ARBEITSGEMEINSCHAFT
PÄDIATRISCHE IMMUNOLOGIE

Universitätsklinikum
Carl Gustav Carus
DIE DRESDNER.

41th API Annual Meeting

May 22nd – 24th 2025
Kloster Nimbschen, Grimma

Meeting Chairs

Prof. Dr. med. Catharina Schütz
Prof. Dr. med. Min Ae Lee-Kirsch
University Hospital Carl Gustav Carus, Dresden

IN COOPERATION WITH

DFG Deutsche Forschungsgemeinschaft

DGfI Deutsche Gesellschaft für Immunologie e.V.

AKPI

Dear friends and colleagues,

It is our great pleasure to welcome you to the 2025 Annual API meeting!

We have put together an exciting program on the topic of New therapeutic approaches for Immune Disorders, ranging from targeted therapies to cell and gene therapies.

The meeting will be held in the beautiful Kloster Nimbschen, a former estate of the Cistercian monastery *Marienthron* near Leipzig, where Katharina von Bora lived her humble life before marrying Martin Luther. This historic site has been transformed into a charming conference venue.

We would like to thank all sponsors for their generous support.

We hope that Kloster Nimbschen will provide a pleasant and inspiring atmosphere that will encourage collegial interaction, scientific exchange and lively discussions between clinicians and scientists for the benefit of our patients suffering from rare diseases that are not always easy to diagnose or treat.

Kind regards,

Prof. Dr. Catharina Schütz
Meeting Chair

Prof. Dr. Min Ae Lee-Kirsch
Meeting Chair

Prof. Dr. Stephan Ehl
API Chairman

Date

May, 22nd – 24th 2025

Venue

Hotel Kloster Nimbschen
Nimbschener Landstraße 1
D-04668 Grimma, Germany

Scientific Society

Arbeitsgemeinschaft
Pädiatrische Immunologie (API) e.V.
info@kinderimmunologie.de
www.kinderimmunologie.de

Scientific Organizers

Prof. Dr. med. Catharina Schütz
Prof. Dr. med. Min Ae Lee-Kirsch
University Hospital Carl Gustav Carus Dresden, Germany

Congress Organization

b4c & solutions GmbH
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Scientific Committee

Anne Rensing-Ehl
Christine Wolf
Min Ae Lee-Kirsch
Julia Körholz
Almut Meyer-Bahlburg
Catharina Schütz

Congress registration

Please use the registration form at the end of this brochure.

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Congress language

Congress language is English.

Thursday, May 22nd

08:30 – 12:00 **Educational Workshop**
Christian Klemann, Horst von Bernuth, Julia Körholz, Timmy Strauss
Room: Kaysersaal

12:00 – 13:30 **API Vorstandssitzung / Board Meeting**
Room: Teamschmiede

12:00 – 13:30 **Registration / Welcome snack & coffee**
Room: Kulturscheune

13:30 – 13:45 **Welcome Note**
Catharina Schütz & Min Ae Lee-Kirsch

13:45 – 15:00 **Session 1: New Approaches to Gene Therapy**
Chairs: Johannes Rädler, Catharina Schütz

13:45 – 14:20 **Keynote speaker: Alessandra Magnani, Barcelona**

14:20 – 14:32 ADA2 genotype and enzymatic activity predict phenotype and decision probability of HCT in DADA2
Phillip Peters, Munich (Abs. 1)

14:32 – 14:44 Hematopoietic Stem Cell Transplantation in Osteopetrosis: an IEWP study of 715 children
Mehtap Sirin, Ulm (Abs. 2)

14:44 – 14:56 A sleeping beauty-based method for re-expressing of LRBA in an in vitro disease model
Catherine Henzgen, Frankfurt (Abs. 3)

15:00 – 15:30 **Coffee break**

Thursday, May 22nd

15:30 – 17:20 **Session 2: Pathogenesis and Management of Autoinflammatory Disorders**
Chairs: Kerstin Felgentreff, Min Ae Lee-Kirsch

15:30 – 16:05 **Keynote speaker: Raphaela Goldbach-Mansky, Bethesda**

16:05 – 16:17 Novel variant in TNFRSF9 induces EBV-specific immunodeficiency and autoinflammation
Carl Christoph Goetzke, Berlin (Abs. 4)

16:17 – 16:29 LOF variants in MAF contribute to an increased inflammatory activity and severe immunological symptoms including atypical hidradenitis suppurativa thereby extending the known developmental phenotype
Ulrike Hüffmeier, Erlangen (Abs.5)

16:29 – 16:41 A Novel Homozygous IFNAR2 Variant in Three Siblings Reveals Clinical and Immunological Heterogeneity in Type I Interferon Signaling Deficiency
Franziska Dunst, Leipzig (Abs. 6)

16:41 – 16:53 Clinical characteristics, treatments and outcomes of an international cohort of 26 patients with biallelic ZNFX1 variants
Julius Köppen, Zurich (Abs. 7)

16:53 – 17:05 Global Disparities in Defining and Treating Colchicine Resistance in Familial Mediterranean Fever: A CLiPS Network Analysis
Helmut Wittkowski, Munster (Abs. 8)

17:05 – 17:17 Infection-triggered erosive mucositis in OSA1-GOF following hematopoietic cell transplantation
Johannes Rädler, Munich (Abs. 9)

17:20 – 18:00 **Break**

Thursday, May 22nd

18:00 – 18:20

Short session: Adolescents and Young Adults – Stem Cell Transplantation for inborn errors of immunity
Chair: Rita Beier

Keynote speaker: Michael Albert, München

18:30 – 20:00

Posterwalk I
Chairs: Ursula Holzer, Christian Klemann

Poster 01: Inflammasome activation in preterm and term neonates
Hannah Siedel, Dresden (Abs. 10)

Poster 02: Deficient anterior pituitary with variable immune deficiency (DAVID Syndrome): 2 unrelated cases due to NFKB2 variants
Carolin Müller, Düsseldorf (Abs. 11)

Poster 03: 6-year-old boy with autoimmune lymphoproliferative immunodeficiency (ALPID)
Marie Gerisch, Leipzig (Abs. 12)

Poster 04: Hyper IgE syndrome and EBV-driven Burkitt leukaemia – a causal relation?
Rita Beier, Hannover (Abs. 13)

Poster 05: HLH and severe CMV pneumonitis as early symptoms of a ZNFX1 defect
Ommo Mauss, Ulm (Abs. 14)

Poster 06: A novel hemizygous nonsense variant in DOCK11 causes systemic inflammation and immunodeficiency
Georgios Sogkas, Hannover (Abs. 15)

Poster 07: The Impact of Dupilumab Treatment on the Respiratory Tract in STAT3 Hyper IgE Syndrome
Natalie Maßberg, Munich (Abs. 16)

Poster 08: Potential of Janus Kinase Inhibition in the Treatment of Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy (APECED)
Lina Igel, Düsseldorf (Abs. 17)

Thursday, May 22nd

Poster 09: An autoinflammatory syndrome with high interferon signature in a child with complete deficiency of the IFN-λ receptor
Christine Wolf, Dresden (Abs. 18)

Poster 10: Hyperinflammation and MPO-ANCA vasculitis due to loss of OAS2 function
Rou Liu, Dresden (Abs. 19)

Poster 11: An atypical case of IPEX syndrome with combined immunodeficiency and anti-IL-6 autoantibodies expands the clinical spectrum of the disease
Tiziana Lorenzini, Zurich (Abs. 20)

Poster 12: Registry Proposal: Anifrolumab Therapy for Type I Interferonopathies
Stavrieta Soura, Krefeld (Abs. 21)

Poster 13: Allogeneic stem cell transplantation in a patient with CTLA-4-defciency and neurologic remitting disease
Ursula Holzer, Tübingen (Abs. 22)

Poster 15: JAK-Inhibition as Treatment Option for severe Alopecia areata: a Case Series.
Christian Karl Braun, Ulm (Abs. 25)

20:00

Dinner
Room: Hotel Restaurant

21:00

Junge API - Get-together
Room: Kulturscheune

Friday, May 23rd

08:30 – 09:30 **Session 3: Update on Immune Dysregulatory Disorders**
Chairs: Maria Faßhauer, Kaan Boztug

08:30 – 09:05 **Keynote speaker: Sophie Hambleton, Newcastle**

09:05 – 09:17 The role of unconventional T cells in autoimmune disorders
Artem Kalinichenko, Graz (Abs. 26)

09:17 – 09:29 LTβR deficiency causes lymph node aplasia and impaired B-cell differentiation
Kaan Boztug, Wien (Abs. 27)

09:30 – 09:50 **Coffee break**

09:50 – 11:30 **Session 4: Thymus Biology after Tissue Transfer**
Chairs: Mehtap Sirin, Horst von Bernuth

09:50 – 10.25 **Keynote speaker: Alexandra Kreins, London**

10:25 – 10:37 Management and outcome of syndromic IEI in TREC-NBS: experiences from the German Screening Program
Lea Graafen, Düsseldorf (Abs. 28)

10:37 – 10:49 A monoallelic PSMB10 mutation impairs immunoproteasome assembly and causes reversible neonatal T lymphopenia due to a thymic epithelial defect
Fabian Hauck, Munich (Abs. 29)

10:49 – 11:01 Post-natal thymectomy vs. congenital defects of thymus development: when a few months really matter
Ondřej Vladyha, Prague (Abs. 30)

11:01 – 11:13 Distinct Immunological and Clinical Profiles in NFKB2-Related Disease: Evidence From 142 Patients Across Three Molecular Subtypes
Christian Klemann, Leipzig (Abs. 31)

11:13 – 11:25 Time from suspected SCID in TREC newborn screening to definitive treatment – can we do better?
Stine Augustin, Berlin (Abs. 32)

Friday, May 23rd

11:30 – 13:30 **Lunch**

13:30 – 15:00 **Session 5: New Concepts of Cell Therapeutics**
Chairs: Sybille Landwehr-Kenzel, Fabian Hauck

13:30 – 14.05 **Keynote speaker: Michael Sieweke, Dresden**

14:05 – 14:17 Exploring human γδ T cell immunity through inborn errors of immunity
Maximilian Tank, Freiburg (Abs. 33)

14:17 – 14:29 Allogeneic stem cell transplantation with treosulfan-based conditioning as appropriate consolidation in Nijmegen breakage syndrome with second diffuse large B-cell lymphoma (DLBCL)
Jörn-Sven Kühl, Leipzig (Abs. 34)

14:29 – 14:41 Phenotypic differences of T Helper subpopulations in patients with inflammatory diseases as a guide for personalized therapy
Ales Janda, Ulm (Abs. 35)

14:41 – 14:53 Extended clinical phenotypes and treatment modalities in 32 JAGN1 deficient patients. A multi-center study by ESID and EBMT IEWP.
Shahrzad Bakhtiar, Frankfurt (Abs. 36)

15:00 – 15:20 **Coffee break**

Friday, May 23rd

15:20 – 16:35 **Session 6: TGFβ links EBV to multisystem inflammatory syndrome in children**
Chairs: Oliver Wegehaupt, Jana Pachlopnik-Schmid

15:20 – 15.55 **Keynote speaker: Tilmann Kallinich, Berlin**

15:55 – 16:07 IKBKB gain-of-function: an inborn error with clinical heterogeneity progressing towards a combined immunodeficiency
Julia Körholz, Dresden (Abs. 37)

16:07 – 16:19 ITPR3-Associated Immunodeficiency: A Novel Multisystem Disorder Challenging Current Diagnostics
Sybille Landwehr-Kenzel, Hannover (Abs. 38)

16:19 – 16:31 HLH in patients with EBV-positive NK/T cells (CAEBV and lymphoma) has distinct features, relevant prevalence in any ethnicity, and poor outcome
Helena Lichtenfeld, Hamburg (Abs. 39)

16:35 – 16:50 **Coffee break**

16:50 – 17:35 **AKPI Frühjahrestreffen / Spring meeting**
Room: Kulturscheune

17:35 – 19:35 **General assembly (API Members only)**
Room: Kulturscheune

20:00 **Dinner & Get-together**
Room: Hotel Restaurant

Saturday, May 24th

08:30 – 09:40 **Session 7: Monogenic Forms of Lupus**
Chairs: Susan Farmand, Henner Morbach

08:30 – 09:00 **Keynote speaker: Min Ae Lee-Kirsch, Dresden**

09:00 – 09:12 Inborn Errors of Immunity Presenting as Rheumatologic Disease: Implications for Diagnostics and Treatment Modification to Improve Patient Outcome
Susan Farmand, Hamburg (Abs. 40)

09:12 – 09:24 JAK inhibitors in the treatment of autoimmune polyendocrine syndrome type 1 (APS1) – chance and challenge
Kerstin Felgentreff, Ulm (Abs. 41)

09:24 – 09:36 High frequency of adult onset neurological manifestations in CVID(-like) patients
Leif Hanitsch, Berlin (Abs. 42)

09:40 **Grab-a-coffee and start of poster walk**

09:45 – 11:15 **Poster walk II**
Chairs: Ellen Renner, Ulrich Baumann

Poster 16: False-Positive SCID Screening in a Newborn with Noonan Syndrome-Associated Juvenile Myelomonocytic Leukemia
Olga Staudacher, Berlin (Abs. 43)

Poster 17: A novel frameshift variant in DIAPH1 in a patient with seizures, cortical blindness, and microcephaly (SCBMS) – a syndrome associated with immunodeficiency and increased sensitivity to radiation
Gisela Fecker, Erlangen (Abs. 44)

Poster 18: Scoring thoracic CT scans of patients with NFKB1-related disease for clinical management and treatment studies using four parameters (adapted from the Hartmann Score)
Katharina Helene Thoma, Freiburg (Abs. 45)

Saturday, May 24th

Poster 19: Optimizing Thymic Allograft Cultivation: Enhancing Viability, Function, and Future Biobanking Potential
Karina Amineva, Dresden (Abs. 46)

Poster 20: Severe ARDS due to PCP-infection as first presentation of Hyper-IgM – or not to forget the zebras
Ommo Mauss, Ulm (Abs. 47)

Poster 21: Phenotypic and pathomechanistic overlap between tapasin and TAP deficiencies
Abdulwahab Elsayed, Hannover (Abs. 48)

Poster 22: Deficiency of RAD17: a novel monogenic disorder with granulomas and defective DNA damage response
Ralf Wiedemuth, Dresden (Abs. 49)

Poster 23: Case report: A complex recovery after a thymus transplant
Adam Klocperk, Prague (Abs. 50)

Poster 24: The role of human LRBA protein in NF-κB regulation
Theresa Füller, Frankfurt (Abs. 51)

Poster 25: Syndrome-associated B cell differentiation defect in a 5-year old with global developmental delay and SPATA5 variants
Svenja Keil, Krefeld (Abs. 52)

Poster 26: Immunophenotypic and functional analysis in a child with Meier-Gorlin syndrome due to a mutation in DONSON gene – suspecting a defect in DNA replication initiation
Stephan Borte, Leipzig (Abs. 53)

Poster 27: Lymphocytic interstitial pneumonia associated with common variable immunodeficiency
Solomiya Tymchyshyn, Lviv (Ukraine) (Abs. 54)

Poster 28: Recurrent nocardiosis in patient with chronic granulomatous disease
Yaryna Romanyshyn, Lviv (Ukraine) (Abs. 55)

Poster 29: Advanced Diagnostic Approaches in Primary Immunodeficiency: The CCI ADU Diagnostics Lab, Freiburg
Jens Wittner, Freiburg (Abs. 56)

PROGRAM

Saturday, May 24th

Poster 30: Low donor chimerism still provides sufficient immune function after allogeneic stem cell transplantation for CARMIL2 immunodeficiency
Sven Starke, Leipzig (Abs. 57)

Poster 31: Impact of Digenic Heterozygous Variants in FHL Genes on HLH Susceptibility
Oliver Wegehaupt, Freiburg (Abs. 58)

11:15 – 12:40 **Session 8: 15 Years of Human Phenotype Ontology**
Chairs: Olga Staudacher, Tim Niehues

11:15 – 11:40 **Keynote speaker: Peter Robinson, Berlin**

11:40 – 12:00 Somatic mosaicism and immune disease
Stephan Ehl, Freiburg (Abs. 59)

12:00 – 12:12 Deep Immunoprofiling for Enhanced Stratification of Paediatric Patients with Severe Inflammatory Response Syndrome
Sebastian Thieme, Dresden (Abs. 60)

12:12 – 12:24 Pulmonary Manifestations in Patients with Inborn Errors of Immunity (IEI) – Insights from an ESID Registry Study
Johanna Krista, Munich (Abs. 61)

12:24 – 12:36 Natural IgM (nIgM) in Autoimmune Diseases and Organ Transplantation
Ulrich Bosch dos Santos, Bochum (Abs. 62)

12:40 – 13:05 **Poster award with industrial support,**
Travel grants,
Next API
Closing remarks

Lunch to-go

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