



2. LÉKAŘSKÁ FAKULTA  
UNIVERZITY KARLOVY



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## SPRING SCHOOL for Primary Immunodeficiencies

Kutna Hora, The Czech Republic

18th – 20th April 2024

GASK, Barborská 51-53, Kutná Hora



## PROGRAMME

## Thursday 18th April

15:00 – 17:15	<b>Arrival, accommodation</b> <b>hotel “U Varhanare”</b> Barborska 578 / 11, Kutna Hora www.uvarhanare.cz
17:15 – 17:30	<b>Welcome and Introductions</b> <i>Anna SEDIVA (Czech Republic)</i>
17:30 – 18:30	<b>Opening lecture – The immune dysregulation in combined Immunodeficiencies</b> <i>Jolan WALTER (USA)</i>
19:00+	<b>Welcome dinner in a traditional Bohemian restaurant „Dacicky“</b> www.dacicky.com

## Friday 19th April

7:30 – 8:30	<b>Breakfast</b> (at the place of accommodation)
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### **Block I – Thymus and T cell disorders**

**Chairs:** *Alexandra KREINS, Adam KLOCPERK*

8:30 – 9:10	<b>Invited lecture – Thymus transplantation</b> <i>Alexandra KREINS (UK)</i>
9:10 – 10:50	<b>Participant presentations</b> (15 min talk + 5 min discussion)  <i>Adam KLOCPERK (Czech Republic)</i> - Atypical complete DiGeorge syndrome caused by diabetic fetopathy  <i>Kristiina TAMM (Estonia)</i> - Co-occurrence of Ultra-rare Immunodeficiency-related Conditions: A Case Report  <i>Ondřej VLADYKA (Czech Republic)</i> - Alterations in T cell Phenotype and Function in Schimke Immuno-osseous Dysplasia  <i>Dharmagat BHATTARAI (Nepal)</i> - ARPC1B deficiency in children with combined immunodeficiency, thrombocytopenia & autoinflammation: Possible founder effect from Nepal  <i>Petra HADLOVÁ (Czech Republic)</i> - Spectral phosphoflow assay to analyze signaling pathways in IEI patients
10:50 – 11:10	<b>Coffee break</b>

## **Block II – Autoinflammatory, dysregulation, innate**

Chairs: *Anna SEDIVA*

- 11:10 – 11:50 **Invited lecture – A novel monogenic human lupus**  
*Fabian HAUCK (Germany)*
- 11:50 – 13:00 **Participant presentations**  
(15 min talk + 5 min discussion)
- Antonia CRISTE (Romania)* - Unravelling Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED): A case study emphasizing the crucial role of Immunodeficiency and Genetic Evaluation in Recurrent Candidiasis and Skin Rashes
- Martin ORLICKÝ (Czech Republic)* - Geranylgeraniol supplementation in patients with hyper IgD syndrome
- Markéta BLOOMFIELD (Czech Republic)* – Fever of (un)known origin
- Mahnaz JAMEE (UK)* - Successful Hematopoietic Stem Cell Transplantation in Interferon Gamma Receptor Deficiency: Case Presentation and Review of the Literature
- 13:00 – 14:00 **Lunch**

## **Block III – B cell disorders**

Chairs: *Jolan WALTER*

- 14:00 – 14:40 **Invited lecture – Human B-cell development, lessons from PID patients**  
*Tomas KALINA (Czech Republic)*
- 14:40 – 16:00 **Participant presentations**  
(15 min talk + 5 min discussion)
- Ana Rita SOARES (Portugal)* - A severe case of humoral immunodeficiency and autoimmune enteropathy with cachexia associated with CTLA4 haploinsufficiency
- Bianca-Maria CAPRIORU (Romania)* - Pulmonary Actinomycosis in a Patient with Common Variable Immunodeficiency
- Marcelo TEOCCHI (Italy)* - Mild Kabuki Syndrome or Common Variable Immunodeficiency and Asperger Syndrome: a Case Report Highlighting Familial Segregation and Immune Dysregulation
- Jorge Diogo DA SILVA (Portugal)* - A complex genetic condition with a simple clinical presentation: overlapping TACI-associated CVID and GATA2 deficiency
- 16:00 – 16:20 **Coffee break**

- 16:20 – 18:00 **Free social programme**  
Visit to the gallery, chocolate museum, mining museum, sightseeing...
- 18:00 – 20:00 **Dinner restaurant v Ruthardce**  
[www.v-ruthardce.cz](http://www.v-ruthardce.cz)
- 20:30+ **Visit of Saint Barbara's Church with organ concert**  
[www.kutna-hora.net/barbora.php](http://www.kutna-hora.net/barbora.php)

## Saturday 20th April

- 7:30 – 9:00 **Breakfast**  
(at the place of accommodation)

### **Block IV – Miscellaneous**

**Chairs:** *Markéta BLOOMFIELD*

- 9:00 – 10:40 **Participant presentations**  
(15 min talk + 5 min discussion)
- Elena DOLAPCIU (Moldova)* - Chronic granulomatous disease with neonatal onset.
- Karla SKUKAN (Croatia)* – Chronic granulomatous diseases presenting with multiple liver abscesses
- Lucija ŠIMUNIĆ (Croatia)* - Bicytopenia and splenomegaly in a patient with hypodontia – could there be a common cause?
- Konstantinos VAZOURAS (Greece)* - Undefined immunodeficiency with lymphoproliferation and severe viral infections
- Vesna VUKIČEVIĆ LAZAREVIĆ (Croatia)* - A Complex Case: Primary Ciliary Dyskinesia, Selective Immunoglobulin A Deficiency, and Their Impact on Respiratory Health
- 10:40 – 10:50 *Anna SEDIVA (Czech Republic)* - **APDS overview**
- Wrap-up and farewell**  
*Anna SEDIVA*
- 10:50 – 11:10 **Coffee break**
- 11:45 **Departure back to Prague**