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AUDIENCE

Physicians & scientists interested in therapies of rare diseases
Young investigators are particularly welcome!
Limited number of travel grants available.

Venue
Evangelische Akademie Tutzung
Schlossstr. 2+4, 82327 Tutzing, Lake Starnberg

Organisation/ Registration
Katja Franke-Rupp
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www.research4rare.de

CONFIRMED SPEAKERS

- **ODILE BOESPFLUG-TANGUY, Centre Hospitalière Universitaire Robert Debré, Paris (France)**
- **DAVID COX, Broad Institute, Cambridge, Massachusetts (USA)**
- **BILL GAHL, Undiagnosed Diseases Program, NIH, Washington (USA)**
- **ILSE GANTOIS, McGill University, Montreal (Canada)**
- **VOLKMAR GIESELMANN, University of Bonn (Germany)**
- **DAN KASTNER, Intramural Research, NIH, Washington (USA)**
- **ROBERT KLETA, UCL London (Great Britain)**
- **PETRA KAUFMANN, National Center for Advancing Translational Sciences (NCATS), Washington (USA)**
- **ALEIXO MUISE, The Hospital for Sick Children, Toronto (Canada)**
- **JOSEF PENNINGER, Institute of Molecular Biotechnology, Vienna (Austria)**
- **ANITA RAUCH, University of Zürich, Zürich (Switzerland)**
- **LAURA DE ROSA, Univ. of Modena and Reggio Emilia (Italy)**
- **SCOTT SNAPPER, Boston Children's Hospital, Boston (USA)**
- **NICO WULFFRAAT, University Medical Center Utrecht, Utrecht (The Netherlands)**

Presentations from the Research Networks on Rare Diseases in Germany

REGISTRATION
www.rare2care2018.wordpress.com
(until March 30, 2018)
ABSTRACTS INVITED!

Picture: 4design-ersart - Fotolia

From Rare to Care III
April 11 - 13, 2018
Tutzing, Lake Starnberg (Bavaria)

SCIENTIFIC ORGANISATION:

RESEARCH FOR RARE
Forschung für seltene Erkrankungen

Federal Ministry of Education and Research

ABSTRACTS INVITED!
www.rare2care2018.wordpress.com

Dear guests, colleagues and friends,

on behalf of the German National Research Networks on Rare Diseases we would like to invite you to our international symposium "Translational Science of Rare Diseases" in Tutzing / Lake Starnberg.

Research on Rare Diseases is not only of critical relevance to patients suffering from one of more than 7.000 rare disease entities but may open new horizons to understand fundamental biological principles and to develop novel therapeutic strategies for common disorders. This symposium will focus on cutting-edge insights and their relevance for the development of therapies.

We are grateful that the German Ministry of Education and Research has initiated a special funding program on rare diseases. After a decade of support we wish to share exciting results and to increase enthusiasm for rare disease research.

We are looking forward to welcome you in Tutzing, Lake Starnberg!

With best regards,

Christoph Klein

Professor of Pediatrics
Dr. von Hauner Children's Hospital
Ludwig-Maximilians-University Munich
Christoph.Klein@med.uni-muenchen.de

With kind support of:

Care-for-Rare Foundation

Wed, April 11, 2018

▸ Session 1 - 2:00 p.m.

Welcome Address

Petra Kaufmann: „NCATS - Catalyzing Translational Innovation"

William Gahl: „Clinical challenges in patients with RD and International Networks"

Anita Rauch: „New insights into the etiology of epileptic encephalopathy"

Reiner Siebert: „Epimutations at imprinted loci: from maternal effect mutations to potential treatment targets"

▸ Session 2 - 4:30 p.m.

Odile Boespflug-Tanguy: „Genetic disorders of the white matter: from animal models to patients for the development of diagnosis and therapies"

Andre Reis: „Chromatin-Net – empowering patients, discovering genes and dissecting mechanisms in disorders with defective chromatin dynamics"

Frank Kaiser: „Novel genes and pathomechanisms in Cornelia de Lange syndrome"

Jens Volkmann: „DysTract - Dystonia Translational Research and Therapy Consortium"

Thu, April 12, 2018

▸ Session 3 - 9:00 a.m.

Dan Kastner: „Rare Autoinflammatory Disorders"

Johannes Roth: „Novel molecular mechanisms of auto-inflammatory diseases"

Nico Wulfraat: „Early Anakinra therapy for Systemic Juvenile Idiopathic Arthritis (SIJA): Standard of care?"

Ilse Gantois: „Metformin, a promising drug for the treatment of Fragile X Syndrome and Autism Spectrum Disorders"

Poster session

▸ Session 4 - 11:20 a.m.

Volkmar Gieselmann: „New developments in the therapy of lysosomal storage diseases"

Angela Schulz: „Longterm safety and efficacy of intracereb roventricular enzyme replacement therapy in children with CLN2 disease: Two year results from an ongoing extension study"

Martin Konrad / Jens König: „The phenotypic spectrum of early onset hereditary cystic kidney diseases"

▸ Session 5 - 1:30 p.m.

Successful collaboration between academia & pharmacy – fact or fiction?

Patrick Bäuerle, Managing Director MPM

Peter-Andreas Löschmann, Senior Med. Director, Pfizer Germany

Ingmar Hoerr, Chairman CureVac

Olaf Ritzeler, Director External Innovation RD, Sanofi

Bert Klebl, Director Lead Discovery Center

Thomas Kramps, Cluster Med. Manager Rare Diseases, Novartis

Round Table Debate

▸ Session 6 - 4:30 p.m.

Presentation of selected abstracts

Scott Snapper: „Cytokine defects in children with rare inflammatory bowel diseases"

Aleixo Muise: „New genetic defects in very early onset inflammatory bowel diseases"

Christoph Klein: „Rare Diseases with disorders in intestinal homeostasis"

Josef Penninger: „Engineering human blood vessels"

Fri, April 13, 2018

▸ Session 7 - 9:00 a.m.

Robert Kleta: „Renal Fanconi Syndromes, 100 years later"

Michael Sereda: „Translational Medicine in CMT"

Rudolf Martini: „Inflammation in models for inherited peripheral neuropathies-commonalities, disease-specific features and possible translational aspects"

Marcus Moeller: „FSGS, a rare disease to understand a common condition, chronic kidney disease"

Tobias Huber: „Systems biology reveals novel mechanisms in podocyte disease and FSGS"

▸ Session 8 - 11:10 a.m.

David Cox: „RNA editing with CRISPR-Cas13"

Laura de Rosa: „Epidermal stem cell-mediated cell and gene therapy for Junctional Epidermolysis Bullosa"

Nicole Endlich: „Cutting edge imaging to analyze a rare degenerative disorder, FSGS, in zebrafish"

Chi Wang Ip: „Rodent models of dystonia - from gene to disease and therapeutic approaches"