

## Meeting of the ADF Working Group Translational Dermatological Genetics

February 23. 2022, 14:00-18:00

- 14:00 – 14:10** Introduction Cristina Has (Freiburg), Vinzenz Oji (Münster)
- 14:10 - 14:45** Univ.-Prof. Dr. Katharina Wimmer Institut für Humangenetik, Medizinische Universität Innsbruck  
**"Neurofibromatosen: Genetik, diagnostische Kriterien und neues aus der Forschung"**
- 14:45 - 15:00** I. Petkovic, J. Bischof, B. Liemberger, O. March, J. Reichelt, H. Binder, D. Strunk, J. Bauer, T. Kocher, U. Koller, Salzburg  
**Efficient homology-directed repair CRISPR/Cas9-based gene editing in junctional epidermolysis bullosa keratinocytes**
- 15:00 - 15:15** M. Zulal, Y. Wang, T. Obser, C. Mess, K. Opitz, A. Bauer, S. Schneider, C. Gorzelanny, Hamburg  
**CRISPR/Cas9-based multiplex gene editing to determine the impact of heparan sulfate on the melanoma microenvironment**
- 15:15 - 15:30** V. Banicka, M. Martens, S. Emmert, A. Thiem, Rostock  
**Generation and characterization of different CRISPR/Cas9-mediated XPA-knockouts in A375 melanoma cells**
- 15:30 - 15:45** M. Martens, W. Sponholz, N. Di Donato, J. Porrmann, M. Hasanvodzic, T. Scholz, M. Hempel, S. Emmert, L. Boeckmann, Rostock, Dresden, Tuzla, Hamburg  
**Xeroderma pigmentosum: Genetic und functional analyses of eight new patients**
- 15:45 - 16:15** Break
- 16:15 - 16:30** A. Reimer-Taschenbrecker, A. Künstner\*, M. Hirose, S. Hübner, S. Gewert, S. Ibrahim, H. Busch, C. Has, Freiburg, Lübeck  
**Predominance of Staphylococcus correlates with wound burden and disease activity in dystrophic epidermolysis bullosa: a prospective case-control study**
- 16:30-16:45** H. Stanisz, C. Mitteldorf, M. Schoen, J. Frank, Goettingen  
**Subcellular compartmentalization of STIM1 may distinguish Darier disease from Hailey-Hailey disease**
- 16:45-17:00** A. Zaremba, Essen  
**Molekularpathologie und Methylierungsprofile als diagnostische Unterstützung bei schwer zu klassifizierenden melanozytären Tumoren mit spitzoider Morphologie**
- 17:00-17:15** L. Hake, K. Suessmuth, C. Drerup, K. Komlosi, J. Kopp, D. Metze, H. Traupe, I. Hausser, K. Eckl, H. Hennies, J. Fischer, V. Oji, Buxtehude, Muenster, Freiburg, Heidelberg, Huddersfield, Innsbruck, Cologne  
**Quality of life and clinical characteristics of self-improving congenital ichthyosis within the disease spectrum of autosomal recessive congenital ichthyosis**
- 17:15-17:30** M. Krumbiegel, C. Kraus, A. Reis, H. Seybold, P. Wörl, F. Kiesewetter, J. Fischer, J. Küsel, B. Hartmann, U. Hüffmeier, Freiburg, Erlangen  
**Blended phenotype in a family with Netherton syndrome**
- 17:30-17:45** V. Moosbrugger-Martinz, S. Blunder, C. Barbieux, M. Bonnet des Claustres, M. Schmuth, A. Hovnanian, R. Gruber, Innsbruck, Paris  
**Treatment of Netherton syndrome with omalizumab**
- 17:45-18:00** Discussion
- 18:00** End of the meeting

**Zoom-Meeting beitreten**

**<https://us06web.zoom.us/j/84778976835?pwd=UTgyUUFeRzFnMW5wQ0U0d0hPZ0EvQT09>**

**Meeting-ID: 847 7897 6835**

**Kenncode: 789308**

**Meeting-ID: 847 7897 6835**

**Ortseinwahl suchen: <https://us06web.zoom.us/u/kjKVFjl7>**