



## Programm

### 33. Jahrestagung der API

*zusammen mit dem 7.  
Jahrestreffen des  
Arbeitskreises  
Pädiatrische  
Immunologie der DGfI*

5.5. – 7.5.2016

Kartause  
Ittingen (CH)

Tagungspräsident:

*Horst v. Bernuth, Berlin*

Kongressorganisation:

*DI-TEXT Frank Digel*

## AG-Sitzungen

9:00 – 12:00 Uhr  
PID-NET meeting

10:00 – 12:00 Uhr  
AG Neugeborenencreening

## Freitag | 6. Mai 2016

## Mitgliederversammlung

18:30 – 20:00

**Mitgliederversammlung**

Vorsitz: Tim Niehues, Krefeld & Volker Umlauf, Sankt Augustin

- TOP 1      Festsetzung der Tagesordnung und Feststellung der Beschlussfähigkeit**
- TOP 2      Genehmigung des Protokolls 2015**
- TOP 3      Bericht des Vorsitzenden**  
*Tim Niehues, Krefeld*
- TOP 4      Bericht des Schatzmeisters und der Geschäftsführung**  
*Volker Umlauf, Sankt Augustin*
- TOP 5      Bericht der Kassenprüfer**  
*Klaus Schwarz, Ulm & Nikolaus Rieber, München*
- TOP 6      Entlastung des Vorstands**
- TOP 7      Vorstandswahlen**
- TOP 8      Wahl des Kongresspräsidenten/-in 2018**
- TOP 9      Berichte aus den Arbeitsgruppen**
- *DGfI*
  - *ESID*
  - *IEWP*
  - *Neugeborenen-Screening*
  - *DGKJ-Konvent*
  - *AG Transition*
- TOP 10    Verschiedenes**

## 1. Sitzung

13:00 – 15:15 Uhr

**Innate Immunity I**

Vorsitz: Horst von Bernuth, Berlin &amp; Jana Pachlopnik-Schmid, Zurich

30' + 10'      **Immunodeficiencies predisposing to fungal infections**  
Anne Puel, Paris

10' + 5'      **Freie Vorträge**  
**Update on extrapulmonary Aspergillus infections in patients with CARD9 deficiency**  
*Nikolaus Rieber, Tübingen & Munich*

**Diverse macrophage phenotypes during pulmonary staphylococcal infection in mice with myeloid STAT3-deficiency**  
*Susann Farmand, Stockholm*

**Anti-IL-6-autoantibodies cause bacterial infections with low penetrance**  
*Stephanie Heller, Berlin*

**NF-kappaB mutations in common variable immunodeficiency**  
*Manfred Fliegau, Freiburg*

5' + 5'      **Kasuistiken**  
**Common variable immunodeficiency, impaired neurological development and reduced numbers of T-regulatory cells in a 10 year-old boy with a STAT1 gain-of-function mutation**  
*Robbin Kobbe, Hamburg*

15:15 – 15:45

**Kaffeepause**

## 2. Sitzung

15:45 – 18:30 Uhr

**Innate Immunity II**

Vorsitz: Catharina Schütz, Ulm &amp; Fabian Hauck, München

30' + 10'      **Extracellular chromatin in infections and autoimmunity**  
*Arturo Zychlinsky, Berlin*

10' + 5'      **Freie Vorträge**  
**A novel complex immunodeficiency with impaired cytoskeletal dynamics responsive to lenalidomide**  
*Elisabeth Salzer, Vienna*

**Reactive oxygen species permit nuclear entry of hydrolases during NETosis**  
*Virginia M Spaccamela, Zürich*

**Burst Screen: a diagnostic feasibility study for newborn screening of severe congenital neutropenias and of phagocytic function**  
*Stephan Borte, Leipzig*

**Flow cytometric based analysis of monocytes as a platform for diagnosis of primary immunodeficiencies**  
*Sandra Ammann, Freiburg*

5' + 5'      **Kasuistiken**  
**Acute Granulomatous necrotizing colitis in a 4-month-old infant**  
*Shahrazad Bakhtiar, Frankfurt*

**Pneumocystis jirovecii pneumonia (PJP) in a child with Hoyeraal Hreidarsson Syndrom**  
*Gregor Dückers, Krefeld*

ab 18:30

**Abendessen**

Abend zur freien Verfügung

## 3. Sitzung

13:00 – 15:15 Uhr

**Innate Immunity III**

Vorsitz: Kaan Boztug, Vienna &amp; Philipp Henneke, Freiburg

30' + 10'

**Intracellular DNA-sensing mechanisms**

Andrea Ablasser, Lausanne

10' + 5'

**Freie Vorträge****A pore ring mutation in the protein-conducting Sec61 channel is causing plasma cell deficiency in humans**

Bodo Grimbacher, Freiburg

**The clinical phenotype and treatment options in a large cohort of CTLA4 mutation carriers**

Charlotte Schwab, Freiburg

**Development of drug screening approaches for primary immunodeficiencies**

Birgit Höger, Vienna

**Cellular and humoral influenza-specific immune response upon vaccination in patients with common variable immunodeficiency and unclassified antibody deficiency**

Leif G Hanitsch, Berlin

**Long-term safety, efficacy, and tolerability of recombinant human hyaluronidase-facilitated subcutaneous infusion of immunoglobulin G in patients aged < 18 years with primary immunodeficiencies**

Norbert Zessack, Munich

**A novel macrophage subset instructs immunity in staphylococcal skin infections**

Philipp Henneke, Freiburg

10:00 – 10:30 Uhr

**Kaffeepause**

## 4. Sitzung

10:30 – 12:45 Uhr

**Novel therapeutic approaches I**

Vorsitz: Manfred Hönig, Ulm &amp; Tim Niehues, Krefeld

30' + 10'

**Present and future potential of stem cell transplantation**

Tayfun Güngör, Zurich

10' + 5'

**Freie Vorträge****The German PID-NET registry**

Sabine El-Helou

**Adoptive transfer of regulatory T-cells ameliorates chronic graft-versus host disease and improves lymphocyte engraftment**

Sybille Landwehr-Kenzel, Berlin

5' + 5'

**Kasuistiken****A novel Wiskott-Aldrich-like syndrome treated by sequential maternal lymphocyte infusions and TCRAB/CD19 depleted peripheral stem cell transplantation**

Markus Seidel, Graz

**Erythrocyte exchange transfusion and early stem cell transplantation to treat prenatally diagnosed PNP-deficiency**

Michael Albert, Munich

**Thymic transplant in complete DiGeorge Syndrome (DGS)**

Florence Junghans, Ulm

**Thymic transplant for a 10 month old boy with complete DiGeorge syndrome; severe vaccine associated Rota Virus infections and pneumonia**

Gregor Dückers, Krefeld

12:45 – 13:30

**Mittagessen**

## 5. Sitzung

13:30 – 16:00 Uhr

**Novel therapeutic approaches II**

Vorsitz: Janine Reichenbach, Zurich &amp; Christoph Klein, Munich

30' + 10'

**Novel strategies for efficient editing of the genome***Boris Fehse, Hamburg*

10' + 5'

**Freie Vorträge****Incidence of SCID – German pediatric surveillance unit (ESPED) study***Tim Niehues, Krefeld***API and GPOH registry for the therapy of patients with SCID***Manfred Hönig, Ulm***Differences in long-term immunological outcome in sib pairs transplanted for SCID***Catharina Schütz, Ulm***The profound combined immunodeficiency (PCID) study – update on the first 100 recruited patients***Carsten Speckmann, Freiburg***Phenotypic spectrum of 9 patients with Hyper-IgM syndrome***Mehtap Sirin, Ulm*

5' + 5'

**Kasuistiken****Heterozygote RAG2 mutation in a boy with immune dysregulation – when functional analysis is required to assess the significance of genetic findings***Susann Farmand, Stockholm*

16:00 – 16:30 Uhr

**Kaffeepause**

## 6. Sitzung

16:30 – 18:30 Uhr

**Innate Immunity IV**

Vorsitz: Ulrich Baumann, Hannover &amp; Bodo Grimbacher, Freiburg

30' + 10'

**Mendelian immunodeficiencies in NK-cells***Jordan S. Orange, Houston*

10' + 5'

**Freie Vorträge****Human CD27 deficiency: Follow-up, further characterization, registry***Ruy Perez-Becker, Krefeld***Rapid flow cytometry-based test for the diagnosis of LRBA deficiency***Sophie Jung, Freiburg***Human RLTPR-deficiency***Fabian Hauck, Munich*

5' + 5'

**Kasuistiken****Immunological abnormalities in an atypical case of chronic active EBV (CAEBV) infection with fulminant hepatitis***Elke Lainka, Essen***Maternal administration of parenteral noxa in Munchausen Syndrome by proxy can elicit the full picture of hemophagocytic lymphohistiocytosis***Kai Lehmborg, Hamburg*

18:30 – 20:00 Uhr

**Mitgliederversammlung**

ab 20:00 Uhr

**Abendessen****Abend zur freien Verfügung**

## 7. Sitzung

13:30 – 16:00 Uhr

**Autoinflammation I**

Vorsitz: Angela Rösen-Wolff, Dresden &amp; Tilmann Kallinich, Berlin

30' + 10'

**Genetically defined autoinflammatory diseases: A journey in Autoinflammation***Raphaela Goldbach-Mansky, Bethesda (USA)*

10' + 5'

**Freie Vorträge****Pro-inflammatory signaling by enzymatically inactive caspase-1 in vivo***Angela Rösen-Wolff, Dresden***Screening of 180 CVID patients for DADA2 sheds new light on the disease in adulthood***Johanna Schepp, Freiburg*

5' + 5'

**Kasuistiken****Heterozygote RAG2 mutation in a boy with immune dysregulation – when functional analysis is required to assess the significance of genetic findings***Susann Farmand, Stockholm***Fever of unknown origin, hematological, cutaneous and neurological symptoms in children: rule out ADA2 deficiency***Gregor Dückers, Krefeld***Metabolic abnormalitis in a patient presenting with recurrent septic episodes and severe lymphopenia at birth***Artem Kalinichenko, Vienna***Interleukin-1 blockade with canakinumab for Hyper-IgD syndrome (HIDS)***Jürgen Brunner, Innsbruck***Juvenile arthritis in a patient with a CVID phenotype – association with a BANK1 mutation***Maria Fasshauer, Leipzig*

10:00 – 10:30 Uhr

**Kaffeepause**

## 8. Sitzung

10:30 – 12:40 Uhr

**Autoinflammation II**

Vorsitz: Dirk Föll, Münster &amp; Carsten Speckmann, Freiburg

30' + 10'

**Familial Mediterranean Fever: Gene-dose does matter***Tilmann Kallinich, Berlin*

10' + 5'

**Freie Vorträge****Evolution of disease activity and biomarkers on and off rapamycin in 27 ALPS patients***Christian Klemann, Freiburg***S100A12 as diagnostic tool in the differential diagnosis of sJIA associated MAS vs hereditary or acquired HLH***Dirk Holzinger, Münster***Validation of MRP8/14 serum levels as biomarker for the diagnosis of systemic juvenile idiopathic arthritis in fever of unknown origin***Dirk Holzinger, Münster***In patients with activated PI3 kinase delta syndrome, CD57 identifies CD8+ T-cells with exhausted proliferative capacity before terminal differentiation and pronounced telomere shortening***Paola Cura Daball, Freiburg*

5' + 5'

**Kasuistiken****Accumulation of  $\gamma\delta$  T-cells in a patient with autoimmune lymphoproliferative syndrome (ALPS)***Stefano Vavassori, Zurich***Hemophagocytic Lymphohistiocytosis (HLH) due to Lysinuric Protein Intolerance (LPI)***Amelia Rösel, Berlin*

**Anne Puel**

*Laboratory of Human Genetics of Infectious Diseases, Necker Branch; French Institute of Health and Medical Research (INSERM) U1163; Imagine Institute, Paris Descartes University, Paris, France*

**Arturo Zychlinsky**

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**Andrea Ablasser**

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**Tayfun Güngör**

*University Children's Hospital Zurich, Division of Immunology, Zurich, Switzerland*

**Boris Fehse**

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**Jordan S. Orange**

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**Raphaela Goldbach-Mansky**

*Translational Autoinflammatory Diseases Section, National Institute of Arthritis, Musculoskeletal and Skin diseases (NIAMS), National Institutes of Health (NIH), Bethesda, MD, USA*

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