THE TRANSLATIONAL SCIENCE OF RARE DISEASES

From Rare to Care IV

3 - 5 May 2023

Evangelische Akademie Tutzing
Germany

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SCIENTIFIC PROGRAMME

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SCIENTIFIC ORGANISATION:

RESEARCH FOR RARE
German Networks on Rare Diseases

LMU KLINIKUM

SPONSORED BY THE

Federal Ministry of Education and Research

Coordinators of the Research Networks on Rare Diseases:

Christian Kratz - Hannover Medical School
ADDRess - *Disorders with abnormal DNA damage response*

Frank Leypoldt - University Hospital Schleswig-Holstein, Kiel
CONNECT-GENERATE - *Autoimmune encephalitis*

Bodo Grimbacher - Medical Center – University of Freiburg
GAIN - *Multi-organ autoimmune diseases*

Martin Zenker - University Hospital Magdeburg
GeNeRARe - *RASopathies*

Verena Keitel-Anselmino - University Hospital Magdeburg
HiChol - *Hereditary intrahepatic cholestasis*

Thomas Klopstock - LMU Klinikum, Munich
mitoNET - *Mitochondrial diseases*

Miriam Erlacher - Medical Center – University of Freiburg
MyPred - *Syndromes predisposing to myeloid malignancies*

Tobias B. Huber - University Medical Center Hamburg-Eppendorf
STOP-FSGS - *Focal segmental glomerulosclerosis*

Marcus Möller - University Hospital of RWTH Aachen
STOP-FSGS - *Focal segmental glomerulosclerosis*

Jens König - University Hospital Münster
NEOCYST - *Hereditary cystic kidney diseases*

Rebecca Schüle - Heidelberg University Hospital
TreatHSP.net - *Hereditary spastic paraplegias*

Holger Lerche - University Hospital Tübingen
Treat-ION - *Neurological ion channel and transporter disorders*

Christoph Klein - LMU Klinikum, Munich
PID-NET - *Primary immunodeficiency disorders (2009-2018)*

WELCOME ADDRESS

Dear guests, colleagues and friends,

On behalf of the German National Research Networks on Rare Diseases, we welcome you cordially to our international symposium „The Translational Science of Rare Diseases – from Rare to Care IV".

The Federal Ministry of Education and Research has been funding such Networks on Rare Diseases since 2003, an exceptionally successful program. It has fostered basic and clinical research on rare diseases in Germany, leading to many excellent publications and international visibility, and - most importantly - to marked improvements in diagnostics, treatment and care.

This symposium will focus on recent progress in cutting-edge technologies employed in rare disease research, such as multiOmics, gene therapy (including AAV vectors, RNA therapies, gene editing), machine learning, and others. For this, we were able to attract very renowned national and international speakers.

A specific characteristic of this meeting is its interdisciplinarity. The fact that the Rare Disease Networks operate in different areas of medicine (including hematology, hepatology, human genetics, immunology, nephrology, neurology, and pediatrics) is reflected by the composition of speakers and presentations. We hope that this will stimulate inspiring and inventive discussions among all participants, off the beaten track, and lead to novel ideas and collaborations.

In this spirit, we wish you a successful meeting and enjoyable days here at Lake Starnberg.

Best regards,

Thomas Klopstock

Department of Neurology
University Hospital, LMU Munich

Rebecca Schüle

Department of Neurology
Heidelberg University Hospital

WELCOME ADDRESS Bettina Stark-Watzinger

Federal Minister of Education and Research

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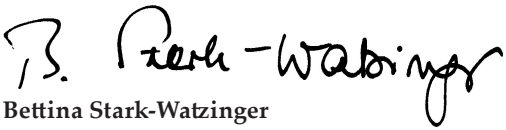
Dear reader,

Researchers of rare diseases often have a more difficult time than others gaining attention, funding and partners which can support their work in many ways. However, there is a number that speaks for all your efforts: the more than 300 million people worldwide who are affected by rare diseases, who lose quality of life and often many years of life itself. This is why your hard work is so valuable, whether it is in the lab, on or off the stage of this symposium.

It is you who tirelessly do investigative work and painstakingly follow up on every clue as to the causes of the disease in question and on what could lead to novel possible treatments. We provide our researchers with the conditions that support them in this endeavour in the best possible way. Our ministry, the Federal Ministry of Education and Research, has been providing funding for rare diseases research for twenty years now. Between 2003 and 2022, we granted a total of €144 million for 39 national research collaborations which draw together capacities and expertise from highly innovative basic as well as clinical research. The knowledge gain has been enormous, which is why we are determined to continue on this successful course and provide another €21.5 million for nine funded networks to continue their research activities. This is our contribution to better diagnostics, effective treatments and, above all, hope for the more than 300 million people who are affected.

I am most pleased that the progress made through our national research programme will once again be channelled into such a top-level international event as yours: the fourth "From Rare to Care" symposium. I am certain that you will make intensive use of this excellent platform for an exchange about the latest methods and formats of cooperation for the benefit of patients – ideally also beyond the borders of disciplines and countries. After all, the rarer the disease, the more exchange is necessary.

To all participants and organizers alike: many thanks for your important work.



Bettina Stark-Watzinger

Member of the German Bundestag
Federal Minister of Education and Research

THE TRANSLATIONAL SCIENCE OF RARE DISEASES

From Rare to Care IV

3-5 May 2023 | Evangelische Akademie Tutzing, Germany

SCHEDULE

WEDNESDAY, 3 MAY 2023

13:00	REGISTRATION
14:00	WELCOME ADDRESS **Thomas Klopstock,** LMU Klinikum, Munich **Rebecca Schüle,** Heidelberg University Hospital

SESSION 1 OMICS I — Chair: Frank Leypoldt

14:15	**Integration of multi-omics data in patients with rare diseases** **Christoph Bock,** Research Center for Molecular Medicine (CeMM), Vienna
14:45	**Omics signatures in patients with common variable immunodeficiency (CVID)** **Bodo Grimbacher,** Medical Center – University of Freiburg - ***GAIN***
15:05	**Childhood brain tumors: a heterogeneous collection of rare diseases** **David Jones,** German Cancer Research Center (DKFZ), Heidelberg - ***ADDRess***
15:25	**Single-cell genomics of childhood brain tumors** **Paul Northcott,** St. Jude Children's Research Hospital, Memphis
15:55	**Reprogrammed tubule cells for kidney disease modeling** **Soeren Lienkamp,** University of Zurich
16:25	BREAK

SESSION 2 LECTURE — Chair: Thomas Klopstock

16:45	**The Human Phenotype Ontology (HPO) and its applications in machine learning** **Peter Robinson,** The Jackson Laboratory for Genomic Medicine, Farmington
17:15	SELECTED POSTER TALKS
17:45	END OF SESSIONS
18:30	DINNER

SCHEDULE

THURSDAY, 4 MAY 2023

SESSION 3	OMICS II	Chair: Holger Lerche
08:15	**Dissecting genetics and pathophysiology of mitochondrial diseases by multi-layered omics** **Dmitrii Smirnov,** Technical University of Munich (TUM) - ***mitoNET***	
08:35	**Advances in omics technologies for rare diseases** **Stephan Ossowski,** University Hospital Tübingen - ***TreatHSP.net***	
08:55	**Multiomics and connectivity mapping approach to decode renal disease** **Tobias Huber,** University Medical Center Hamburg-Eppendorf (UKE) - ***STOP-FSGS***	

09:15	POSTER SESSION / BREAK

SESSION 4	GENE THERAPY	Chair: Christian Kratz
10:30	**Adeno-associated viral vectors for gene therapy of rare diseases** **Hildegard Büning,** Hannover Medical School	
11:00	**Epigenetic editing: engineering the epigenome** **Rutger Gjaltema,** University of Amsterdam	
11:30	**Genome editing of stem cells** **Matthew Porteus,** Stanford University School of Medicine	
12:00	**Personalised RNA therapies for ultrarare diseases** **Annemieke Aartsma-Rus,** Leiden University Medical Center (LUMC)	
12:30	**Gene therapy in ultra rare diseases - a path for industry/academia collaboration** **Petra Kaufmann,** Affinia Therapeutics, Waltham	

13:00	LUNCH BREAK

THURSDAY, 4 MAY 2023

SESSION 5 — PANEL Gene therapy for rare diseases

Chairs: Maggie Walter, Wolfgang Müller-Felber

14:00	**Medical perspective** **Janbernd Kirschner**, Medical Center – University of Freiburg
14:10	**Industry perspective** **Günter Harms**, Novartis Gene Therapies, Munich
14:20	**Ethical perspective** **Ralf Jox**, Lausanne University Hospital (CHUV)
14:30	**Health technology assessment perspective** **Stefan Lange**, Institute for Quality and Efficiency in Health Care, Cologne
14:40	**Patient perspective** **Eva Stumpe**, German Society for Patients with Muscular Diseases e.V. (DGM) / SMA Europe e.V.
14:50	**- Panel discussion -**
16:00	BREAK

SESSION 6 — PATHOMECHANISMS I

Chair: Maja Lindenmeyer

16:30	**Towards gene therapy of mtDNA diseases** **Michal Minczuk**, University of Cambridge
17:00	**Single molecule trafficking in autoantibody-mediated encephalitis** **Laurent Groc**, CNRS - University of Bordeaux
17:30	**Progression from preleukemia to frank leukemia: lessons learned from children with Down Syndrome** **Jan-Henning Klusmann**, University Hospital Frankfurt - ***MyPred***
17:50	**Hyperexcitable interneurons can trigger migraine attacks via rising potassium levels in an *Scn1a* mouse model** **Tobias Freilinger**, Klinikum Passau & University of Tübingen - ***Treat-ION***
18:10	END OF SESSIONS
18:30	DINNER

SCHEDULE

FRIDAY, 5 MAY 2023

SESSION 7	PATHOMECHANISMS II	Chair: Jens König
09:00	**Deep learning 4 kidney pathology... and beyond** **Peter Boor,** University Hospital of RWTH Aachen - ***STOP-FSGS***	
09:30	**Progressive liver, kidney and heart degeneration in adults with genetic ciliary disease** **Carsten Bergmann,** Medizinische Genetik Mainz - ***NEOCYST***	
09:50	**Genotype-phenotype relationships in genetic transport defects affecting bile production and implications for patient care** **Henkjan Verkade,** University Medical Center Groningen (UMCG)	
10:20	**From synapse to trials: translational research in autoimmune encephalitis** **Christian Geis,** Jena University Hospital - ***CONNECT-GENERATE***	
10:40	**iPSC-based modeling of RASopathy-related cardiac hypertrophy reveals new mechanisms and therapeutic options** **George Kensah,** University Medical Center Göttingen - ***GeNeRARe***	
11:00	**Hereditary cholestatic syndroms in adults** **Verena Keitel-Anselmino,** University Hospital Magdeburg - ***HiChol***	
11:20	BREAK	

SESSION 8	PANEL European Reference Networks	Chair: Rebecca Schüle
11:45	**ERN-RND** **Holm Graessner,** University Hospital Tübingen	
11:55	**ERKNet** **Franz Schaefer,** Heidelberg University Hospital	
12:05	**ERN-RITA** **Nico Wulffraat,** University Medical Center (UMC) Utrecht	
12:15	**- Panel discussion -**	
12:45	CLOSING REMARKS AND FAREWELL	
13:00	END	

Annemieke Aartsma-Rus

Prof. Dr. Annemieke Aartsma-Rus is a professor of Translational Genetics at the Department of Human Genetics of the Leiden University Medical Center. She played an important role in the development of antisense mediated exon skipping for Duchenne muscular dystrophy. In 2020 she co-founded the Dutch Center for RNA Therapeutics (DCRT), a non-for-profit academic collaboration aiming to develop clinical treatment with exon skipping therapies for eligible patients with unique mutations. She has published over 210 peer-reviewed papers, 11 book chapters, 15 patents and has edited a book. Awards: Duchenne Award from the Dutch Duchenne Parent Project (2011), Black Pearl Science Award from EURORDIS (2020), Ammodo Science Award and the Lifetime achievement award (2021) from the Dutch Society of Gene and Cell Therapy for her work on antisense oligonucleotide mediated exon skipping.

Carsten Bergmann

Carsten Bergmann, M.D., is a clinical geneticist by training and Professor for Human Genetics. He is in charge of the Medizinische Genetik Mainz and manages the genetic activities of the Limbach group on a global scale. His research lab is located at the University of Freiburg where he holds a part-time faculty appointment. His group is interested in the translation of genetic findings into mechanistic studies to define new therapeutic targets and clinical treatment options. Key to success is the complementary approach of research and diagnostics with outstanding sequencing, clinical genetics and bioinformatics facilities combining the strengths of an academic and private diagnostic setting.

Christoph Bock

Christoph Bock is a Principal Investigator at the CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences and Professor of [Bio]Medical Informatics at the Medical University of Vienna. His research combines experimental biology (high-throughput sequencing, epigenetics, CRISPR screening, synthetic biology) with computational methods (bioinformatics, machine learning, artificial intelligence) – for cancer, immunology, and precision medicine. Before coming to Vienna, he was a postdoc at the Broad Institute of MIT and Harvard and a Ph.D. student at the Max Planck Institute for Informatics. Christoph Bock is also scientific coordinator of the Biomedical Sequencing Facility of CeMM and MedUni Vienna, and he coordinates an EU Horizon 2020 project that contributes single-cell sequencing of organoids to the Human Cell Atlas. Awards: Otto Hahn Medal of the Max Planck Society, ERC Starting and Consolidator grants, Overton Prize of the International Society for Computational Biology.

Peter Boor

Professor Peter Boor, M.D., Ph.D., is senior consultant pathologist, Heisenberg Professor and chair of Translational Nephropathology. He leads Electron Microscopy Facility and Digital Pathology at the University Clinic of RWTH Aachen, Germany, and is the RWTH Lecturer. He is a member of several national and international societies of pathology, renal pathology and nephrology and received several prestigious awards from these societies. His group, the LaBooratory of Nephropathology, focuses on diagnostic biomarkers, particularly imaging, digital pathology and AI, in vivo animal modeling, and chronic kidney diseases and fibrosis. He published >200 original papers, reviews and editorials, and several book chapters.

Hildegard Büning

Hildegard Büning is Professor of Infection Biology and Gene Transfer and Deputy Director of the Institute of Experimental Hematology at Hannover Medical School. She serves as President of the European Society of Gene and Cell Therapy and Scientific Secretary of the German Society for Gene Therapy. She studied Biology in Münster and Munich, and obtained her Ph.D. (Dr. rer. nat.) at the Ludwig-Maximilians-University Munich in 1997. Since then, she is active in the field of adeno-associated virus (AAV) vectors with a particular focus on AAV-host interactions and vector development. Aiming to optimize efficacy and safety of AAV vectors for gene therapy, she is developing strategies to modify vector tropism and immunogenicity using both rational design and library-based approaches.

Tobias Freilinger

Tobias Freilinger, M.D. - Education and clinical career: Medical studies at the Universities of Regensburg and Munich (TU). Residency in Neurology at Klinikum Großhadern, LMU, Munich (2002-2012). Consultant Neurologist at the Centre of Neurology, University of Tübingen (2013-2018); current position: Head of the Department of Neurology, Klinikum Passau, Germany. Research activities: Researcher at Molecular Neurogenetics and Institute of Stroke and Dementia Research, LMU Munich (2002-2013). Research group leader (since 2014) at Hertie Institute for Clinical Brain Research, Tübingen (Migraine and Primary Headache disorders). Research interests: genetics of migraine and other paroxysmal brain disorders, pathophysiology of hemiplegic migraine.

Christian Geis

Professor Christian Geis is a senior Neurologist, Schilling Professor, and Chair of Translational Neuroimmunology at Jena University Hospital, Germany. He is speaker of the Research Unit 'Synaptic pathology of autoimmune encephalitis' funded by the German Research Council and Board member of 'GENERATE' and the 'Center for Sepsis Control and Care'. His research focuses on basic autoimmune and inflammatory mechanisms in the brain, in particular on autoantibody-mediated synaptic pathomechanisms and on cognitive dysfunction after severe systemic inflammation. The research group has specific expertise in single-cell and network electrophysiology, super-resolution imaging techniques, computational analysis, molecular biology, and animal models of autoimmune and inflammatory disorders. Prof. Geis is PI of the randomized multicenter phase II trial GENERATE-BOOST.

Rutger Gjaltema

Rutger Gjaltema is an assistant professor of epigenetics at the Swammerdam Institute for Life Sciences at the University of Amsterdam. He earned his PhD with honors from the University of Groningen for his work on epigenetic editing in cancer cells and fibrotic fibroblasts using zinc finger proteins and the CRISPR/Cas9 platform. He also made significant contributions to the field of brittle bones disease. During his postdoc at the Max Planck Institute for Molecular Genetics in Berlin, he developed and applied (tunable) epigenetic editing tools to study the (cis) regulatory landscape of X-chromosome inactivation in early embryonic development. His recently established lab at the University of Amsterdam focuses on understanding the epigenetic crosstalk underlying transcriptional memory and variability. He is also involved in the interdisciplinary Epi-Guide-Edit consortium, which aims to unravel the (epi)genetic rules for robust and sustained epigenetic editing.

Holm Graessner

Holm Graessner has graduated in Biomedical Engineering, Electrical Engineering, German Language and Literature, Philosophy as well as Business Administration. He received his Ph.D. "Summa cum laude" in 2004 and his MBA degree in 2008. He is Managing Director of the Rare Disease Centre, since 2010, at the University and University Hospital Tübingen, Germany. He is Coordinator of the European Reference Network for Rare Neurological Diseases (ERN-RND). Together with Professor Olaf Riess, he coordinates the H2020 Solve-RD project "Solving the unsolved rare diseases". He was co-leading one of the four working groups of the German Action Plan for Rare Diseases from 2010 until 2013. Since 2020, as a fellow of the European Academy of Neurology (EAN) and in his function as the coordinator of ERN-RND, he is a personal member of the Rare Disease Coordinating Panel of the EAN.

Bodo Grimbacher

Prof. Dr. Bodo Grimbacher is a physician scientist and the Vice-Director of the Institute for Immunodeficiency at the Medical Center – University of Freiburg, Germany. Additionally, he is an honorary consultant for the Department of Immunology at the Royal Free Hospital, University College London, where until 2011, when he returned to work in Freiburg, he was a EU-Marie-Curie team leader. Previously, he was lecturer and senior scientist at the Department of Clinical Immunology, Freiburg. He completed his postdoc at the NIH, National Human Genome Institute. Amongst his awards and honors, he was granted the Emmy-Noether Fellowship of the Deutsche Forschungsgemeinschaft (DFG), the Marie-Curie Excellence Grant of the European Commission, the Georges Köhler Award of the German Immunology Society (DGfI), the Richard Farr Memorial Lectureship at AAAAI, the Rudolf-Schoen Prize from the Medizinische Hochschule Hannover, and the Thieme prize from the Leopoldina, the German Research Society.

Laurent Groc

Laurent Groc is Research Director for the CNRS at the Interdisciplinary Institute for Neurosciences (Bordeaux, France). After receiving his B.Sc. degree from the University of Toulouse (France), he earned his Ph.D. degree (2000) in neurobiology at Wayne State University (MI, USA) and University of Lyon (France), working in the Gossett laboratory of Robert Levine. He took a postdoctoral training from Bengt Gustafsson at the Department of Physiology in the University of Goteborg (Sweden). In 2009, he was nominated Research Director for the CNRS in Bordeaux and is currently heading a research group that investigates how developing synapses are formed in health and psychotic diseases. His lab uses a variety of methods (classical and single molecule imaging, electrophysiological, biochemical, molecular, and behavioral) to investigate the molecular pathways regulating the dynamic organization of synaptic proteins in physiopathological conditions.

Günter Harms

Dr. Günter Harms is Senior Market Access Director for Germany, Austria and Switzerland at Novartis Gene Therapies. He has over 25 years of international management experience in the pharmaceutical industry working at Ferring, Shire, Avexis, and Novartis in Germany, Switzerland and the USA. His areas of work included drug development, launch and life cycle management for global, regional and national markets, including Europe and also the USA. He has many years of experience in the areas of rare diseases and orphan drugs, including gene therapy. Günter studied Pharmacy and Biochemistry. Before joining the pharmaceutical industry he held a management role at the Institute for Public Health Systems Research and Development (IGSF), a consulting firm and WHO collaborating centre in Kiel.

Tobias B. Huber

Professor Tobias B. Huber is Chair of the Center of Internal Medicine and Director of the III. Department of Medicine (Nephrology, Rheumatology, Endocrinology and Transplantation) at the University Medical Center Hamburg-Eppendorf (UKE). Together with his team he is internationally recognized for discovering signalling pathways relevant for kidney development, filtration, maintenance and disease. He received numerous national and international recognitions including the Donald W. Seldin Young Investigator Award of the American Society of Nephrology and is elected member of the American Society of Clinical Investigation, the Association of American Physicians and the National Academy of Sciences – Leopoldina.

David T. W. Jones

David Jones is a biologist, who completed his Ph.D. at the University of Cambridge in 2009, before joining the German Cancer Research Center (DKFZ) in Heidelberg. Now a Division Head at the DKFZ and the Hopp Children's Cancer Center Heidelberg (KiTZ), his group uses cutting-edge genomics techniques for both basic and translational research in the field of pediatric neurooncology, and he has co-authored more than 250 publications on topics such as the identification of novel molecular tumor classes, pre-clinical modelling and the application of molecular diagnostics. David Jones was closely involved in the establishment of DNA methylation analysis as a robust tool for brain tumor classification, and is focused on translating the latest technologies to improve diagnostic accuracy and identify novel treatment approaches for pediatric cancer patients.

Ralf J. Jox

Ralf J. Jox M.D., Ph.D., is a neurologist and bioethicist working as Associate Professor of Medical Ethics at Lausanne University Hospital, Switzerland, affiliated to the Institute of Humanities in Medicine. He also directs the Clinical Ethics Unit of Lausanne University Hospital where he conducts on a regular basis, among others, retrospective ethics case deliberations with the Service of Genetic Medicine. His research activities focus on clinical ethics consultation, treatment decision making for neurological patients, research ethics concerning vulnerable populations, and the ethics of technology use in health care. He is an elected member of the National Advisory Committee on Biomedical Ethics in Switzerland.

Petra Kaufmann

Petra Kaufmann, M.D., is the Chief Medical Officer of Affinia Therapeutics, a gene therapy company using innovative technology to overcome the limitations of current AAV therapies. Prior to joining Affinia, she was Senior Vice President, Clinical Development, Translational Medicine & Analytics at Novartis Gene Therapies, following many years of clinical research in academia, and at the National Institutes of Health (NIH) where she most recently served as Director of the Office of Rare Diseases Research. She earned her M.D. at the University of Bonn, and her M.S. in Biostatistics at Columbia University where she also trained in neurology and served as tenured faculty member, advancing research and caring for patients. She continues to hold an adjunct appointment at Columbia University. She is also an adjunct Professor of Neurology at the University of Rochester.

Verena Keitel-Anselmino

Professor Verena Keitel-Anselmino, M.D., is Head of the Department of Gastroenterology, Hepatology and Infectious Diseases at Magdeburg University Hospital. She studied medicine at Heidelberg University and Imperial College, London (DAAD stipend). In her medical thesis at DKFZ, Heidelberg, she studied the molecular mechanisms of variants in the ABCC2 gene contributing to the rare Dubin-Johnson Syndrome. She received her specialist training in gastroenterology and her scientific training at Duesseldorf University Hospital from 2004 onwards. There, she started her own research group with a special interest in hepatobiliary transport and bile acid signalling and their relevance for cholestatic liver disease. These rare inherited cholestatic syndromes are the research focus of the HiChol consortium.

George Kensah

George Kensah, Dr. rer. nat., is Head of Experimental Research of the Clinic for Cardiothoracic and Vascular Surgery at the University Medical Center Göttingen, Germany. He is biologist by training and received his Ph.D. in the field of pluripotent stem cell-based myocardial tissue engineering from Hannover Medical School, Germany. Since 2013, he works on in vitro myocardial disease modelling using RASopathy patient-derived induced pluripotent stem cells (iPSCs). RASopathies are a family of rare diseases where heterozygous mutations in components of the Ras-MAPK pathway are associated with cardiac anomalies. George Kensah's lab employs iPSC-derived cardiomyocytes to study pathomechanisms underlying manifestations of RASopahy-associated mutations on single cells, or on the level of miniaturized 3D myocardium to identify potential targets for pharmaceutical phenotype rescue approaches.

Janbernd Kirschner

Professor Janbernd Kirschner is Director of the Department of Neuropaediatrics and Muscle Disorders at the Medical Center – University of Freiburg. After studying medicine (M.D.) at the Universities of Freiburg and Newcastle-upon-Tyne (UK), he completed his specialist training in paediatrics and adolescent medicine with a focus on neuropaediatrics at the University Hospital Freiburg. He performed a two years post-doc training with Prof. Carsten Bönnemann at the Children's Hospital of Philadelphia with a DFG research fellowship. After his return to Freiburg he received his habilitation and established a research group focusing on paediatric neuromuscular diseases and the development of innovative therapeutic approaches. From 2019 to 2022 he hold a position as director and professor for neuropaediatrics at the University Hospital Bonn, before accepting the call to a full professorship in Freiburg. He is now deputy chair of the Freiburg Centre for Rare Diseases (FZSE), executive board member of the European Reference Network for Neuromuscular Diseases (EURO-NMD) and principal investigator of the SMArtCARE network for the collection of real-world data in spinal muscular atrophy. Awards: Felix-Jerusalem Prize of the German Muscular Dystrophy Society and Research Prize of the German Duchenne Foundation.

Jan-Henning Klusmann

Professor Jan-Henning Klusmann is Director of the Department of Pediatrics and ERC Research group leader at the University Hospital Frankfurt, Goethe-University Frankfurt. He has been focusing on molecular and translational studies on acute myeloid leukemia especially in infants. Using one of the largest collections of pediatric AML specimens worldwide, his group combines state-of-the-art omics profiling (including whole genome sequencing, global mRNA and ncRNA expression profiling) with functional analyses and disease modeling in primary human and murine cells. He is International Coordinator of the ML-DS 2018 trial.

Stefan Lange

Stefan Lange completed his medical studies at the Heinrich-Heine-University in Düsseldorf in 1989 and received his M.D. in 1994. From 1989-1993 he was initially in practical training at the Ferdinand-Sauerbruch-Clinic in Wuppertal, then assumed the position of intern/resident physician. In 1993 he joined the department of medical biometry at the Ruhr-University in Bochum and was appointed to the position of research assistant in 1995. In 2003 he received his Habilitation (qualification for a professorship) and venia legendi (award of title of Privatdozent) in Medical Biometry and Clinical Epidemiology. He joined the Institute for Quality and Efficiency in Health Care in 2004, and has held the position of Deputy Director of the institute since 2005.

Soeren Lienkamp

Soeren Lienkamp studied Medicine at the University of Freiburg (M.D.), Germany. He interrupted his studies for a research student fellowship at the University of Nagoya, Japan. After a post-doc at the Renal Division of the University Hospital Freiburg, he established his own group funded by a prestigious Emmy-Noether grant from the German research association and completed his residency training for Internal Medicine. In 2019 he became an Assistant Professor at the Institute of Anatomy of the University of Zurich. His research interests include inherited kidney diseases and cell fate determination during renal development using the Xenopus model. His group established a method to directly reprogram fibroblasts into renal tubule cells, a project that is currently supported by an ERC starting grant.

Michal Minczuk

Michal Minczuk is a MRC Investigator in the MRC Mitochondrial Biology Unit (MBU) at the University of Cambridge, leading a research programme in mitochondrial genetics. His programme encompasses the development of methods for controlled editing of the mammalian mitochondrial genome, mechanistic studies of mitochondrial gene maintenance and expression in health and disease, and the development of advanced gene therapies for mtDNA dysfunction. Michal obtained a PhD from the University of Warsaw (2003). From 2004–2007 he was an FEBS Postdoctoral Fellow in the group of Prof. Sir Aaron Klug at MRC Laboratory of Molecular Biology, Cambridge. Michal joined the MRC MBU in 2007, later he became a tenure-track MRC Investigator, with tenure in 2015. In 2019, Michal co-founded Pretzel Therapeutics, a start-up biotechnology company that focuses on the development of therapies to treat unmet needs in diseases driven by mitochondrial dysfunction.

Paul A. Northcott

Paul A. Northcott, Ph.D., is an Associate Member in the Department of Developmental Neurobiology at St. Jude Children's Research Hospital. During his Ph.D. and postdoctoral training, he made numerous important contributions to the medulloblastoma literature, including the description of molecularly and clinically distinct consensus subgroups, comprehensive annotation of genomic and epigenomic landscapes, and the discovery of 'enhancer hijacking' as a novel mechanism of oncogene activation in brain tumors. He is an active Member of the Children's Oncology Group, the American Association of Cancer Research, and the Society for Neuro-Oncology, and serves on the Scientific Advisory Board for The Brain Tumor Charity (UK). He has led several high-impact studies published in Nature, Nature Genetics, Lancet Oncology, and the Journal of Clinical Oncology, disclosing the molecular mechanisms underlying medulloblastoma pathogenesis through high-resolution genomics. Awards/Grants: V Foundation V Scholar Award, Sontag Foundation Distinguished Scientist Award, Pew-Stewart Scholar for Cancer Research, St. Baldrick's Foundation Robert J. Arceci Innovation Award, AACR NextGen Grant for Transformative Cancer Research.

Stephan Ossowski

Stephan Ossowski, M.D., is Professor of Genome Analytics and Head of the Computational Genomics Group at the Faculty of Medicine in Tübingen. His group develops methods for biomedical genomics with a focus on applications of next generation and nanopore sequencing in personalised medicine. Research foci of the Ossowski Lab are rare genetic diseases, the evolution of tumours and the spread of antibiotic resistance genes in hospitals. As Head of the Diagnostic Bioinformatics Group at the University Hospital Tübingen and member of the Centre for Personalised Medicine, Stephan Ossowski translates new diagnostic methods such as genome, exome and transcriptome sequencing or liquid biopsy into clinical practice.

Matthew Porteus

Matthew Porteus, M.D., Ph.D., is the Sutardja Chuk Professor of Definitive and Curative Medicine and a Professor in the Department of Pediatrics, Institute of Stem Cell Biology and Regenerative Medicine and Maternal-Child Health Research Institute at Stanford. His primary research focus is on developing genome editing as an approach to cure disease, particularly those of the blood but also of other organ systems as well. He received his undergraduate degree at Harvard in History and Science where his honors thesis studied the recombinant DNA controversy of the 1970s. He then completed his M.D. and Ph.D. training at Stanford, clinical training in Pediatric Hematology/Oncology at Boston Children's Hospital, and post-doctoral research training with Noble Laureate David Baltimore at CalTech. He works as an attending physician on the Pediatric Hematopoietic Stem Cell Transplant service at Lucile Packard Children's Hospital. His goal is to combine his research and clinical interests to develop innovative curative therapies. He served on the 2017 National Academy Study Committee of Human Genome Editing and serves on the Scientific Advisory Board for WADA on Cell and Gene Doping and the NIH NexTRAC advisory committee evaluating the emergence of new technologies.

Peter Robinson

Peter Robinson, M.D., M.Sc., studied Mathematics and Computer Science at Columbia University and Medicine at the University of Pennsylvania. He completed training as a Pediatrician at the Charité University Hospital in Berlin, Germany. His group developed the Human Phenotype Ontology (HPO), which is now an international standard for computation over human disease that is used by the Sanger Institute, several NIH-funded groups including the Undiagnosed Diseases Program, Genome Canada, the rare diseases section of the UK's 100,000 Genomes Project, and many others. The group develops algorithms and software for the analysis of exome and genome sequences and has used whole-exome sequencing and other methods to identify a number of novel disease genes, including CA8, PIGV, PIGO, PGAP3, IL-21R, PIGT, and PGAP2.

Franz Schaefer

Franz Schaefer is Professor of Pediatrics and Chief of the Pediatric Nephrology Division at Heidelberg University Hospital. He received his M.D. at Würzburg University Medical School. He performed research scholarships at the Institute of Child Health, London, University of Virgina and Stanford University. He served on boards and councils of numerous professional societies and is president-elect of the International Pediatric Nephrology Association. His research interests include the genetic basis of kidney diseases, the prevention of kidney disease progression, the cardiovascular, metabolic and endocrine consequences of kidney disease in children, and the management of end-stage kidney disease by renal replacement therapy. He conducted numerous clinical trials and established the European Study Consortium for CKD in Children (ESCAPE), the International Pediatric Dialysis Network (IPDN), and the IPNA Global Registry for Pediatric Kidney Replacement. He also coordinates the European Rare Kidney Disease Reference Network (ERKNet) and Registry (ERKReg) and the PodoNet SRNS Registry, chairs the Global Registry for atypical HUS, and co-chairs the European Joint Programme on Rare Diseases (EJP-RD). His publication record encompasses more than 600 scientific articles and book chapters and the international textbooks 'Pediatric Kidney Disease' and 'Pediatric Dialysis'.

Dmitrii Smirnov

Dmitrii (Dima) Smirnov is a researcher in the group of Dr Holger Prokisch at the Klinikum rechts der Isar of the Technical University of Munich, where he focuses on the integration of multi-omics data for the diagnosis of Mendelian diseases. He is particularly interested in the implementation of machine learning algorithms to integrate genetic, phenotypic, and functional OMICs data for the automation of molecular diagnostic procedures in rare diseases and the identification of novel biomarkers for mitochondrial conditions. His recent contributions include implementation of proteomics into molecular diagnosis workflow and development of clinical guidelines for RNA sequencing data interpretation as part of the ACMG/AMP framework. Dima recently received an early career award from the European Society of Human Genetics for his work in this area.

Eva Stumpe

Eva Stumpe is a longtime patient advocate engaged in the field of spinal muscular atrophy. She is a contact person at DGM e.V., a board member of Initiative SMA and treasurer and board member of SMA Europe e.V. She is mother of 2 adult kids. Her 27-year old daughter is living with spinal muscular atrophy type II. Eva Stumpe is an alumni of the EUPATI Patient Expert Training Course on R&D in medicines and of the EURORDIS summer and winter school on R&D and translational medicine. On a professional level, she works as a lawyer, supports her husband in the family business and is member of a local housing board.

Henkjan J. Verkade

Professor Henkjan J. Verkade, M.D, Ph.D, is a pediatric gastro/hepatologist at the Beatrix Children's Hospital of the University Medical Center Groningen. He received his Ph.D. in Medicine at the University of Groningen on the thesis entitled „Lipid absorption and metabolism". He was a post-doctoral fellow at the University of Alberta, Edmonton, Canada. Henkjan Verkade combines clinical work in pediatric gastro/hepatology with clinical and fundamental research projects. He is a member of national and international professional associations and organisations. For the European Society Pediatric Gastroenterology Hepatology and Nutrition (ESPGHAN), he served as Chair of the Hepatology Committee and member of the Council (2016-2020). His current research projects involve intestinal lipid absorption and metabolism, the enterohepatic circulation, and pediatric liver disease. He has authored more than 250 peer-reviewed publications and more than 15 book chapters. Since 2019, he is associate editor of the Journal of Pediatric Gastroenterology and Nutrition. He has supervised 45 Ph.D. students.

Nico Wulffraat

Nico Wulffraat is full professor of pediatric Rheumatology at the department of pediatrics, University Medical Center, Utrecht. He is involved in several research fields, including vaccination in paediatric rheumatologic diseases, methotrexate treatment in children with juvenile idiopathic arthritis, biologic treatment in paediatric rheumatologic diseases, stem cell therapy in juvenile idiopathic arthritis and international paediatric rheumatology trials with PRINTO. Over the years he gained much experience with large multicenter international research projects. One of his current biggest projects as principal investigator is an ongoing study of an EU funded pharmacovigilance project in which juvenile patients treated with biologicals are followed for over 10 years in more than 50 countries. He leads the EU project SHARE on the network for paediatric rheumatology centers and recommendations for diagnosis and therapy. He was principal investigator of an international study for evaluation of behavioural therapy for treatment of methotrexate intolerance in juvenile idiopathic arthritis patients completed in 2014. Since 2017 he is co-PI together with Professor R. Yeung of a multicentre precision medicine research grant (UCAN-CANDU) in which all Dutch and Canadian pediatric rheumatology centers collaborate.

RESEARCH POSTERS

Back to schedule

1. **Rare variant co-occurrence in gnomAD – A resource to aid clinical variant interpretation for recessive disease**
   Sarah Stenton, Broad Institute of MIT and Harvard, Cambridge

2. **RIOK1 - a novel gene associated with Diamond Blackfan anemia**
   Sheila Bohler, Medical Center - University of Freiburg - *MyPred*

3. **Patient-Specific Hepatobiliary Organoids Elucidate Functional Alterations in Intrahepatic Cholestasis**
   Tobias Cantz, Hannover Medical School (MHH) - *HiChol*

4. **Skeletal muscle transcriptomics dissects the pathogenesis of Friedreich´s Ataxia**
   Silvia Boesch, Medical University of Innsbruck - *TreatHSP.net (associated partner)*

5. **Systemic and intracellular iron starvation response in Friedreich´s Ataxia**
   Elisabetta Indelicato, Medical University of Innsbruck - *TreatHSP.net (associated partner)*

6. **Compassionate use of VAL-1221 i.v., a Fab/rhGAA fusion protein, in a Lafora disease patients**
   David Brenner, Ulm University

7. **UCHL1 missense and loss-of-function variants cause autosomal dominant optic atrophy with a shift in glycolytic homeostasis**
   Claudio Fiorini, IRCCS Istituto delle Scienze Neurologiche di Bologna

8. **Allogeneic BK virus-specific T cell therapy in patients with progressive multifocal leukoencephalopathy (CurePML)**
   Günter Höglinger, LMU Klinikum, Munich

9. **Systematic evaluation of olfaction in patients with renal ciliopathies**
   Mareike Dahmer-Heath, University Hospital Münster - *NEOCYST*

10. **Ultrastructural characterization of human motile cilia in renal ciliopathies**
    Mareike Dahmer-Heath, University Hospital Münster - *NEOCYST*

11. **Refining kidney survival in 383 genetically characterized patients with nephronophthisis – conclusions from the NEOCYST clinical registry**
    Jens König, University Hospital Münster - *NEOCYST*

12. Towards human retinal organoids as a test model for virus mediated gene therapy
Teresa Rogler, Ludwig-Maximilians-Universität (LMU), Munich

13. The German Human Genome-Phenome Archive (GHGA) – A national infrastructure for secure archival and community-driven analysis of omics data
Janika Kiltz, University Hospital Tübingen, GHGA

14. Comprehensive, Orchestrated, National Network to Explain, Categorize and Treat autoimmune encephalitis and allied diseases within the GErman NEtwork for Research on AuToimmune Encephalitis
Frank Leypoldt, University Hospital Schleswig-Holstein, Kiel - *CONNECT-GENERATE*

15. HiChol: Hereditary Intrahepatic Cholestasis Translational Network
Verena Keitel-Anselmino, University Hospital Magdeburg - *HiChol*

16. German network for mitochondrial diseases (mitoNET)
Boriana Büchner, LMU Klinikum, Munich - *mitoNET*

17. MyPred - Optimizing care for young individuals with syndromes predisposing to myeloid malignancies
Ursula Kern, Medical Center - University of Freiburg - *My Pred*

18. The NEOCYST Network of Early Onset Cystic Kidney Disease
Jens König, University Hospital Münster - *NEOCYST*

19. Speed Translation-Oriented Progress to Treat FSGS
Sybille Köhler & Maja Lindenmeyer, University Medical Center Hamburg-Eppendorf - *STOP-FSGS*

20. Treat-ION: New Therapies for Neurological Ion Channel and Transporter Disorders
Holger Lerche, University Hospital Tübingen - *Treat-ION*

21. Research for Rare – German Networks on Rare Diseases
Katja Franke-Rupp & Corinna Schultheis, LMU Klinikum, Munich - *Research for Rare*

Thomas Klopstock

LMU Klinikum, Munich
*Research network **mitoNET** - Mitochondrial diseases*

Jens König

University Hospital Münster
*Research network **NEOCYST** - Hereditary cystic kidney diseases*

Christian Kratz

Hannover Medical School
*Reseach network **ADDRess** - Disorders with abnormal DNA damage response*

Holger Lerche

University Hospital Tübingen
*Research network **Treat-ION** - Neurological ion channel and transporter disorders*

Frank Leypoldt

University Hospital Schleswig-Holstein, Kiel
*Research network **CONNECT-GENERATE** - Autoimmune encephalitis*

Maja Lindenmeyer

University Medical Center Hamburg-Eppendorf
*Research network **STOP-FSGS** - Focal segmental glomerulosclerosis*

Wolfgang Müller-Felber

LMU Klinikum, Munich
*Research network **MD-Net** - Hereditary neuromuscular diseases*

Rebecca Schüle

Heidelberg University Hospital
*Research network **TreatHSP.net** - Hereditary spastic paraplegias*

Maggie Walter

LMU Klinikum, Munich
*Research network **MD-Net** - Hereditary neuromuscular diseases*

RESEARCH FOR RARE
German Networks on Rare Diseases

Since 2003, the Federal Ministry of Education and Research (BMBF) has been funding research for rare diseases by supporting national networks in their efforts to understand the underlying causes of rare diseases and to develop new therapeutic approaches. The networks bring together excellent scientific and clinical capacities in order to create best conditions for systematic basic research and its translation into patient care.

The cooperation of specialists, located nationwide at German university hospitals, overcomes the hurdles for medical research caused by small patient groups and expertise spreaded over different locations. Networking activities enable a structured and coordinated compilation of data as well as evaluation of study results and new findings. The joint effort is intended first and foremost to help patients achieve a more timely and exact diagnosis and to continue developing the best personalized treatments.

For the funding period 2019-2022/2023, 11 research networks have been supported, and 9 of them entered into the current funding period 2023-2026. Together with the consortia financed in previous years, the BMBF has been investing 165.5 million Euro in this very successful programme.

A board of speakers serves as decision-making and coordinating body. Elected spokespersons are Prof. Dr. Rebecca Schüle, principal investigator of the TreatHSP.net and Prof. Dr. Thomas Klopstock, principal investigator of the mitoNET research network. A coordinating office, located in Munich at the Friedrich-Baur Institute at the Department of Neurology, University Hospital, LMU Munich, assists the board of speakers to manage joint tasks and to foster collaboration on national and international level.

For further information, please visit the website: www.research4rare.de

ADDRess

A common characteristic of patients with Disorders with Abnormal DNA Damage Response (DADDR) is a highly increased cancer risk. The scientific projects of the research network **ADDRess (Translational Research for Persons with Abnormal DNA Damage Response)** join forces to improve medical and psychosocial care, early cancer detection, cancer diagnosis and therapy for people with impaired DNA repair. The research is fascilitated by the cancer predisposition syndrome (CPS) registry.

www.krebs-praedisposition.de/register/address

CONNECT-GENERATE

The established German Network for Research on Autoimmune Encephalitis (GENERATE) contributes significantly to the national networking of therapists, researchers and patients and offers a server-based registry and biomaterials. The research network **CONNECT-GENERATE, for the explanation, categorisation and treatment of autoimmune encephalitis and related diseases**, is part of this network and aims at improving the diagnosis and therapy for patients with autoimmune encephalitis, a rare but treatable form of brain inflammation. The scientific projects focus on genetic causes, imaging techniques as well as immunological and neuronal network mechanisms. In addition, the efficacy of plasma cell-targeted therapeutics is being investigated in a clinical trial (GENERATE-BOOST). The optimisation and expansion of the patient registry is another important activity.

www.generate-net.de/connect-generate.html

GAIN

The research efforts of the **German genetic multi-organ Auto-Immunity Network (GAIN)** concentrate on "ultra-rare" multi-organ autoimmune diseases. Scientific projects focus on the underlying molecular and cellular pathomechanisms as well as possible molecular interventions as a therapeutic option. Another focus is on the development of a uniform approach to the identification, diagnosis and treatment of multi-organ autoimmune diseases. A biobank and patient registry have been established. Through this registry, patients could be recruited for a clinical trial investigating the safety and efficacy of the immune-modulatory drug Abatacept. The quality of life of patients will be investigated through an epidemiological study with patient involvement.

www.g-a-i-n.de

GeNeRARe

The objective of the scientific projects of the **German Network of RASopathy Research (GeNeRARe)** is to increase knowledge on molecular pathogenesis and pathophysiology of RASopathies in order to explore novel clues for a causal treatment. Previous work led to the development of a database to better assess the effects of certain gene mutations. New genes for RASopathies have been identified and special studies have contributed to a better understanding of the causes and effects of the gene mutations. Further goals are the establishment of a collection of standardized clinical data from RASopathy patients, the analysis of genotype phenotype correlations, and the development of innovative diagnostics.

www.generare.de

HiChol

The research network **HiChol** focuses on the genetic defects in patients with severe **hereditary intrahepatic cholestasis**, a disease caused by disrupted bile flow and resulting in progressive liver damage. The aim of the consortium is to better understand these diseases and, in the long term, to provide patients with better predictions of the disease progression and tailored treatment options. The scientific projects analyse the biological consequences of novel mutations in silico and in organoid models and include the establishment of a patient registry allowing investigating genotype-phenotype relationships. Results should provide new insights regarding effective therapies.

www.hichol.hhu.de

mitoNET

The **German Network for Mitochondrial Diseases (mitoNET)** is dedicated to the heterogeneous group of mitochondrial diseases. The mitoNET builds on a horizontal network of clinical experts in pediatric and neurological university hospitals who enroll patients into a comprehensive registry and, by regular follow-up visits, into a natural history study. DNA, RNA and serum is collected in a central biobank. The basic science experts in the network work inter alia on genomics, transcriptomics, proteomics and complexome profiling. A current project explores the benefit of wearables to assess exercise intolerance in the real life setting. Since the mitoNET has been funded by BMBF for the maximum of 3 periods, its sustainability has been secured by different measures. Most importantly, mitoNET structures have been rolled out to an international level via the EU-funded GENOMIT project including the setup of a global mitochondrial registry. Of note, mitoNET/GENOMIT sites provide excellent infrastructure for clinical studies, and are accordingly involved in most of the current and upcoming therapy trials in mitochondrial diseases.

www.mitonet.org * www.genomit.eu

MyPred

The research network **MyPred** cares for young individuals with **syndromes predisposing to myeloid malignancies,** including myelodysplastic syndromes, myeloid leukemias and other myeloproliferative diseases. The consortium's overall goal is to understand specific genetic alterations that lead to myeloid neoplasms. This includes the description of still unknown syndromes that predispose to myeloid neoplasms. The research results will be used to help patients by establishing and improving early detection and treatment strategies in a targeted manner. One focus is on the comparison of different aspects of carcinogenesis and the treatment results of individual therapeutic measures in the different syndromes.

www.mypred.de

NEOCYST

Hereditary cystic kidney diseases are among the most important causes of chronic renal failure in childhood. The goals of the **Network for Early Onset Cystic Kidney Disease (NEOCYST)** are to increase the knowledge on the epidemiology, the phenotypical spectrum, the molecular background and the clinical prognoses of the individual disease entities. Based on an online patient registry and a central biobank, several scientific studies have been performed and allowed new insights into clinical and molecular details as well as the identification of a previously unknown genetic disease entity. Further work focuses on the prediction of individual disease courses and the identification and development of therapeutic approaches improving the clinical management and personal counselling of patients.

www.neocyst.de

STOP-FSGS

The research network **STOP-FSGS (Speed Translation-Oriented Progress to Treat FSGS)** concentrates on the idiopathic focal segmental glomerulosclerosis (FSGS). The focus is on understanding the molecular basis and developing new diagnostic and therapeutic approaches to treat FSGS. Previous work has helped to reveal underlying mechanisms of the disease by identifying new disease-causing genes, biomarkers and signaling pathways. The goal of the network is to combine the identified disease mechanisms, biomarkers and therapeutic approaches, develop them further and translate them into improved diagnostics and therapy for patients. In addition, STOP-FSGS will provide a central information, contact and exchange platform for FSGS for physicians, scientists and patients in Germany.

www.research4rare.de/forschungsverbuende/stop-fsgs/

TreatHSP.net

The research network **Treat-HSP.net (Translational Research in Hereditary Spastic Paraplegias)** emphasises on hereditary spastic paraplegias (HSP), which are highly heterogeneous neurodegenerative disorders. The network concentrates its translation-oriented research approach on pathophysiological key pathways of HSP. To make substantial progress towards implementation of novel therapies, TreatHSP.net generates a shared infrastructure that provides access to clinical data, biological samples and OMICS data. Scientific projects develop and validate outcome parameters for clinical trials, identify shared pathways and novel therapeutic targets and prioritise drug-repurposing strategies to evaluate novel therapeutic approaches in preclinical trials.

www.treathsp.net

Treat-ION

The research network **Treat-ION** specialises on **rare neurological ion channel and transporter disorders**. The focus of the network is on improving diagnostics and establishing individualised therapeutic treatment options for these diseases. Scientific projects concentrate on molecular and cellular pathophysiology, the effects of mutations on channel functions and the mechanisms of specific drugs. In explorative n-of-1 studies, research results in clinical subprojects are to be transferred into practice. A translational process accompanies the therapeutic use, in particular of already licensed and available drugs (drug repurposing). An established registry on ion channel diseases supports the work and results will be delivered to patients through a structured molecular therapeutic board.

www.treat-ion.de

Imprint

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