

## **Meeting of the ADF Working Group Translational Dermatological Genetics February 22. 2023, 14:00-18:00**

- 14:00 – 14:10** Introduction Cristina Has (Freiburg), Vinzenz Oji (Münster)
- 14:10 - 15:00** Prof Dr. Heiko Traupe  
**Ichthyosen als Fenster in die Epidermis**
- 15:00 - 15:15** Kira Süßmuth\*, C. Klein, R. Sommer, M. Augustin, S. Ständer, S. Salzmann, K. Kiekbusch, J. Bodes, M. Danzer, H. Traupe, J. Fischer, S. Steinke, V. Oji (Münster)  
**Personal, financial and time burden in inherited ichthyoses: A survey of 144 patients in a university-based setting**
- 15:15 - 15:30** Kevin Koschitzki, Mark Berneburg (Regensburg)  
**Trichothiodystrophie, klinische Beispiele & Exkurs: Der Struwelpeter**
- 15:30 - 15:45** Basmanav FB, Cesarato N, Kumar S, Borisov O, Kokordelis P, Ralser DJ, Wehner M, Axt D, Xiong X, Thiele H, Dolgin V, Gossmann Y, Fricker N, Dewenter MK, Weller K, Suri M, Reichenbach H, Oji V, Addor MC, Ramirez K, Stewart H, Garcia Bartels N, Weibel L, Wagner N, George S, Kilic A, Tantcheva-Poor I, Stewart A, Dikow N, Blaumeiser B, Medvecz M, Blume-Peytavi U, Farrant P, Grimalt R, Bertok S, Bradley L, Eskin-Schwartz M, Birk OS, Bygum A, Simon M, Krawitz P, Fischer C, Hamm H, Fritz G, Betz RC.Regina (Bonn)  
**Das genetische Spektrum des Syndroms der unkämmbaren Haare in einer Kohorte von 107 Individuen**
- 15:45 - 16:00** Y. Wang, A. Hotz, P. R. Esser, J. Fischer, C. Has (Freiburg)  
**Amino acid substitution in the cysteine-rich region of the integrin beta 4 subunit causes mild late onset junctional epidermolysis bullosa without extracutaneous involvement**
- 16:00 - 16:30** Break
- 16:30 - 16:45** Holm Schneider, Christine Schweikl, Florian Faschingbauer, Smail Hadj-Rabia, Nicolai Peschel, Pascal Schneider (Erlangen)  
**Short-term correction of deficient ectodysplasin A signaling with a replacement protein: long-term results in nine patients**
- 16:45 - 17:00** Thomas Trafoier (Innsbruck)  
**CRISPR/Cas9 double nicking treatment restores intermediate filament integrity in keratinocytes derived from patients with EPPK**
- 17:00 - 17:15** Saptaswa Dey, Peter Wolf (Graz)  
**STAT3/5 locus gain is a disease marker and therapeutic target in leukemic/cutaneous T cell lymphoma**
- 17:15 - 17:30** Ulrich Koller (Salzburg)  
**Gene Editing für die junktionale Epidermolysis bullosa**
- 17:30 - 18:00** Discussion
- 18:00** End of the meeting