Young Researchers Workshop on Rare Diseases 2025

24th - 25th March 2025

Kardinal-Wendel-Haus

Kath. Akademie in Bayern

Mandlstr. 23, 80802 München

Schloss Suresnes, © Kath. Akademie in Bayern

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Bundesministerium für Bildung und Forschung

The **Young Researchers Workshop on Rare Diseases 2025** gives young researchers and clinicians in the field of rare diseases the opportunity to learn from experienced colleagues and to put their own scientific work into a broader context. Main topics will be in the field of innovative genetics and therapy, bioinformatics, and research ethics. The seminar will be **complemented by a workshop on scientific writing (26th March)**, which can be chosen additionally.

Participants:

The Young Researchers Workshop is organised for members of the "Research for Rare" (BMBF) network. Scientists not belonging to the above-mentioned networks can participate upon request.
Participants are asked to hand in a poster abstract and present the poster during the seminar. From the submitted abstracts, participants will be selected for short talks. The workshop will be held in English.

Registration:

Please register with a short bio under research4rare.de/young-researchers-workshop/.
The workshop is free of charge. Registration needs to be confirmed. Rooms are reserved in the academy until 20th January 2025 and have to be paid by the participants (approx. 110€/night).

CME credits will be applied for.

About us:

Research for Rare - Translational research on rare diseases
Many of the approximately 8000 rare diseases are still poorly understood. For this reason, research projects in the field of rare diseases are working together at many different locations in Germany and with international partners to jointly improve diagnostics and develop new therapies. The aim is to translate promising scientific findings into clinical application in order to improve the situation for people with rare diseases. The "Research for Rare" networks are funded by the Federal Ministry of Education and Research (BMBF). The networks are represented by their chairs Prof. Rebecca Schüle, Heidelberg University Hospital (speaker) and Prof. Thomas Klopstock, LMU Klinikum München (co-speaker).

The workshop is organised by a team of young researchers involved in the research networks: Midhuna Joseph Maran, **TreatHSP.net** (Saarland University), Tabita Ghete, **MyPred** (University Hospital Erlangen), and Dr. Justina Dargvaniene, **CONNECT-GENERATE** (University Hospital Schleswig-Holstein, Kiel). They are supported by the co-ordinating office to the research networks on rare disease.

Contact:

Research for Rare
Co-ordinating office
LMU Klinikum, Friedrich-Baur-Institut an der Neurologischen Klinik, Ziemssenstraße 1a, 80336 München
Responsible: Prof. Dr. Thomas Klopstock
Management: Katja Franke-Rupp and Dr. Corinna Schultheis

Scientific Advisors:
Prof. Dr. Rebecca Schüle, Heidelberg University Hospital (TreatHSP.net)
PD Dr. Frank Leypoldt, University Hospital Schleswig-Holstein, Kiel (CONNECT-GENERATE)

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Monday, 24th March 2025

11:15 – 11:30	Welcome and Introduction
11:30 – 12:00	**Unravelling the genetic basis of rare diseases. Challenges and strategies in neurogenetics** Prof. Dr. med. Rebecca Schüle - Heidelberg University Hospital
12:00 – 12:30	**Unchartered territory: How technology and informatics might transform diagnostics and therapy. Examples from rare autoimmune diseases** PD Dr. med. Frank Leypoldt - University Hospital Schleswig-Holstein
12:30 – 13:30	Lunch
13:30 – 14:00	**Current strategies in RNA-seq for Rare Diseases** Dr. Vicente Yépez - Technical University of Munich
14:00 – 15:30	**Ethics in Research and Medicine** Dr. phil. Christoph Schickhardt - Heidelberg University Hospital
15:30 – 17:00	**Poster Session** and Coffee
17:00 – 18:30	**Short presentations from participants**
19:00	Dinner

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Tuesday, 25th March 2025

7:00 – 8:30	Breakfast
8:30 – 10:00	**CNV/SV Detection and Exome data analysis** German Demidov, PhD - University Hospital Tübingen
10:00 – 10:15	Coffee Break
10:15 – 12:15	**Bioinformatics Workshop** German Demidov, PhD / Dr. Vicente Yépez
12:15 – 13:15	Lunch
13:15 – 14:15	**Panel Q&A: How to PhD?** Prof. Dr. med. Rebecca Schüle / PD Dr. med. Frank Leypoldt / German Demidov, PhD
14:15 – 15:30	**How to review and be reviewed?** Thomas Lemberger, PhD - European Molecular Biology Organization (EMBO) / Review Commons, Heidelberg
15:30 – 15:45	Coffee Break
15:45 – 16:45	**Good Data Management Practices** Anandhi Iyappan, PhD - European Molecular Biology Laboratory (EMBL), Heidelberg; German Human Genome-Phenome Archive (GHGA)
16:45	Closing remarks
17:00	End

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Optional workshop for a limited number of participants (max. 12 people):

Wednesday, 26th March 2025

7:00 – 8:00	Breakfast
8:30 – 17:00	**Scientific Writing Workshop** Dr. Avril Arthur-Goettig, Munich Venue: Friedrich-Baur-Institute, LMU Klinikum, Ziemssenstraße 1a, 80336 Munich

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SPEAKERS

Prof. Dr. med. Rebecca Schüle

Rebecca Schüle is an Adult Neurologist and Neuroscientist with special expertise in neurodegenerative motoneuron diseases and movement disorders. She serves as the Director of the Division of Neurodegenerative Diseases at the Heidelberg University Hospital and is Full Professor at the Ruprecht Karl University of Heidelberg. As a clinician-researcher the objective of her research is to advance novel treatments and cures for rare and ultrarare hereditary movement disorders.

Prof. Dr. Schüle received her M.D. from Ruprecht Karl University in Heidelberg. After graduating, she trained in Neurology and Movement disorders at the University of Tübingen and intensified her research into genomics of rare movement disorders at the John P. Hussman Institute for Human Genomics in Miami, Florida. Upon her return to the University of Tübingen, she served as Residency Program Director and Clinical Practice Director at the Center for Neurology and, as senior faculty, took over the lead of the research group 'Functional Genomics of Rare Movement Disorders' at the Hertie Institute for Clinical Brain Research.

Rebecca Schüle has built a translational research program for ultrarare diseases with a particular focus on Hereditary Spastic Paraplegias (HSP) and Ataxias. She also leads international networks for these diseases and is the coordinator of national research network TreatHSP.net. In recent years she has pioneered development and application of tailored RNA therapies for ultrarare diseases as a founding member of the 1 Mutation 1 Medicine network.

PD Dr. med. Frank Leypoldt

Frank Leypoldt is a clinical neuroimmunologist with board specialization in neurology and laboratory medicine. He is head of the neuroimmunology section at the University Hospital Schleswig-Holstein, Campus Kiel (UKSH) and a member of the faculty at the Christian-Albrechts-University Kiel. He was trained in Hamburg, Bern, and Toronto and worked as a research fellow in Josep Dalmau's lab (IDIBAPS, Barcelona).

PD Dr. Leypoldt is a board member and organizer of the German Network for Research on Autoimmune Encephalitis GENERATE e.V. Since 2019, he has been the spokesperson for the national research network CONNECT-GENERATE, which is funded by the German Federal Ministry of Education and Research. The physician and scientist has given over 180 national and international teaching and training lectures, published more than 130 scientific papers, is a member of various professional societies, part of the editorial board of the renowned neuroimmunology journal Neurology N2 and he is involved in the development of national and international guidelines.

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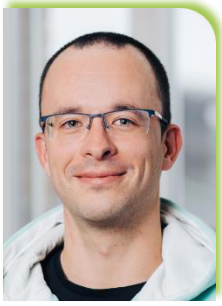

German Demidov, PhD

German Demidov (Diploma in Maths, 2013, Moscow State University; MSc in Algorithmic Bioinformatics, 2015, Saint Petersburg Academic University; PhD in Biomedicine, Universitat Pompeu Fabra in Barcelona) is a bioinformatics postdoc at the Institut für Medizinische Genetik und angewandte Genomik, Tübingen.

Previously, his main specialization was bioinformatics tools development, namely structural variants callers, but currently he concentrates on a task of diagnostics of undiagnosed rare disease patients. He worked in large scientific consortia such as PanCancer Analysis of Whole Genomes and Solve-RD.

Dr. Vicente Yépez

Vicente Yépez is a researcher and Scientific Manager of the Chair of Computational Molecular Medicine at the Technical University of Munich (TUM). He was the co-lead of the European Solve-RD RNA-seq analysis working group and is now part of the Multi omics group in ERDERA.

Dr. Yépez did his bachelor in Industrial Engineering in Mexico and his MSc in Mathematical Modelling in Engineering in Italy, followed by a PhD in Bioinformatics at the TUM where he is currently based. His main expertise is the analysis of RNA-seq data and its interpretation in combination with DNA and phenotype. He is also actively pushing for open source software and promoting the Solvathon concept.

Dr. phil. Christoph Schickhardt

Christoph Schickhardt is a Post Doc and Senior Scientist in the Department of Translational Medical Ethics at the National Center for Tumor Diseases (NCT), University Hospital Heidelberg, and the German Cancer Research Center (DKFZ) in Heidelberg (since 2013).

One of his research focuses is ethics in biomedical research (e.g. ethics of genomics, data sharing, secondary research use of health data) and in general the digitalization of medicine and governance of biomedical research. Further research focuses include public health ethics and child ethics.

As an ethicist or ethical advisor, Dr. Schickhardt is involved in several large biomedical research consortia and biotechnological initiatives. Christoph Schickhardt studied philosophy and was awarded a PhD degree in philosophy for a dissertation on child ethics by the University of Düsseldorf in 2011. Since then, he has been teaching courses in philosophy and ethics, particularly in applied ethics and medical ethics at the University of Heidelberg.

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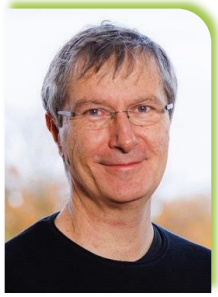
Anandhi Iyappan, PhD

Anandhi Iyappan started her education as a Microbiologist and later pursued master's in Bioinformatics. She further specialized in developing computational models for neurodegenerative diseases (PhD). She particularly enjoys working with structured interoperable data models and provide easy-to-use solutions for biologists and bioinformaticians.

Furthermore, she has worked as a science/project manager where she managed multiple projects relevant to Artificial intelligence and Machine Learning as well as involved in community outreach events, organizing mid and large-scale conferences and workshops. Apart from that, she has also experience in organizing and establishing scientific outreach activities for strengthening the scientific community.

Within GHGA, Anandhi Iyappan plays the role of metadata workstream coordinator, where she is in-charge of data harmonization, standardization, and generation of data by abiding to standardised principles and policies.



Thomas Lemberger, PhD

Thomas Lemberger is Head of Open Science Implementation and Deputy Head of Scientific Publications at EMBO in Heidelberg. Trained as a molecular biologist, Thomas received his PhD at the University of Lausanne, Switzerland, and did his postdoc at the German Cancer Research Center in Heidelberg.

He joined EMBO as a scientific editor to launch the open access journal Molecular Systems Biology. He was Chief Editor of the journal for more than ten years. Thomas is leading the development of the preprint peer-review platform Review Commons and is responsible for data and technology initiatives at EMBO Press.

Dr. Avril Arthur-Goettig

Avril Arthur-Goettig was born in Scotland and graduated from Edinburgh University with a BSc (hons) in Molecular Biology. She gained a PhD in Molecular Genetics from Sussex University, UK, followed by a postdoctoral fellowship at Glasgow University, UK. After three months as a visiting scientist at Dalhousie University, Halifax, Nova Scotia, Canada, she joined the Department of Biochemistry at the LMU Munich moving on to the Munich Gene Center with her own funding to lead a small research group.

In 1998, Dr. Arthur-Goettig became a freelance writer, editor and translator of scientific texts for research institutes (research papers, funding proposals), biotech startups, clinical agencies, and biotech/pharmaceutical companies, including 10 years of writing articles for Bayer. Later she decided to consolidate her experience and expertise into workshop training in scientific writing and communication for young researchers to help them optimize their writing and presentation skills.

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