34th Annual Meeting of the German Society of Human Genetics in Kassel, Germany from March 15–17, 2023 (Subject to change)

Kassel 2023
34. gfh-Jahrestagung

Wednesday, March 15, 2023

12:00–01:00 PM	Blauer Saal **Technical Workshop** **Life & Brain GmbH**
12:00–01:00 PM	Kolonnadensaal 2+3 **Technical Workshop** **Ilumina**
12:00–01:00 PM	Kolonnadensaal 4+5 **Joint Symposia Industry** **Covaris** **Devyser AB** **EnGenome srl** **Integrated DNA Technologies GmbH**
12:00–01:00 PM	Kolonnadensaal 1 **Technical Workshop** **Bionano Genomics**
12:00–01:00 PM	Kolonnadensaal 6 **Technical Workshop** **MetaSystems GmbH**
01:15–02:45 PM	Blauer Saal **Symposium 1** **Extrachromosomal Circular DNA (eccDNA)**
	Chair: Michael Speicher, Graz (Österreich) Malte Spielmann, Lübeck
01:15–01:45 PM	eccDNA in normal tissue (or in the human germline) *Birgitte Regenberg, Copenhagen (Denmark)*
01:45–02:15 PM	Prenatal *Dennis Lo, Hongkong (China) - Remote*
02:15–02:45 PM	eccDNA in cancer cells *Elias Rodriguez-Fos, Berlin*
01:15–02:45 PM	Kolonnadensaal 2+3 **Symposium 2** **Genetics of Aging**
	Chair: Sven Cichon, Basel (Schweiz) Alicja Pacholewska (Köln)
01:15–01:45 PM	Possible functional contribution of somatic mutations in the arterial wall to age-associated vascular disease *Maria Eriksson, Huddinge (Sweden)*
01:45–02:15 PM	Somatic mutations reveal individual histories of aging and development *Alexej Abyzov, Rochester, Minnesota (USA)*
02:15–02:45 PM	Somatic mutation and selection in ageing *Iñigo Martincorena, Cambridge (UK)*
01:15–02:45 PM	Kolonnadensaal 4+5 **EDU1** **Mutationen, die wir beinahe verpasst hätten**
	Bernd Wollnik, Göttingen, Maria Kuzyakova, Göttingen, Martin Zenker, Magdeburg, Rami Abou Jamra, Leipzig, Matthias Begemann, Aachen Dagmar Wieczorek, Düsseldorf, Alexander Hoischen, Nijmegen(The Netherlands)

03:00–04:00 PM	Blauer Saal **Selected Presentations**
	Chair: Brunhilde Wirth, Köln Ingo Kurth, Aachen
03:00–03:15 PM	SEL-01: De novo variants in KDM2A cause a syndromic neurodevelopmental disorder *Konrad Platzer, Leipzig*
03:15–03:30 PM	SEL-02: LHX2 loss of function causes neurodevelopmental deficits in humans and flies *Anne Gregor, Bern (Schweiz)*
03:30–03:45 PM	SEL-03: BTRR complex deficiency is a driver for genomic instability in Bloom syndrome *Ipek Ilgin Goenenc, Göttingen*
03:45–04:00 PM	SEL-04: Biallelic loss-of-function variants in CYHR1 cause syndromic microcephaly *Maria Asif, Köln*
04:00–05:30 PM	Foyer/Industry Exhibition **Get together**
04:30–05:30 PM	Blauer Saal **Sitzung der Jungen Humangenetik**
05:30–06:00 PM	Blauer Saal **Opening Session**
	The future of rare disease therapies *Brunhilde Wirth, Köln*
06:00–07:00 PM	Blauer Saal **Keynote Lecture**
	Chair: Brunhilde Wirth, Köln
	How therapies and newborn screening changed SMA: A paradigma for other rare diseases *Laurent Servais, Oxford (UK)*

Thursday, March 16, 2023

08:45–09:30 AM	Blauer Saal **Plenary Session 1** **Genome Regulation**
	Chair: Bernd Wollnik, Göttingen
	Genome transcription and regulation *Patrick Cramer, Göttingen*
10:00–11:30 AM	Blauer Saal **EDU 2** **TA-Fortbildung: Pitfalls in der NGS-Diagnostik**
	Doreen William, Dresden
10:00–11:30 AM	Kolonnadensaal 2+3 **Workshop 1** **Clinical Genetics**
	Chair: Alma Küchler, Essen Elisabeth Mangold, Bonn
10:00–10:15 AM	W1-01: Homozygous loss-of-function variants in FILIP1 cause autosomal recessive arthrogryposis multiplex congenita with microcephaly *F. Schnabel, Göttingen*
10:15–10:30 AM	W1-02: Heterozygous variants in LAMA5 are associated with short stature by affecting the focal adhesion pathway *A.Schulz, Erlangen*
10:30–10:45 AM	W1-03: Elucidating the clinical and molecular spectrum of SMARCC2 associated NDD in a cohort of 65 affected individuals *G. Vasileiou, Erlangen*
10:45–11:00 AM	W1-04: Assessment of the Genetic Spectrum of Uncombable Hair Syndrome in a Cohort of 107 Individuals *N. Cesarato, Bonn*
11:00–11:15 AM	W1-05: Investigation of Optical Genome Mapping Capabilities and its Potential to be Implemented as a Routine Diagnostic Test *U. Koehler, München*
11:15–11:30 Am	W1-06: GestaltMatcher Database – a medical imaging database for rare Mendelian disorders *H. Lesmann Bonn*
10:00–11:30 AM	Kolonnadensaal 1 **Workshop 2** **Cancer Genetics & Basic Mechanisms of Disease**
	Chair: Stefan Aretz, Bonn Goekhan Yigit, Köln
10:00–10:15 AM	W2-01: Chromosomal imbalances define a site-specific pattern in distant metastases of colorectal cancer *M. Golas, Augsburg*
10:15–10:30 AM	W2-02: APC-specific variant classification rules reduce the burden of variants of uncertain significance in ClinVar and locus-specific databases considerably *S. Aretz, Bonn*
10:30–10:45 AM	W2-03: Epigenetically regulated microRNA-449a inhibits triple negative breast cancer by inducing chromosomal instability *B. Vajen, Hannover*
10:45–11:00 AM	W2-04: Single-nucleus RNA sequencing in a fetal lamb model of hypoplastic left heart syndrome *M. Reuter, Toronto (Canada)*
11:00–11:15 AM	W2-05: Elucidating the genetic network in SHOX2-dependent atrial arrhythmias using human iPSC-derived cardiomyocytes *K. Rädecke, Heidelberg*

11:15–11:30 AM	W2-06: Impact of hypermannosylation on the structure and functionality of the ER and the Golgi complex in AAMR syndrome *P. Franzka, Jena*
10:00–11:30 AM	Kolonnadensaal 4 **Workshop 3** **Bioinformatics**
	Chair: Michael Speicher, Graz (Österreich) Martin Kircher, Lübeck/Kiel
10:00–10:15 AM	W3-01: Systematic analysis and prediction of genes associated with monogenic disorders on human chromosome X *E. Leitão, Essen*
10:15–10:30 AM	W3-02: Deep learning-assisted selection of regulatory variants for high-throughput experimental testing *M. Schubach, Berlin*
10:30–10:45 AM	W3-03: Deep Learning for Pediatric Bone Age Assessment on Patients with Genetically-confirmed Bone Disorders *B. Javanmardi, Bonn*
10:45–11:00 AM	W3-04: GestaltMatcher supports delineation of novel syndromes to facilitate lumping and splitting decision-making by facial phenotype descriptors *H. Klinkhammer, Bonn*
11:00–11:15 AM	W3-05: Enhancing applicability of PEDIA by integrating it into the variant prioritization pipeline with open source GestaltMatcher, CADA and VarFish. *Meghna Ahuja Bhasin, Bonn*
11:15–11:30 AM	W3-06: Face2HPO: Automated HPO Labeling of Syndromic Faces *A.Hustinx, Bonn*
11:45 AM–12:45 PM	Blauer Saal **Talk nach 12** **RNA-Sequenzierung in der Diagnostik**
	RNA-Sequenzierung in der Diagnostik *Tobias Haack, Tübingen*
	Improvements to the Detection of RNA Outliers Pipeline and its application to multi-tissue rare disease cohorts *Vicente Yépez, München*
11:45 AM–12:45 PM	Kolonnadensaal 2+3 **Sitzung der Naturwissenschaftler**
	Chair: Ulrich Zechner, Köln
	Aktivitäten des Netzwerks der Fachwissenschaftler in der Medizin *Heinz Gabriel, Nagold*
	Bericht der Kommission „Fachhumangenetiker (GfH) *Christine Neuhaus, Stadecken-Elsheim*
11:45 AM –12:45 PM	Kolonnadensaal 1 **Technical Workshop** **Limbus Medical Technologies GmbH**
11:45 AM –12:45 PM	Kolonnadensaal 4 **Technical Workshop** **Agilent Technologies Sales & Service GmbH & Co. KG c/o GBT Deutschland GmbH**
11:45 AM –12:45 PM	Kolonnadensaal 5 **Technical Workshop** **AstraZeneca GmbH**

11:45 AM –12:45 PM	Kolonnadensaal 6 **Technical Workshop** **PacBio**
11:45 AM –12:45 PM	Konferenzzimmer 1 **GHGA-Workshop**
01:00–02:00 PM	Blauer Saal **Plenary Session 2** **New Therapies for Rare Diseases**
	Chair: Brunhilde Wirth, Köln Nataliya Di Donato, Dresden
01:00–01:30 PM	From Genetic Basis to Pathophysiology, Preclinical Modelling and Therapeutic Trials for Vascular Anomalies *Miikka Vikkula, Bruxelles (Belgien)*
01:30–02:00 PM	Antisense oligonucleotide silencing of FUS expression as a therapeutic approach in amyotrophic lateral sclerosis *Neil Shneider, New York (USA)*
02:15–03:45 PM	Foyer **Posterwalk 1**
04:00–05:30 PM	Blauer Saal **Pro & Contra I** **Präkonzeptionelles Carrierscreening**
	Chair: Johannes Zschocke, Innsbruck (Österreich) Sabine Rudnik, Innsbruck (Österreich)
04:00–04:03 PM	Begrüßung & Einleitung *Johannes Zschocke, Innsbruck (Österreich)*
04:03–04:13 PM	Entwicklung und aktueller Stand *Sabine Rudnik, Innsbruck (Österreich)*
04:13–04:23 PM	Ziele des PCS in der Reproduktionsmedizin *Thomas Strowitzki, Heidelberg*
04:23–04:33 PM	Welche Methoden stehen zu welchen Kosten zur Verfügung? *Angela Abicht, München*
04:33–04:43 PM	Welche gesellschaftlich-sozialen Aspekte sind zu berücksichtigen? *Ingrid Metzler, Krems an der Donau (Österreich)*
04:43–04:53 PM	Lässt sich das PCS ethisch akzeptabel umsetzen? *Anette Grüters-Kieslich, Berlin*
04:53–04:58 PM	Zusammenfassung *Johannes Zschocke, Innsbruck (Österreich)*
04:58–05:00 PM	Diskussion
04:00–05:30 PM	Kolonnadensaal 2+3 **EDU 3** Bioinformatik
	Peter Krawitz, Bonn
04:00–05:30 PM	Kolonnadensaal 1 **Workshop 4** **Monogenic Disease**
	Chair: Ruthild Weber, Hannover Rami Abou Jamra, *Leipzig*
04:00–04:15 PM	W4-01: A novel intronic GAA repeat expansion in FGF14 causes autosomal dominant adult-onset ataxia *K. Lohmann, Lübeck*
04:15–04:30 PM	W4-02: Pathogenic variants in the gene SPTLC1 cause hyperkeratosis lenticularis perstans

	S. Alter, Freiburg
04:30–04:45 PM	W4-03: TSHZ3 is mutated in human CAKUT *E. Kesdiren, Hannover*
04:45–05:00 PM	W4-04: TMCO3, a putative K+:proton antiporter at the Golgi apparatus, is important for longitudinal growth in mice and humans *T. Holling, Hamburg*
05:00–05:15 PM	W4-05: de novo MAP3K3 variants in patients with abnormalities of the lymphatic system cause attenuation of MAP3K3/MEKK3 downstream signaling pathways *I.G. Kreimer, Hamburg*
05:15–05:30 PM	W4-06: Premature cognitive decline in a mouse model of tuberous sclerosis *V. Engelhardt, Mainz*
04:00–05:30 PM	Kolonnadensaal 4 **Workshop 5** **Complex Diseases**
	Chair: Stefanie Heilmann-Heimbach, Bonn Ulrike Hüffmeier, Erlangen
04:00–04:15 PM	W5-01: Mono- and biallelic variant effects on disease at biobank scale *H. Heyne, Potsdam*
04:15–04:30 PM	W5-02: Potential clinical utility of polygenic risk scores in disease prognosis *J. Wanner, Postdam*
04:30–04:45 PM	W5-03: Saturation mutagenesis data facilitate interpretation of noncoding variants in the IRF6 enhancer associated with orofacial clefting *R. Hollstein, Bonn*
04:45–05:00 PM	W5-04: entfällt
05:00–05:15 PM	W5-05: Genetic associations underlying susceptibility to SARS-CoV-2 and severity of COVID-19: Results of the global COVID-19 Host Genetics Initiative *K. Ludwig, Bonn*
05:15–05:30 PM	W5-06: Novel pharmacological approaches for non-alcoholic steatohepatitis based on a single patient with a de novo variant in FASN *M Musfeldt, Hamburg- Eppendorf*
04:00–05:30 PM	Kolonnadensaal 5 **Workshop 6** **Disease Models and Therapy**
	Chair: Anne Gregor, Bern (Schweiz) Hans Zempel, Köln
04:00–04:15 PM	W6-01: Morphological alterations of microglia in the Magel2 KO mouse model of Prader-Willi and Schaaf-Yang syndromes *F. Althammer, Heidelberg*
04:15–04:30 PM	W6-02: DEGS1-associated leukodystrophy: using patient-derived iPSCs to unravel neuro- and gliopathology *N. Haag, Aachen*
04:30–04:45 PM	W6-03: Human models for White Sutton syndrome: POGZ mutations change the transcriptome and induce defects in neural progenitor cell biology *A. Soliman, Mainz*
04:45–05:00 PM	W6-04: Survival of Glioblastoma patients vaccinated with personalized peptides: a retrospective analysis *S. Biskup, Tübingen*
05:15–05:30 PM	W6-05: 1 Mutation 1 Medicine: a European platform for ASO development and treatments for individuals with nano-rare neurological diseases *H. Graessner, Tübingen*

05:00–05:15 PM	W6-06: Patient and variant stratification for individualized genetic therapies for ultra-rare diseases *M. Lauffer, Leiden (Niederlande)*
06:00–07:30 PM	Blauer Saal **General Meeting GfH**
From 20:00	**Geselliger Abend** **Kongress Palais Kassel**

Friday, March 17, 2023

08:45–09:30 AM	Blauer Saal **Plenary Session 3** **Cancer Genomes**
	Chair: Johanna Tecklenburg, Hannover Michael Speicher, Graz (Schweiz)
	Somatic mutations in cancer: Tracking mutational footprints to study the origin of cancer *Ruben van Boxtel, Utrecht (Niederlande)*
10:00–11:00 AM	Blauer Saal **Symposium 3** **New Developments in Old Syndromes**
	Chair: Sabine Hoffjan, Bochum Miriam Elbracht, Aachen
10:00–10:30 AM	New insights and ASO treatment developments for Angelman Syndrome *Ype Elgersma, Rotterdam (Niederlande)*
10:30–11:00 AM	New players explaining cognitive impairments in Down syndrome *Mara Dierssen, Barcelona (Spanien)*
10:00–11:00 AM	**Kolonnadensaal 2+3** **Symposium 4** **Completing Human Genomes**
	Chair: Ingo Kurth, Aachen Alex Hoischen, Nijmegen (Niederlande)
10:00–10:30 Am	The Human Pangenome Project: Creating a Reference that Better Represents Human Global Genetic Diversity *Karen Miga, San Diego (USA)*
10:30–11:00 AM	The advantages of complete, telomere-to-telomere genomes *Adam Philippy, Bethesda (USA)*
11:00 AM –12:30 PM	Foyer **Posterwalk 2**
12:45–01:45 PM	Blauer Saal **Technical Workshop** **MGI International Sales Co., Limited.**
12:45–01:45 PM	**Kolonnadensaal 2+3** DFG-Fördermöglichkeiten
	Lisa Nalbach (DFG), Bonn Christian Kubisch, Hamburg
12:45–01:45 PM	Kolonnadensaal 1 **Technical Workshop** **Singleron Biotechnologies**
12:45–01:45 PM	Kolonnadensaal 4 **Technical Workshop** **Twist Bioscience**
12:45–01:45 PM	Kolonnadensaal 5 **Technical Workshop** **QIAGEN GmbH**
12:45–01:45 PM	Kolonnadensaal 6 **Technical Workshop** **Oxford Nanopore Technologies**
02:00–03:30 PM	Blauer Saal **Pro & Contra II** **Teilen von genetischen Daten**
	Peter Krawitz, Bonn

02:00–03:30 PM	Kolonnadensaal 2+3 **EDU 4** **ACMG-AMP Klassifizierungsrichtlinien**
	Andreas Laner, München
02:00–03:30 PM	Kolonnadensaal 1 **Workshop 7** **Neurodevelopmental Disorders**
	Chair: Mert Karakaya, Köln Silke Redler, Düsseldorf
02:00–02:15 PM	W7-01: De novo variants in CNOT9 cause a neurodevelopmental disorder with or without epilepsy *R. Abou Jamra, Leipzig*
02:15–02:30 PM	W7-02: DOCK4 loss-of-function variants lead to neurodevelopmental delay *V. Bothe, Leipzig*
02:30–02:45 PM	W7-03: De novo variants in DLGAP1 cause a variable neurodevelopmental disorder *H. Lyubenova, Berlin*
02:45–03:00 PM	W7-04: de novo PHF5A variants cause craniofacial abnormalities and developmental delay *F.L. Harms, Hamburg*
03:00–03:15 PM	W7-05: CUX1-related neurodevelopmental disorder: Deep insights into phenotype-genotype spectrum and underlying pathology *H. Oppermann, Leipzig*
03:15–03:30 PM	W7-06: Mutant SSU processome component C1orf131 causes ribosome biogenesis dysfunction leading to primary microcephaly *M.S. Hussain, Köln*
02:00–03:30 PM	Kolonnadensaal 4 **Workshop 8** **Technologies and Clinical Application**
	Chair: Per Hofmann, Konstanz Larissa Arning, Bochum
02:00–02:15 PM	W8-01: Intra-operative prediction of brain tumor classes from Nanopore data *H. Kretzmer, Berlin*
02:15–02:30 PM	W8-02: Parallel In-Depth Analysis of Repeat Expansions in Ataxia Patients by Long-Read Sequencing *H. Erdmann, München*
02:30–02:45 PM	W8-03: Combined somatic copy number alteration and fragmentation analysis for treatment monitoring in colorectal cancer patients using liquid biopsy *A. Hallermayr, München*
02:45–03:00 PM	W8-04: Meta-Analysis of Multiomics-Neurodegeneration data *S. Klingenberg, Mainz*
03:00–03:15 PM	W8-05: Prenatal Trio-Exome-Analysis: A powerful routine diagnostic tool *U. Ahting, Martinsried*
03:15–03:30 PM	W8-06: 2000 trio exome sequencing on research base: output and outlook *A. Golod, Leipzig*
02:00–03:30 PM	Kolonnadensaal 5 **Workshop 9** **Functional Genomics**
	Chair: Andreas Forstner, Bonn Anja Weise, Jena
02:00–02:15 PM	W9-01: Disruption of the topologically associated domain at Xp21.2 is related to 46,XY gonadal dysgenesis *V. Yumiceba, Lübeck*

02:15–02:30 PM	W9-02: Enhancer hijacking at the ARHGAP36 locus is associated with connective tissue to bone transformation *U. Melo, Berlin*
02:30–02:45 PM	W9-03: Frameshifts of intrinsically disordered regions alter nucleolar phase separation and cause genetic disease *M.A. Mensah, Berlin*
02:45–03:00 PM	W9-04: Effects of TXNIP-variants to systemic and cellular lipid and glucose handling *J.-J. Scholz, Hamburg*
03:00–03:15 PM	W9-05: SEC24Cdeficiency causes trafficking and glycosylation abnormalities and leads to a congenital syndrome including anemia, microcephaly, and cataracts *N. Bögershausen, Göttingen*
03:15–03:30 PM	W9-06: Clinical and molecular characterization of MAU2 variants uncover an essential role of MAU2 in transcriptional regulation *I. Parenti, Essen*
03:45–04:00 PM	Blauer Saal **Closing remarks**