

Curriculum Vitae

Personal Data

Title	Prof. Dr. med
First name	Stephan
Name	Ehl
Current position	Full Professor (W3) for Pediatric Immunology, Director, Institute for Immunodeficiency Medical Director, Center for Chronic Immunodeficiency permanent position
Current institution(s)/site(s), country	Center for Chronic Immunodeficiency (CCI), Institute for Immunodeficiency & Center for Pediatrics and Adolescent Medicine Medical Center – University of Freiburg Breisacher Str. 115 D-79106 Freiburg
Identifiers/ORCID	0000-0002-9265-2721

Qualifications and Career

Stages	Periods and Details
Degree programme	Study of Medicine in Aachen, Erlangen and Munich 1985-1992
Doctorate	1992 Prof. Dr. D. Seiler Friedrich-Alexander Universität Erlangen-Nürnberg
Stages of academic/professional career	since 2018 Director, Institute for Immunodeficiency, Medical Center - University of Freiburg since 2012 Medical Director, Center for Chronic Immunodeficiency, Freiburg 2008 - 2011 Scientific Director, Center for Chronic Immunodeficiency, Freiburg since 2002 Attending physician, Center for Pediatrics, Freiburg 1998 - 2002 Assistant physician, Center for Pediatrics, Freiburg 1994 - 1997 Postdoctorate, Institute for Experimental Immunology, University of Zurich, Switzerland 1993 - 1994 Intern in Pediatrics, Children's Hospital Ulm

Activities in the Research System

2024	Initiation of a DFG funded partnership between Faculty of Medicine and Institute IMAGINE, Paris
since 2019	Co-speaker Clinician Scientist Programs IMMEDIATE and IMPACT
since 2015	Speaker CRC 1160 "Immunopathology as a consequence of impaired immune reactions"
2021-2025	President, German working group for Pediatric Immunology (API)
2015-2018	Coordinator EU-Consortium "Euro-CID"
2012-2014	Board member and head of HLH working group, Histiocyte Society
2010-2014	Board member and head of ESID registry, European Society for Immunodeficiencies

2008-2018 Co-Founder and Speaker of the BMBF-funded Integrated Research and Treatment Center (IFB) Center for Chronic Immunodeficiency

Supervision of Researchers in Early Career Phases

I have supervised 3 Bachelor, 4 Master and 2 MD student dissertations within the last 5 years. I have hosted 3 postdoctoral researchers and 3 clinician scientists within the last 5 years. I am mentor in various (Advanced) Clinician Scientist programs and member of the CIBSS Launchpad programme

Scientific Results

Category A.

1. Beyond genotype: challenges in predicting disease risk for carriers of biallelic perforin variants. Wegehaupt O, Borisov O, Sieni E, Oyen F, Mann J, Coniglio ML, Chinnici A, Pegoraro F, Beneforti L, Gilmour K, Moshous D, de Saint Basile G, Zhang W, Marsh R, De Fusco C, Wustrau K, Timeus F, Micalizzi C, Gunsilius E, Hosking L, Choo S, Ghosh S, Köttgen A, Lehmberg K, Ehl S (2025). **Blood** 145. doi: 10.1182/blood.2024027954.
2. Clinical manifestations, disease penetrance, and treatment in individuals with SOCS1 insufficiency: a registry-based and population-based study. Hadjadj J, Wolfers A, Borisov O, et int., Ehl S (2025). **Lancet Rheumatology** Feb 27. doi: 10.1016/S2665-9913(24)00348-5.
3. Diagnostic evaluation of pediatric autoimmune-lymphoproliferative immunodeficiencies (ALPID): A prospective cohort study. Hägele P, Staus P, Scheible R, et int., Rensing-Ehl A, Ehl S (2024). **Lancet Hematology** 11. doi: 10.1016/S2352-3026(23)00362-9.
4. Abnormal biomarkers predict complex FAS or FADD defects missed by exome sequencing. Rensing-Ehl A, Lorenz MR, Führer M, Willenbacher W, Willenbacher E, Sopper S, Abinun M, Maccari ME, König C, Haegele P, Fuchs S, Castro C, Kury P, Pelle O, Klemann C, Heeg M, Thalhammer J, Wegehaupt O, Fischer M, Goldacker S, Schulte B, Biskup S, Chatelain P, Schuster V, Warnatz K, Grimbacher B, Meinhardt A, Holzinger D, Oommen PT, Hinze T, Hebart H, Seeger K, Lehmberg K, Leahy TR, Claviez A, Vieth S, Schilling FH, Fuchs I, Groß M, Rieux-Laucat F, Magerus A, Speckmann C, Schwarz K, Ehl S (2024). **J Allergy Clin Immunol** 153. doi: 10.1016/j.jaci.2023.11.006.
5. JAK inhibitor treatment for inborn errors of JAK/STAT signaling: An ESID/EBMT-IEWP retrospective study (2024). Fischer M, Olbrich P, Hadjadj J, et int., Ehl S, Neven B, Speckmann C (2023). **J Allergy Clin Immunol** 153. doi: 10.1016/j.jaci.2023.10.018.
6. Activated phosphoinositide 3-kinase δ syndrome: Update from the ESID Registry and comparison with other autoimmune-lymphoproliferative inborn errors of immunity. Maccari ME, Wolkewitz M, Schwab C, et int., Ehl S (2023). **J Allergy Clin Immunol** 152. doi: 10.1016/j.jaci.2023.06.015.
7. STAT3-confusion-of-function: Beyond the loss and gain dualism. Lodi L, Faletti LE, Maccari ME, Consonni F, Groß M, Pagnini I, Ricci S, Heeg M, Simonini G, Azzari C, Ehl S (2022). **J Allergy Clin Immunol**. 150. doi: 10.1016/j.jaci.2022.06.007.
8. Early-onset autoimmunity associated with SOCS1 haploinsufficiency. Hadjadj J, Castro CN, Tusseau M, Stolzenberg MC, Mazerolles F, Aladjidi N, Armstrong M, Ashrafian H, Cutcutache I, Ebetsberger-Dachs G, Elliott KS, Durieu I, Fabien N, Fusaro M, Heeg M, Schmitt Y, Bras M, Knight JC, Lega JC, Lesca G, Mathieu AL, Moreews M, Moreira B, Nosbaum A, Page M, Picard C, Ronan Leahy T, Rouvet I, Ryan E, Sanlaville D, Schwarz K, Skelton A, Viillard JF, Viel S, Villard M, Callebaut I, Picard C, Walzer T, Ehl S, Fischer A, Neven B, Belot A, Rieux-Laucat F (2020) **Nat Commun** 11. doi: 10.1038/s41467-020-18925-4.
9. A distinct CD38+CD45RA+ population of CD4+, CD8+, and double-negative T cells is controlled by FAS. Maccari ME, Fuchs S, Kury P, et int., Ehl S, Rensing-Ehl A (2021). **J Exp Med** 218. doi: 10.1084/jem.20192191.
10. Distinct molecular response patterns of activating STAT3 mutations associate with penetrance of lymphoproliferation and autoimmunity. Jäggle S, Heeg M, Grün S, Rensing-Ehl A, Maccari ME, Klemann C, Jones N, Lehmberg K, Bettoni C, Warnatz K, Grimbacher B, Biebl A, Schauer U, Hague R, Neth O, Mauracher A, Pachlopnik Schmid J, Fabre A, Kostyuchenko L, Führer M, Lorenz MR, Schwarz K, Rohr J, Ehl S (2020). **Clin Immunol**. 210. doi: 10.1016/j.clim.2019.108316.

These publications specifically refer to our work on JAK/STAT and FAS signalling relevant to CIBSS. Two important publications on STAT signalling are pending resubmission after revision (both J Exp Med).

Academic Distinctions

2020	Elected member, German National Academy of Sciences (Leopoldina)
2001	Kind-Phillip-Prize of the German Society for Pediatric Hematology and Oncology
1999	Research Prize of the German Society for Pediatric Infectious Diseases
1998	Research Prize of the German-Austrian Society for Neonatology and Pediatric Intensive Care