant.*, 2023

Peter Svec, Reem Elfeky, Jacques-Emmanuel Galimard, Christine S Higham, Arnaud Dalissier, Troy C Quigg, David Bueno Sanchez, Su Han Lum, Maura Faraci, Theresa Cole, Herbert Pichler, Maria Isabel Benítez-Carabante, Julia Horakova, Marta Gonzalez-Vicent, Asaf Yanir, Franca Fagioli, Matthias Wölfl, Nicolas von der Weid, Rachel Protheroe, Gergely Krivan, Carsten Speckmann, Beki James, Simona Lucija Avcin, Yves Bertrand, Marta Verna, Petr Riha, Katharine Patrick, Simone Cesaro, Krzysztof Kalwak, Marc Bierings, Jochen Büchner, Karin Mellgren, Zoltán Prohászka, Bénédicte Neven, Arjan Lankester, Selim Corbacioglu

PATIENTS RELEVANT FOR IEWP



IEWP SESSIONS AT ANNUAL MEETING 2024

TUESDAY, APRIL 16 (LOMOND)

Inborn Errors Working Party Session 11:00 AM – 12:15 PM

Chair: Bénédicte Neven

Paed2-1 Retroviral and lentiviral vectors based Gene therapy in PIDs; were do we stand?

Sung-Yun Pai (United States)

Paed2-2 New strategies of gene therapy in PIDs

Thomas Fox (United Kingdom)

Paed2-3 Gene therapy in metabolic diseases

Rob Wynn (United Kingdom)

Paed2-4 Discussion

Inborn Errors Oral Session 04:30 PM – 05:45 PM

Chairs: Bénédicte Neven (France), Michael Albert (Germany)

IEWP Business Meeting of the Inborn Errors Working Party

12:45 PM – 02:00 PM

Chair: Bénédicte Neven (France)

FUTURE EVENTS

Inborn Errors Working Party Spring School 2024

23-26 June 2024

Montserrat | Barcelona, Spain

Inborn Errors Working Party Annual Conference

27-29 September

Helsinki, Finland

visit ebmt.org/events for more info

THE IEWP TEAM

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