



# 36<sup>th</sup> API ANNUAL MEETING

17<sup>th</sup> – 19<sup>th</sup> May 2019

Abbey Frauenwörth, Frauenchiemsee, Germany

*together with the 10<sup>th</sup> Annual Meeting of the  
Working Group Pediatric Immunology of the German Society of Immunology*

[www.kinderimmunologie.de](http://www.kinderimmunologie.de)

## Organ-specific

## Features of Inborn Errors of Immunity

## Program

Congress President:  
Fabian Hauck, MD, PhD (Munich)



ARBEITSGEMEINSCHAFT

**PÄDIATRISCHE IMMUNOLOGIE**

**Dear API members and friends,**

The ever-exciting immune system is going on to teach us about its complexity. While we all have appreciated the systemic character of immunity, recently the organ-specific particularities are coming back into the spotlight.

This is why we have dedicated the 36th annual meeting of the Arbeitsgemeinschaft Pädiatrische Immunologie (API), i.e. the German Speaking Working Group for Paediatric Immunology, to the main topic of Organ-specific Features of Inborn Errors of Immunity. We will have outstanding keynote presentations on particular immunological compartments by internationally renowned physician-scientists and are counting on your active participation.

We are looking forward to your most recent translational and basic research findings on inborn errors of immunity.

As our meeting venue we have chosen the idyllic Abbey *Frauenwörth* located on the island *Fraueninsel* placed in the heart of the lake *Chiemsee* in southeast Bavaria. Even though the beautiful landscape and the Mountain View might distract you, we are daring to take the risk.

We are looking forward to welcome you, to listen to your latest findings and to discuss them with you in the most interactive setting of our 36<sup>th</sup> annual API meeting.

Fabian Hauck, MD, PhD

## Joint Venture

10th Annual Meeting of the Working Group Pediatric Immunology of the German Society of Immunology (DGfI)

## Date

17th – 19th of May 2019

## Congress President

Fabian Hauck, MD, PhD

Dr. von Haunersches Kinderspital

Kinderklinik und Kinderpoliklinik der Ludwig Maximilian Universität München

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D-80337 München

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## Organizer / Scientific Society

Arbeitsgemeinschaft Pädiatrische Immunologie (API) e.V.

Arnold-Janssen-Str. 25

53757 Sankt Augustin

E-Mail: [info@kinderimmunologie.de](mailto:info@kinderimmunologie.de)

Web: [www.kinderimmunologie.de](http://www.kinderimmunologie.de)

## Congress Organization

DI-TEXT Frank Digel

Ruhwarder Str. 19

D-26969 Butjadingen-Ruhwarden

E-Mail: [Digel.F@t-online.de](mailto:Digel.F@t-online.de)

## Congress registration

Deadline: 1<sup>st</sup> of April 2019

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

## Congress language

Congress language is English.

## Abstract submission

Deadline: 1<sup>st</sup> of April 2019, extended to 14<sup>th</sup> of April 2019

Submitting an abstract is mandatory if you intend to hold a lecture or case report. Please submit the abstracts in English by e-mail to the following address: [Fabian.Hauck@med.uni-muenchen.de](mailto:Fabian.Hauck@med.uni-muenchen.de).

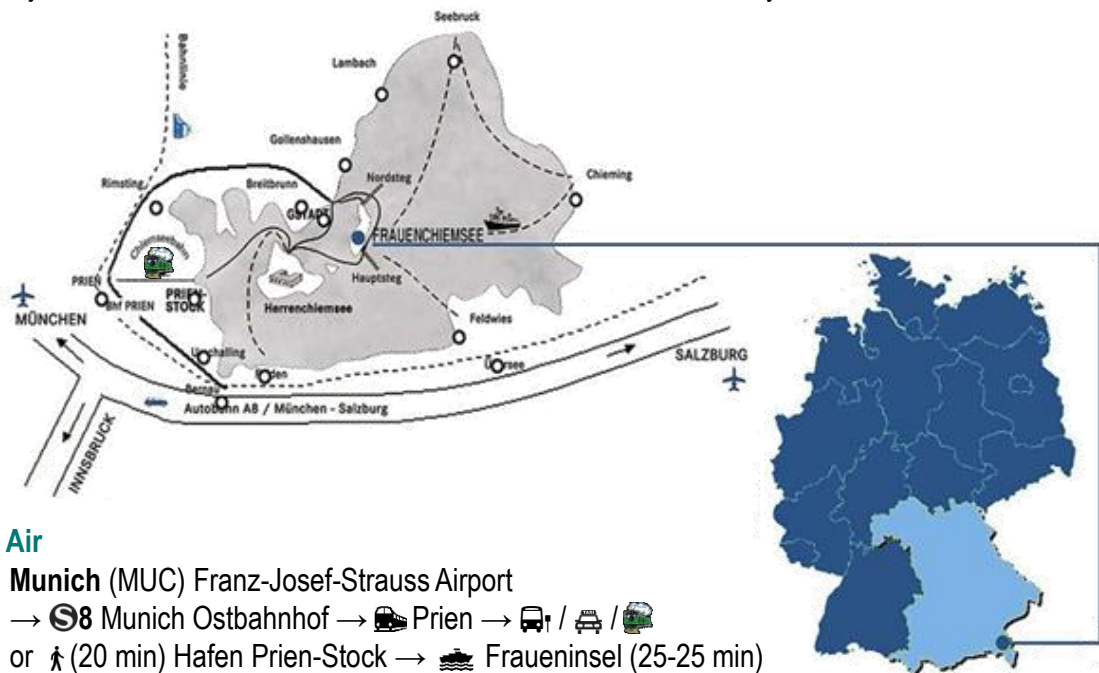
Please structure your abstract into "Background, Methods, Results, Conclusions" and limit the text to 250 words.

Lecture: 10 min + 5 min discussion; presentations with more than 10 slides will not be accepted.

Case report: 5 min + 5 min discussion; presentations with more than 5 slides will not be accepted.

## Venue

Abbey Frauenwörth, Frauenchiemsee 50, 83256 Chiemsee, Germany



## By Air

✈ **Munich (MUC) Franz-Josef-Strauss Airport**  
 → Munich Ostbahnhof → Prien → / /   
 or (20 min) Hafen Prien-Stock → Fraueninsel (25-25 min)

✈ **Salzburg (SZG) W. A. Mozart Airport** → / Salzburg Main Station → Prien  
 → / / or (20 min) Hafen Prien-Stock → Fraueninsel (25-25 min)

## By Train

**Munich - Salzburg**, runs every hour; → Prien → / / or (20 min) Hafen Prien-Stock  
 → Fraueninsel (25-25 min)

## By Car

Munich – Salzburg A8 → Bernau, via Prien to direction Rimsting/Breitbrunn to Gstadt  
 → **P** → Fraueninsel (10 min)

Car Parking (w/ fee) near by Gstadt harbour, e.g.:

- Seeplatz 4 · 83257 Gstadt
- Reiterstr. 9 / Breitbrunner Str. 12, 83257 Gstadt

## Chiemsee Ferry

Gstadt - Fraueninsel: 07:20 | 08:05 | 08:50 | 09:20 | 09:50 | 10:20 | 10:50 | 11:20 | 11:50 | 12:20 | 12:50 | 13:20 | 13:50 | 14:20 | 14:50 | 15:20 | 15:50 | 16:20 | 16:50 | 17:20 | 17:50 | 18:55 | 19:50 | 20:50 | 20:50 | 22:50

2-way-ticket Gstadt-Fraueninsel: 6,30 €

See latest timetable and fees at: <https://www.chiemsee-schiffahrt.de/>

10:00 – 13:00

**Pre-meeting**

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10:00 – 11:00

AG Neugeborenenenscreening

10:00 – 12:00

Registration for the API meeting

12:00 – 12:45

Pre-meeting lunch

12:45 – 13:00

Welcome address

*Fabian Hauck (Munich)*

13:00 – 15:00

**Session 1: The lymphatic organs**

*Chairs: Carsten Speckmann (Freiburg); Ursula Holzer (Tübingen)*

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13:00 – 13:40 (30'+10')

The lymphatic organs

*Frédéric Rieux-Laucat (Paris)*

13:40 – 13:55 (10'+5')

Human interleukin-2 receptor  $\beta$  mutations are associated with defects in immunity and peripheral tolerance

*Florian Gothe (Munich)*

13:55 – 14:10 (10'+5')

Functional clustering of activating germline STAT3 mutations associates with disease penetrance

*Maximilian Heeg (Freiburg)*

14:10 – 14:25 (10'+5')

Long-lived and stable evolution of a protective human T cell system by a putative single T cell precursor

*Stephan Ehl (Freiburg)*

14:25 – 15:40 (10'+5')

Late thymic deficiency after HLA-haploidentical hematopoietic stem cell transplantation for severe combined immunodeficiency

*Catharina Schütz (Dresden)*

14:40 – 14:50 (5'+5')

T-cell impairment is predictive for a severe clinical course in NEMO-deficiency

*Horst von Bernuth (Berlin)*

14:50 – 15:00 (5'+5')

A heterozygous IKBKG mutation causes incontinentia pigmenti and a fever syndrome in a female patient with variable NEMO expression in blood mononuclear cells

*Kerstin Felgentreff (Ulm)*

**15:00 – 15:30**      **Coffee break and Poster Session 1**

**15:30 – 17:30**      **Session 2: The lungs**

*Chairs: Henner Morbach (Würzburg), Jana Pachlopnik-Schmid (Zurich)*

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- 15:30 – 16:10 (30'+10')**      **The lungs**  
*Matthias Griese (Munich)*
- 16:10 – 16:25 (10'+5')**      **Heterozygous OAS1 Gain-of-Function Variants Cause a Polymorphic Autoinflammatory and Immunodeficiency Syndrome**  
*Fabian Hauck (Munich)*
- 16:25 – 16:40 (10'+5')**      **Interstitial lung disease in COVID patients – a cross sectional study in 102 adult patients**  
*Leif Hanitsch (Berlin)*
- 16:40 – 16:55 (10'+5')**      **Clinical and immunological phenotype of patients with primary immunodeficiency due to damaging mutations in NFKB2**  
*Christian Klemann (Hannover)*
- 16:55 – 17:10 (10'+5')**      **Nanopore sequencing as tool to analyze total genes, to phase parental alleles and to detect large indels in DNA and RNA: Useful in PID?**  
*Marita Führer (Ulm)*
- 17:10 – 17:20 (5'+5')**      **Novel genetic and immunological aspects of COPA syndrome**  
*Martin Wetzke (Hannover)*
- 17:20 – 17:30 (5'+5')**      **Lethal ARDS after stem cell transplantation in a child with homozygous STAT1 LOF**  
*Lisa-Maria Köhler (Munich)*

**18:00**      **Dinner at Klosterwirt**

8:00 – 8:45

**Breakfast**

9:00 – 10:30

**Session 3: The liver**

*Chairs: Kai Lehmborg (Hamburg), Shahrzad Bakhtiar (Frankfurt)*

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9:00 – 9:40 (30'+10')

The liver

*Philip Buefler (Berlin)*

9:40 – 9:55 (10'+5')

Recurrent Inflammatory Disease due to a heterozygous Mutation in CD48

*Jana Pachlopnik Schmid (Zurich)*

9:55 – 10:10 (10'+5')

Genetic causes and new therapeutic approach in monogenic systemic lupus erythematosus

*Sarah Koss (Dresden)*

10:10 – 10:20 (5'+5')

Conventional treatment versus stem cell transplantation – outcome in 104 patients with chronic granulomatous disease

*Horst von Bernuth (Berlin)*

10:20 – 10:30 (5'+5')

A novel NCF4 family highlights the mild disease phenotype in p40phox autosomal-recessive chronic granulomatous disease

*Sujal Ghosh (Düsseldorf)*

10:30 – 11:00

**Coffee break and Poster Session 2**

11:00 – 12:30

**Session 4: The Brain**

*Chairs: Fabian Hauck (Munich), Kerstin Felgentreff (Ulm)*

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11:00 – 11:40 (30'+10')

The brain

*Shen-Ying Zhang (New York)*

11:40 – 11:55 (10'+5')

Identification and characterization of a new genetic cause of a type 1 interferonopathy

*Min Ae Lee-Kirsch (Dresden)*

11:55 – 12:10 (10'+5')

Neuroimaging findings in children with hereditary hemophagocytic lymphohistiocytosis – report from the German reference center

*Kai Lehmborg (Hamburg)*

- 12:10 – 12:20** (5'+5') Predominant neurological manifestations accompanied by hepatosplenomegaly in a case of late-onset familial hemophagocytic lymphohistiocytosis type 5  
*Nora Naumann-Bartsch (Erlangen)*
- 12:20 – 12:30** (5'+5') Allogeneic Hematopoietic Stem Cell Transplantation in Ataxia Telangiectasia Patients without Malignancy  
*Shahrazad Bakhtiar (Frankfurt)*
- 12:30 – 13:15** **Lunch**
- 13:30 – 15:00** **Session 5: The blood vessels**  
*Chairs: Helmut Wittkowski (Münster), Min Ae Lee-Kirsch (Dresden)*
- 
- 13:30 – 14:10** (30'+10') The blood vessels  
*Marco Gattorno (Genoa)*
- 14:10 – 14:25** (10'+5') Autoinflammatory syndrome due to Interferon Lambda Receptor (IFNLR1) deficiency  
*Marc Bienias (Dresden)*
- 14:25 – 14:40** (10'+5') Hyperactivation of TLR signaling in a family with monogenic systemic lupus erythematosus  
*Barbara Kind (Dresden)*
- 14:40 – 14:50** (5'+5') STING-associated vasculopathy in mice requires adaptive immunity but not type I interferon or cGAS  
*Angela Rösen-Wolff (Dresden)*
- 14:50 – 15:00** (5'+5') Ruxolitinib for the treatment of immune dysregulation in PID: bridge or alternative to transplant? Experiences from a case series  
*Carsten Speckmann (Freiburg)*
- 15:00 – 15:30** **Coffee break and Poster Session 3**



15:30 – 17:00

**Session 6: The neoplasms**

*Chairs: Sujal Ghosh (Düsseldorf), Susan Farmand (Stockholm)*

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15:30 – 16:10 (30+10')

The neoplasms

*Jürgen Ruland (Munich)*

16:10 – 16:25 (10+5')

Novel inborn error of immunity causes immune dysregulation with predisposition to lymphomagenesis

*Marini Thian (Vienna)*

16:25 – 16:40 (10+5')

Successful Hematopoietic Stem Cell Transplantation in a patient with recurrent EBV-induced lymphoproliferation due to a homozygous missense mutation in 4-1BB

*Christine Wildermann (Ulm)*

16:40 – 16:50 (5+5')

Unusual presentation and early transplant course of a JAK3-SCID patient

*Jörn-Sven Köhl (Leipzig)*

16:50 – 17:00 (5+5')

Dysgammaglobulinemia, progressive B cell deficiency and loss of T cell naivety in a family with a novel heterozygous, haploinsufficient IKZF1 mutation

*Ann Bökstegers (Munich)*

17:00 – 17:15

**Presentation of the Walter Hitzig Award 2019**

17:30 – 18:45

**API members' general assembly**

19:00

**Dinner at Inselwirt**

8:00 – 8:45

**Breakfast**

9:00 – 10:30

**Session 7: The body surface**

*Chairs: Michael Albert (Munich), Catharina Schütz (Dresden)*

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9:00 – 9:40 (30'+10')

The body surface

*Joshua Milner (Bethesda)*

9:40 – 9:55 (10'+5')

Identification of an inborn error of immunity with atopy and abnormal inflammatory responses

*Sevgi Köstel Bal (Vienna)*

9:55 – 10:10 (10'+5')

Novel inborn error of immunity linking aberrant cytoskeletal dynamics and severe immune dysregulation

*Kaan Boztug (Vienna)*

10:10 – 10:20 (5'+5')

Infantile-onset monogenetic acute generalized pustular psoriasis

*Anna Eichinger (Munich)*

10:20 – 10:30 (5'+5')

Neonatal inflammatory bowel disease as presenting manifestation of chronic granulomatous disease caused by EROS deficiency psoriasis

*Julian Thalhammer (Freiburg)*

10:30 – 11:00

**Coffee break and Poster Session 4**

11:00 – 12:30

**Session 8: The bone**

*Chairs: Kaan Boztug (Wien), Vera Binder (Munich)*

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11:00 – 11:40 (30'+10')

The bone

*Ansgar Schulz (Ulm)*

11:40 – 11:55 (10'+5')

Wiskott-Aldrich Syndrome (WAS): A retrospective study on 575 patients defines the genotype as a predictive biomarker for disease severity and survival

*Michael Albert (Munich)*

11:55 – 12:10 (10'+5')

A retrospective study of 84 transplanted LAD-I and LAD-III patients. Dealing with an Underestimated Inflammatory Disease?

*Shahrzad Bakhtiar (Frankfurt)*

**12:10 – 12:20** (5'+5')

A retrospective study of treatment forms and clinical courses of patients with LPS-responsive beige-like anchor protein (LRBA) deficiency using the Immune deficiency and –dysregulation activity (=IDDA) score

*Markus Seidel (Graz)*

**12:20 – 12:30** (5'+5')

MIRAGE syndrome – A new clinical entity with myelodysplasia, recurrent infections and various syndromic features

*Sujal Ghosh (Düsseldorf)*

**12:30 – 13:30**

**Farewell and take-away lunch**

*Fabian Hauck (Munich)*

## Poster Session 1

Chair: Kerstin Felgentreff (Ulm)

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Impact of Immune Checkpoint Inhibitors (CPI) on B-cell selection checkpoints

*Joshua Boeckers (Münster)*

Immunodeficiency with susceptibility to Morbus Hodgkin lymphoma and variants in RAB27A, ACAD9 and FBP1

*Sandra Ammann (Cambridge)*

CRISPR/Cas9-mediated ELANE gene correction as a gene therapy for Severe Congenital Neutropenia

*Van Trung Chu (Berlin)*

Severe form of Roifman syndrome with congenital diarrhea, agammaglobulinemia and chronic inflammation due to a homozygous RNU4ATAC mutation

*Johannes Dirks (Würzburg)*

Complex, chronic aphthosis and infection with mycobacteria tuberculosis as hallmarks for STAT1 gain of function-associated primary immunodeficiency

*Gregor Dückers (Krefeld)*

STAT-dependent primary immunodeficiencies and environmental signalling

*Renate Effner (Munich)*

## Poster Session 2

Chair: Carsten Speckmann (Freiburg)

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Chromosomal deletion 13q33 with syndromic immunodeficiency in a preterm infant

*Elisabeth Förster-Waldl (Vienna)*

A family with hypogammaglobulinemia, ACTH deficiency, ectodermal dysplasia, Pneumocystis jirovecii pneumonia and a novel NFKB2 mutation

*Linda Geberzahn (Dresden)*

Challenges to Identify the Molecular Basis of Hyper-IgE Syndromes

*Ellen Renner (Munich)*

Severe gastric manifestation of autoimmunity in an immune dysregulation syndrome of an infant

*Marcus Jakob (Regensburg)*

Neonatal Autoimmune-Hypothyroidism as presenting sign of IPEX-like primary immunodeficiency due to CD25 deficiency

*Christian Klemann (Hannover)*

### Poster Session 3

*Chair: Susan Farmand (Stockholm)*

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Unusual infectious complications in a 64 year old patient with epithelioid-cell-like-granulomatous organizing pneumonia

*Julia Körholz (Dresden)*

A systematic review of the literature assessing the frequency and patterns of immune dysregulation in various Primary Immunodeficiencies

*Andrea Mauracher (Zurich)*

Hypomorphic CD40LG-deficiency leading to duodenitis with nontuberculous mycobacteria

*Anita Rack-Hoch (Munich)*

Genetic Diagnostic Elucidation Of A Patient With Multiorgan Granulomas, Facial Peculiarities, And Psychomotor Retardation

*Jochen Roesler (Dresden)*

Severe eczema, hyper IgE and hypogammaglobulinemia in infancy – all due to filaggrin deficiency?

*Elisabeth Rolfes (Berlin)*

### Poster Session 4

*Chair: Christian Klemann (Hannover)*

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IgG4 related disease leading to severe solitary lung fibrosis in a girl

*Katharina Schütz (Hannover)*

Syndrome-associated, congenital neutropenia with splenomegaly and selective mutism due to a heterozygous de novo mutation in SRP54

*Kathrin Siepermann (Krefeld)*

Haploidentical hematopoietic stem cell transplantation for two siblings with STK4-deficiency?

*Stephanie Thee (Berlin)*

Dermatological manifestations of MALT1 deficiency mimicking Netherton- and Omenn syndrome

*Helmut Wittkowski (Münster)*

Manifestation of LRBA- deficiency during the first year of life – a case report

*Christina Zachbauer (Vienna)*

Anmeldung zur 36. Jahrestagung der Arbeitsgemeinschaft Pädiatrische Immunologie (API) e.V.  
17. – 19. Mai 2019, Abtei Frauenwörth (D-83256 Frauenchiemsee)



Anmeldung bis spätestens 01.04.2019  
Die Plätze sind begrenzt!

Titel	Vorname
Name	Insitution
Straße	PLZ und Ort
Telefon-Nr.	Fax-Nr.
E-Mail	

### Anmeldung Vortrag / Fallvorstellung

Vortrag      10 Min. + 5 Min. Diskussion, max. 10 Folien. Vorträge mit >10 Folien werden nicht aufgespielt!

Fallvorstellung      5 Min. + 5 Min. Diskussion, max. 5 Folien. Vorträge mit >5 Folien werden nicht aufgespielt!

Titel: \_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

Für die Anmeldung eines Vortrages/einer Fallvorstellung ist eine Zusammenfassung erforderlich. Bitte reichen Sie die **Abstracts für Ihre Beiträge bis 14.04.2019** in englischer Sprache per E-Mail unter folgender Adresse ein: [Fabian.Hauck@med.uni-muenchen.de](mailto:Fabian.Hauck@med.uni-muenchen.de)

Bitte gliedern Sie Ihre Abstracts in „Background, Methods, Results, Conclusions“ und beschränken Sie den Text auf 250 Wörter.

Hiermit bestelle ich bei der Abteil Frauenwörth verbindlich:

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| <input type="checkbox"/> im Einzelzimmer, Kategorie A, Seeblick, Du/WC     | € 60,00 p.P |
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Weitere Bemerkungen:

\_\_\_\_\_

\_\_\_\_\_

Die Verteilung der Zimmer in der Abtei Frauenwörth wird entsprechend dem Eingang der Anmeldung vorgenommen!

Ich habe zur Kenntnis genommen, dass ich im Falle meiner Verhinderung für die Stornierungskosten selbst aufkommen muss.

\_\_\_\_\_, den \_\_\_\_\_, \_\_\_\_\_  
Ort Datum Unterschrift

### Rücksendung an: Frank Digel

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