

Sitzung der Arbeitsgruppe Genetik der ADF

Donnerstag 10. März 2016

9.30 - 13.00 Uhr

9:30 – 9:40 h Begrüßung

Cristina Has, Klinik für Dermatologie und Venerologie, Universitätsklinikum Freiburg

Vinzenz Oji, Klinik für Dermatologie und Venerologie, Universitätsklinikum Münster

9:40 – 10:10 Identifikation neuer primärer Immundefekte - was Pädiater und Dermatologen voneinander lernen können?

Prof. Priv.-Doz. Dr. Kaan Boztug, CeMM Principal Investigator / Associate Professor, Dept. of Pediatrics and Adolescent Medicine, Medical University of Vienna, Director Vienna Center for Rare and Undiagnosed Diseases (CeRUD)

10:10-10:20 Increased prevalence of filaggrin deficiency in recessive X-linked ichthyosis

Kira Süssmuth, Tatjana Tarinski, Frederic Valentin, Elke Rodriguez, Alberto Sánchez-Guijo, Natalia Straub, Heiko Traupe, Dieter Metze, Stefan Werner Schneider, Ingrid Hausser-Siller, Stephan Weidinger, Amler Susanne, Vinzenz Oji

Münster, Kiel, Giessen, Mannheim, Heidelberg

10:20-10:30 IL36RN, other genes coding for members of the IL-36 pathway, CARD14 and AP1S3 in pustular psoriasis

Oji V., Mössner R. Wilsmann-Theis D., Schulz P., Körber A., Prinz J., Renner S., Schäkel K., Peters K.-P, Philip S., Reich K., Ebertsch L., Ekici A., Uebe S., Thiel C., Sticherling M., Traupe H., Sticht H., Hüffmeier U.

Münster, Erlangen

10:30-10:40 BRAF and RAS Mutations in Sporadic and Secondary Pyogenic Granuloma

Leopold Größer, Mark Berneburg, Regensburg

10:40-10:50 Photosensitive form of trichothiodystrophy associated with a novel mutation in the XPD gene

Brauns B, Schubert S, Lehmann J, Laspe P, Körner A, Brockmann K, Schön MP, Emmert S

Göttingen, Aachen, Osnabrueck, Rostock

10:50-11:00 Mutational spectrum in a large cohort of families with hypohidrotic ectodermal dysplasia and breakpoint mapping in four of five independent cases of rare genomic rearrangements

Sigrun Wohlfart, Johanna Hammersen, Holm Schneider

Erlangen

11:00-11:30 Pause

11:30-11:40 TALEN-mediated elimination of mutant keratin 14 as a gene therapy for epidermolysis bullosa simplex

Magomet Aushev, Claudio Mussolino, Toni Cathomen, Julia Reichelt

Newcastle upon Tyne, Freiburg, Salzburg

- 11:40-11:50 Optimised TALEN-mediated Gene Editing of Keratinocyte Stem Cells for a novel ex vivo Epidermolytic Ichthyosis Therapy**
Oliver March, Magomet Aushev, Ulrich Koller, and Julia Reichelt
Salzburg, Newcastle
- 11:50-12:00 Single amino acid deletion in kindlin-1 results in partial protein degradation which can be rescued by chaperone treatment**
Kristin Maier, Yinghong He, Philipp R. Esser, Kerstin Thriene, Daniela Sarca, Jürgen Kohlhase, Jörn Dengjel, Ludovic Martin, Cristina Has
Freiburg, Angers
- 12:00-12:10 Clinical spectrum in Patients with Ichthyosis with Confetti (IWC)**
Bettina Burger, Peter Itin
Basel
- 12:10-12:20 Assoziation von Cole-Krankheit mit neuen heterozygoten Mutationen in der Somatomedin B-Domäne des ENPP1 Gens: notwendig, aber nicht immer ausreichend**
H. Traupe, N. Schlipf, Y. Gilaberte, W.K. Peitsch, I. Hausser, v. Oji, A. Schmieder, S. W. Schneider, P. Demmer, B. Rösler J. Fischer
Münster, Freiburg ,Mannheim und Heidelberg
- 12:20-12:30 Red scaly Baby**
Cristina Has, Agnes-Schwieger-Briel, Nina Schlipf, Ingrid Hausser, Nadja Chmel, Bernd Rösler, Kristin Technau, Thilo Jakob, Andreas Zimmer, Judith Fischer
Freiburg
- 12:30-13:00 Organisatorisches; Anregungen und Wünsche**
Cristina Has und Vinzenz Oji
- 13:00 Ende der Tagung**