The Translational Science of Rare Diseases

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

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

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TreatHSP.net - Hereditary spastic paraplegias

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Responsible
Prof. Dr. Thomas Klopstock
Friedrich-Baur-Institut an der
Neurologischen Klinik
LMU Klinikum
Ziemssenstraße 1a
80336 Munich, Germany

Organisation/Registration:
Katja Franke-Rupp; Dr. Corinna Schultheis
Phone: +49 (0)89 4400-57063 /-57064
E-Mail: info@research4rare.de
www.research4rare.de
### WEDNESDAY, SEPTEMBER 29, 2021

<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
<th>Event</th>
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<tbody>
<tr>
<td>01:00 PM</td>
<td>Registration</td>
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<tr>
<td>02:00 PM</td>
<td>Welcome Address</td>
<td>Thomas Klopstock, University Hospital LMU, Munich, Germany</td>
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</tbody>
</table>
| 02:15 PM | Session 1: Omics I       | Integration of multi-omics data in patients with rare diseases
Bodach Bock, Research Center for Molecular Medicine, Vienna, Austria |
| 02:45 PM |                          | Omics signatures in patients with monogenic 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                    |                                                                      |
| 04:45 PM | Session 2: Artificial Intelligence | 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|
| 05:45 PM | Patients in Research     | 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 Proksch, 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  Multimics and connectivity mapping approach to decode renal disease  
           Tobias Huber, University Medical Center Hamburg-Eppendorf, Germany

09:00 AM  Hyperrexcitable 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
## SESSION 7  PATHOMECHANISMS II

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

## SESSION 8  PANEL European Reference Networks

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<thead>
<tr>
<th>Time</th>
<th>Panel Name</th>
<th>Speaker</th>
<th>Institution</th>
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<tbody>
<tr>
<td>11:00 AM</td>
<td>ERN-RND</td>
<td>Holm Graessner, University Hospital Tübingen, Germany</td>
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<tr>
<td>11:10 AM</td>
<td>ERN GENTURIS</td>
<td>Nicoline Hoogerbrugge, Radboud university medical center, Nijmegen, the Netherlands</td>
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<td>11:20 AM</td>
<td>ERKNet</td>
<td>Franz Schaefer, Heidelberg University Medical Center, Heidelberg, Germany</td>
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<tr>
<td>11:30 AM</td>
<td>ERN-RTA</td>
<td>Nico Wulffraat, University Medical Center Utrecht, the Netherlands</td>
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<tr>
<td>11:40 AM</td>
<td>- Panel discussion -</td>
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12:45 PM CLOSING REMARKS AND FAREWELL

01:00 PM END