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# THE TRANSLATIONAL SCIENCE OF RARE DISEASES

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Research on Rare Diseases is of critical relevance to 300 million people affected worldwide by more than 8.000 rare disease entities. Additionally, it may open new horizons to understand fundamental pathomechanisms and to develop new therapies for common disorders. This symposium will focus on cutting-edge insights from basic and clinical science and their translation into the development of novel therapeutic strategies for rare diseases. The Research for Rare Consortium, supported by the German Federal Ministry of Education and Research, is looking forward to sharing innovative and exciting results and hopes to increase enthusiasm for rare diseases research.

# THE TRANSLATIONAL SCIENCE OF RARE DISEASES

## From Rare to Care IV

SPONSORED BY THE



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and Research

**LMU**KLINIKUM



September 29 - October 01, 2021

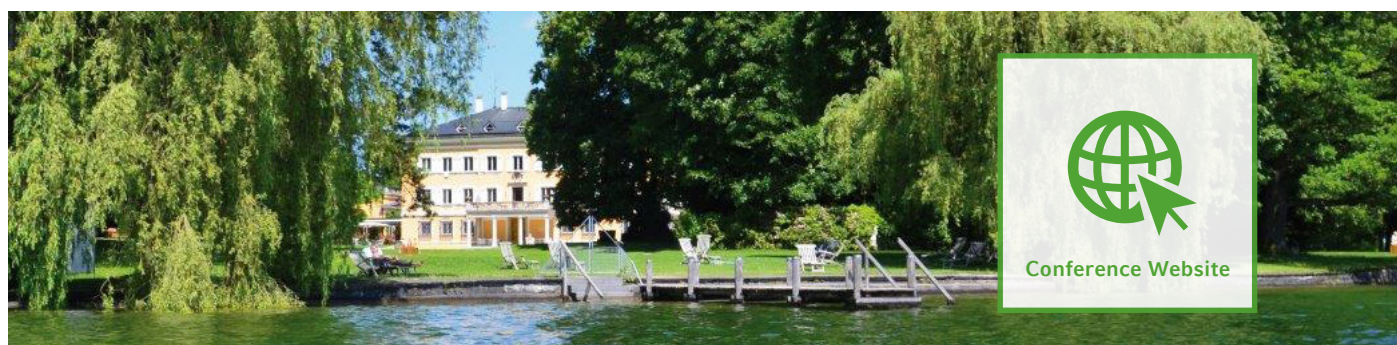


Evangelische Akademie Tutzing  
Schlossstr. 2+4, 82327 Tutzing (Lake Starnberg)



Participate: [>>online registration<<](#)

Participation is free of charge - registration is required



## SCIENTIFIC ADVISORY BOARD

**CHRISTIAN KRATZ**, Hannover Medical School  
ADDRess - Disorders with abnormal DNA damage response

**FRANK LEYPOLDT**, University Hospital Schleswig-Holstein  
CONNECT-GENERATE - Autoimmune encephalitis

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TreatHSP.net - Hereditary spastic paraplegias

**CHRISTOPH KLEIN**, University Hospital LMU, Munich



RESEARCH FOR RARE  
Forschung für seltene Erkrankungen

**Responsible:**

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## WEDNESDAY, SEPTEMBER 29, 2021

01:00 PM REGISTRATION

02:00 PM WELCOME ADDRESS

**Thomas Klopstock**, University Hospital LMU, Munich, Germany

## SESSION 1 OMICS I

02:15 PM **Integration of multi-omics data in patients with rare diseases**  
**Christoph Bock**, Research Center for Molecular Medicine, Vienna, Austria

02:45 PM **Omics signatures in patients with monogenetic multi-organ autoimmunity**  
**Bodo Grimbacher**, Medical Center – University of Freiburg, Germany

03:05 PM **Childhood brain tumors: a heterogeneous collection of rare diseases**  
**David Jones**, German Cancer Research Center (DKFZ), Heidelberg, Germany

03:25 PM **Single-cell genomics of childhood brain tumors**  
**Paul Northcott**, St. Jude Children's Research Hospital, Memphis, USA

03:55 PM **Reprogrammed tubule cells for kidney disease modeling**  
**Soeren Lienkamp**, University of Zurich, Switzerland

04:25 PM BREAK

## SESSION 2 ARTIFICIAL INTELLIGENCE

04:45 PM **Artificial intelligence in pathology and beyond**  
**Peter Boor**, University Hospital of RWTH Aachen, Germany

05:15 PM **The Human Phenotype Ontology (HPO) and its applications in machine learning**  
**Peter Robinson**, The Jackson Laboratory for Genomic Medicine, Farmington, USA

## PATIENTS IN RESEARCH

05:45 PM **Patient involvement in research projects**  
**Virginie Bros-Facer**, EURORDIS, Paris, France

06:15 PM END OF THE DAY

THURSDAY, SEPTEMBER 30, 2021

**SESSION 3 OMICS II**

- 08:00 AM **Integrated omics in the diagnosis of mitochondrial diseases**  
Holger Prokisch, Technical University of München, Munich, Germany
- 08:20 AM **Advances in omics technologies for rare diseases**  
Stephan Ossowski, University Hospital Tübingen, Germany
- 08:40 AM **Multimomics and connectivity mapping approach to decode renal disease**  
Tobias Huber, University Medical Center Hamburg-Eppendorf, Germany
- 09:00 AM **Hyperexcitable interneurons can trigger migraine attacks via rising potassium levels in an *Scn1a* mouse model**  
Tobias Freilinger, Klinikum Passau & University Tübingen, Germany
- 09:20 AM - Selected poster talks -

09:45 AM **POSTER SESSION****SESSION 4 GENE THERAPY**

- 11:00 AM **Adeno-associated viral vectors for gene therapy of rare diseases**  
Hildegard Büning, Hannover Medical School, Hanover, Germany
- 11:30 AM **Epigenetic editing**  
Marianne Rots, University of Groningen, the Netherlands
- 12:00 PM **Crisp editing of stem cells**  
Matthew Porteus, Stanford University School of Medicine, Stanford, USA
- 12:30 PM **Gene therapy in ultra rare diseases - a path for industry/academia collaboration**  
Petra Kaufmann, Novartis Gene Therapies, Bethesda, USA

01:00 PM **BREAK****SESSION 5 PANEL Gene therapy in spinal muscular atrophy**

- 02:00 PM **Medical perspective**  
Janbernd Kirschner, University Hospital Bonn, Germany
- 02:10 PM **Industry perspective**  
Uwe Ernst, Novartis Gene Therapies, Munich, Germany
- 02:20 PM **Ethical perspective**  
Georg Marckmann, Ludwig-Maximilians-University Munich, Germany
- 02:30 PM **Health technology assessment perspective**  
Stefan Lange, Institute for Quality and Efficiency in Health Care, Cologne, Germany
- 02:40 PM **Patient perspective**  
Eva Stumpe, German Society for Muscular Dystrophy Association (DGM), Germany
- 02:50 PM - Panel discussion -

04:00 PM **BREAK****SESSION 6 PATHOMECHANISMS I**

- 04:30 PM **Molecular therapies in mitochondrial diseases**  
Carlos Moraes, University of Miami Miller School of Medicine, Miami, USA
- 05:00 PM **Single molecule trafficking in autoantibody-mediated encephalitis**  
Laurent Groc, University of Bordeaux, France
- 05:30 PM **Axon degeneration: mechanistic insights yield therapeutic opportunities**  
Aaron DiAntonio, Washington University School of Medicine, St. Louis, USA *(to be confirmed)*
- 06:00 PM **Progression from preleukemia to frank leukemia: lessons learned from children with Down syndrome**  
Jan-Henning Klusmann, University Hospital Halle (Saale), Germany

06:20 PM **END OF THE DAY**

FRIDAY OCTOBER 01, 2021

**SESSION 7 PATHOMECHANISMS II**

- 08:30 AM **Genotype correlates with the natural history of severe bile salt export pump deficiency**  
**Henkjan Verkade**, University Medical Center Groningen (UMCG), the Netherlands
- 09:00 AM **From synapse to trials: translational research in autoimmune encephalitis**  
**Christian Geis**, Jena University Hospital, Jena, Germany
- 09:20 AM **Molecular and clinical characterization of a new ciliary protein**  
**Carsten Bergmann**, Limbach Genetics - Medizinische Genetik Mainz, Germany
- 09:40 AM **iPSC-based modeling of RASopathy-related cardiac hypertrophy reveals new mechanisms and therapeutic options**  
**George Kensah**, University Medical Center Göttingen, Germany
- 10:00 AM **Hereditary intrahepatic cholestasis**  
**Verena Keitel-Anselmino**, University Hospital Düsseldorf, Germany

10:20 AM BREAK

**SESSION 8 PANEL European Reference Networks**

- 11:00 AM **ERN-RND**  
**Holm Graessner**, University Hospital Tübingen, Germany
- 11:10 AM **ERN GENTURIS**  
**Nicoline Hoogerbrugge**, Radboud university medical center, Nijmegen, the Netherlands
- 11:20 AM **ERKNet**  
**Franz Schaefer**, Heidelberg University Medical Center, Heidelberg, Germany
- 11:30 AM **ERN-RITA**  
**Nico Wulffraat**, University Medical Center Utrecht, the Netherlands
- 11:40 AM **- Panel discussion -**

12:45 PM CLOSING REMARKS AND FAREWELL

01:00 PM END