Kassel 2023
34. gfh-Jahrestagung

Posterwalk 1

16.03.2023 von 14:15–15:45 Uhr

Slot 1: 14:15–14:45 Uhr	
Terminal	**Titel**
Basic Mechanisms and Epigenetics	
1	P1-1: Evidence of a quantum physics layer of epigenomics from modeling dissipative charge-transfer dynamics along DANN **M. Rossini**
1	P1-2: Functional analyses of *SEPHS2* variants link ferroptosis to cholesterol metabolism **J. Ridder**
1	P1-3: Comparative DNA methylation profiling of human and murine ALK+ LBCL with aberrant ALK expression reveals similarities to plasma cell neoplasms **S.Glaser**
1	P1-4: Nuclear envelope proteins in myotonic dystrophy: extending evidence for a relevant contribution to the phenotype **P. Meinke**
2	P1-5: Transcriptome analysis on Emery-Dreifuss muscular dystrophy primary tissue cultures reveals alterations of metabolic, fibrotic, and splicing pathways **S. Hintze**
2	P1-6: *TERT* Promoter DNA Methylation Differs Between Subtypes of Malignant Lymphomas **A.G. Kouroukli**
2	P1-7: Characterizing isoform-specific activities of thyroid hormone receptor α **N. Härting**
2	P1-8: Altered DNA methylation profiles in *SF3B1* mutated CLL patients **A. Pacholewska**
3	P1-9: Engineering of an inducible iPSC line to study imprint establishment at the PWS-SRO **J. Haake**
3	P1-10: Functional characterization of microRNA-375-3p in triple-negative breast cancer **F. Henning**
3	P1-11: Huntington's disease-like phenotype and dystonia: are pathogenic variants in *STUB1* and *TUBB4A* a common cause? **P. C. Panitz**
3	P1-12: Features of lncRNA expressions and regulation across different brain regions of primates **M. Navandar**
3	P1-13: Comprehensive RNA sequencing demonstrates that endothelial differentiation of *CCM1* knockout iPSCs triggers a specific gene expression signature **R. A. Pilz**
Cancer Genetics	
4	P2-1: Integrated mRNA and miRNA expression profiling on archived tissue of high-grade B-cell lymphomas with 11q aberrations **G. Di Stefano**
4	P2-2: Identification of regulatory variants in genes predisposing for hereditary breast and ovarian cancer **B. A. Allister**
4	P2-3: Exome sequencing reveals clonal evolution in tyrosine kinase inhibitor-resistance cell line models with evolving mutations in *PTPN11*, *KRAS* and *NRAS* **I. Nagel**
4	P2-4: Highly sensitive Liquid Biopsy Duplex Sequencing complements tissue biopsy to enhance detection of clinically relevant genetic variants **A. Hallermayr**
4	P2-5: Targeted RNA analysis classified potential germline splice variants in tumour risk genes **A. K. Sommer**
5	P2-6: Genetic and epigenetic characterization of B-cell neoplasms with *IGH*::*BCL3*-translocation **C. Drewes**

5	P2-7: Liquid Biopsy in a clinical setting - evidence from real world data **S. Biskup**
5	P2-8: Identification of human papillomavirus 16 genomic integration sites in oropharynx squamous cell carcinoma and their impact on host gene expression **E. Chteinberg**
5	P2-9: CRISPR/Cas9-mediated generation of cell lines with one and two defined fusion transcripts for Next Generation Sequencing based molecular analyses **M. F. Hossain**
5	P2-10: Ongoing V(D)J recombination leads to structural aberrations in B-lymphoblastic leukemia/lymphoma with *IGH::IL3* fusion **D. Marx**
6	P2-11: Gene expression profile of eccrine porocarcinoma point to biologically heterogeneous diseases and gene expression signatures of general downregulation **S. Redler**
6	P2-12: Spectrum of somatic variants and clinical phenotypes in RUNX1-associated familial platelet disorder with predisposition to hematologic malignancies **A. Förster**
6	P2-13: A functional characterization of *hsa-miR-129-5p* in triple-negative breast cancer cells **R. Steinmetz**
6	P2-14: Establishment of a functional assay for the classification of VUS in *FANCJ* **A. Droste**
6	P2-15: Malignant rhabdoid tumors in patients conceived following assisted reproduction technologies (ART) – genetic, epigenetic and clinical features **J. Kolarova**
7	P2-16: Unmet need for interdisciplinary cooperation: Case series of patients diagnosed with hereditary leiomyomatosis and renal cell cancer (HLRCC) **M. Kachanov**
7	P2-17: Comparison of glioblastoma tissue *vs* glioblastoma stem-like cells: genetic, biophysical and bioinformatical investigations **V.-P. Brandt**
7	P2-18: An inherited common PTPN11 variant associated with neuroblastoma but without characteristic features of Noonan syndrome **J. Hoyer**
7	P2-19: Diagnostic yield and clinical relevance of expanded germline genetic testing for HBOC patients **J. Henkel**
7	P2-20: Monitoring Cancer Treatment Response with two different NGS-based strategies **B. Küchler**
Slot 2: 14:45–15:15 Uhr	
Cancer Genetics	
1	P2-21: Implementation of microarray-based genome-wide somatic CNV analyses into molecular tumor board recommendations for targeted cancer therapy **K. Oehl-Huber**
1	P2-22: *DDX41*-associated donor cell leukemia **R. Meyer**
1	P2-23: Capture RNA-seq as supplement to DNA germline testing to increase diagnostic yield **J. Romic-Pickl**
1	P2-24: Integrating functional analysis into *RUNX1* germline variant classification **M. Decker**
1	P2-25: *TP53* single hit and double hit events and their impact on prognosis in hematological malignancies **A. Stengel**
Clinical Genetics, Genetic Counselling and Prenatal Diagnosis	
2	P3-1: Single centre experience with diagnostic whole genome sequencing in 70 patients with rare diseases **D. William**
2	P3-2: Compound-heterozygous *GRIN2A* null variants associated with severe developmental and epileptic encephalopathy **V. Strehlow**

2	P3-3: SysNDD is an expert-curated map of neurodevelopmental disorders to study gene disease connections **B. Popp**
2	P3-4: Parallel deletion and duplication at 7q11.23 in a silent carrier for two reciprocal syndromic disorders **J. L. Lühmann**
3	P3-5: Resolving pathogenicity of non-truncating *ARID1B* variants in Coffin-Siris syndrome **E. Bosch**
3	P3-6: Biallelic variants in the Guanine Exchange Domain Expanding the Spectrum of *EEF1D*-related Neurodevelopmental Disorders **D. Wieczorek**
3	P3-7: Biallelic *KITLG* variants lead to a distinct spectrum of hypomelanosis and sensorineural hearing loss **B. Vona**
3	P3-8: Detection of 5q-Spinal Muscular Atrophy by short-read next-generation sequencing - unexpected results **A. Abicht**
4	P3-9: Exome sequencing individuals and families with suspected monogenic primary immunodeficiencies – experiences of a single center **U. Hüffmeier**
4	P3-10: *Specialised Dup15Q clinic at Heidelberg University Hospital* **C. Gabriel**
4	P3-11: Clinical genetic assessment and medical care of children with tumor predisposition syndromes. An interdisciplinary approach **A. L. Nitschke**
4	P3-12: Whole-genome sequencing of 65 families with inherited retinal dystrophies **C. Kiel**
4	P3-13: Mutations affecting Glycine-X-Y repeats in the *COL4A6* gene cause hearing loss **D. Owrang**
5	P3-14: Austrian Exome meetings – a nationwide competence sharing initiative **S. Verheyen**
5	P3-15: Long-read genome sequencing and RNA sequencing resolve an intronic LINE-1 insertion in the APC gene in a so far unsolved adenomatous polyposis family **A. A. Baumann**
5	P3-16: Towards targeted therapy in Li-Fraumeni syndrome: Case report of a 62-year-old patient with osteosarcoma **V. I. Gaidzik**
5	P3-17: Using genome sequencing to diagnose unsolved cases exemplified by an atypical case of Rett syndrome **S. Schuhmann**
5	P3-18: Novel de novo *POLR3B* variant responsible for demyelinating Charcot-Marie-Tooth disease **A.-S. Onuk**
6	P3-19: Whole exome sequencing identifies a heterozygous *TRPC3* missense variant in a patient with adult onset spinocerebellar ataxia **T. Bender**
6	P3-20: *RYR2* and atrial fibrillation – it is not always a disease of old age **D. S. Westphal**
6	P3-21: Prenatally detected complex mosaicism in a baby of uncertain sex **F. Stock**
6	P3-22: Refining the phenotypic spectrum of Poretti-Boltzhauser syndrome caused by biallelic variants in *LAMA1* **B. Greiten**
6	P3-23: Infantile acute liver failure in *EFL1*-associated Shwachman-diamond syndrome **A. van der Ven**
7	P3-24: Heterozygous *ADAR* pathogenic variant associated with variable neurological symptoms and incomplete penetrance in one family **A.-K. Bauer**
7	P3-25: Blended phenotype in an infant with developmental delay, bilateral cleft lip and palate and coloboma due to *de novo* deletions at 2q24.2q24.3 and 6p24 **M. Walther**
7	P3-26: Another case of Cutis Marmorata Teleangiectatica Congenita caused by postzygotic *GNA11* mutation **K. Komlosi**
7	P3-27: Whole genome sequencing offers new possibilities for the identification of disease-causing gene variants escaping detection by whole exome analysis **M. A. H. Hofrichter**

Slot 3: 15:15–15:45 Uhr	
Clinical Genetics, Genetic Counselling and Prenatal Diagnosis	
1	P3-28: Comparison of three patients with variants in histone methyltransferase complex genes SETD1B, KMT2A and KMT2B **G. Miltenberger-Miltenyi**
1	P3-29: First Patient With X-linked Developmental Disorder Caused By A Partial Duplication Of The *ZDHHC9* Gene **V. K. Berge**
1	P3-30: KMT2B associated intellectual disability without dystonic features: an unusual phenotype for a dystonia gene **C.C. Becker**
1	P3-31: Severe psychomotor and developmental delay due to a homozygous deletion of *CTNND2* **M. Rieger**
2	P3-32: Combined Partial Monosomy 9p and Partial Trisomy 20p: A Case Report and Review of the Literature **D. Wand**
2	P3-33: Missense variants in *MAGEL2* as a potential cause of Schaaf-Yang syndrome related phenotypes **S. Unser**
2	P3-34: Male patient with a novel likely pathogenic variant in *HDAC4* causing Neurodevelopmental disorder with central hypotonia and dysmorphic facies **J. Köhler**
2	P3-35: When you hear hoofbeats, think zebra, not horse **J. Tecklenburg**
3	P3-36: *COL4A1*/*2*-related disorders - A report of two affected children with rare multisystem diseases **M. K. Moddemann**
3	P3-37: Establishment of a Genetic Counsellor Association for the German-speaking Countries **G. Schwaninger**
3	P3-38: Clinical exome analysis revealed a mosaic *KMT2A* deletion in a patient with unexplained developmental delay **S. Döhnert**
3	P3-39: Siblings with two different molecular diagnoses – a challenge for a family-based exome sequencing **B. Eichhorn**
3	P3-40: Case report: Rapid progressive ataxia and mild cognitive impairment due to digenic inheritance of pathogenic variants in *TGM6* and *PRNP*? **M. Neidhardt**
4	P3-41: *CYLD* Cutaneous Syndrome. A case presentation **D. González Fassrainer**
4	P3-42: *PRNP* octapeptide insertion in a familial case of neurodegenerative disease elusive in routine neuropathological examination **V. G. Paul**
4	P3-43: Atypical or particularly mild Rett syndrome? - A family with a novel frameshift mutation in *MECP2* **S. Riedel**
4	P3-44: One Year of Genome Sequencing in Diagnostic Routine **C. Du**
4	P3-45: Only one beer can be mortal: a case report of two sisters with cardiac arrest due to a homozygous *PPA2* mutation **S. Rudnik-Schöneborn**
5	P3-46: Progressive congenital microcephaly, growth retardation and dysmorphisms in a male infant with an intronic splice variant in *GPKOW* **J. Lisfeld**
5	P3-47: Hydatidiform mole with coexistent fetus **K. Bahlke**
5	P3-48: Clinical phenotype in a male patient with biallelic pathogenic deletions in *PTEN,* a case report **A. Möllring**
5	P3-49: Case report of a hemizygous intragenic deletion of *WDR13* - Further evidence for the role of *WDR13* in X-linked intellectual disability **K. Cremer**

5	P3-50: Rare diagnosis of acromicric dysplasia in an adult woman **N. Dragicevic Babic**
6	P3-51: Prenatal Presentation of *ARCN1*-associated Skeletal Dysplasia **A. Busche**
6	P3-52: Colon cancer in a 41-year-old patient with *MORC2*-related neurodevelopmental disorder **B. Herr**
6	P3-53: Three sisters with Schwartz-Jampel syndrome and an additional cardiac phenotype **H. Surowy**
6	P3-54: Novel homozygous *LAMB1* in-frame deletion in a pediatric patient with brain anomalies and cerebrovascular event **L. Toutouna**
6	P3-55: Vascular Ehlers-Danlos syndrome with co-occuring metachondromatosis in a German family **L. Wankner**
7	P3-56: Novel pathogenic variant in PUF60 detected in a male patient with Verheij syndrome **D. Kaschta**
7	P3-57: Clinical and etiological differences between alopecia areata patients with and without immune-mediated comorbidities **Y. Gossmann**
7	P3-58: 16q12.2 microdeletion as a rare cause of developmental delay **C. Barkhaus**
7	P3-59: Panel versus whole Exome genetic testing in a real-world cohort of patients with amyotrophic lateral sclerosis (ALS) **K. P. Lubieniecki**
7	P3-60: Delineation of *AMMECR1* as a candidate gene for idiopathic short stature **C. T. Thiel**

Posterwalk 2

17.03.2023 von 11:00–12:30 Uhr

Slot 1: 11:00–11:30 Uhr	
Terminal	**Titel**
Clinical Genetics, Genetic Counselling and Prenatal Diagnosis	
1	P3-61: Characterization of CNVs and SNVs in Known and Candidate Genes in Patients with Idiopathic Short Stature **A. Fink**
1	P3-62: Implementation of a high-risk Familial Breast and Ovarian Cancer prevention network in sparsely populated Western Pomerania **L. Stark**
1	P3-63: *ATXN2* gene variants in neurodegenerative diseases **J. M. Lubieniecka**
1	P3-64: Combined features of short-rib-polydactyly syndrome and oral-facial-digital syndrome in a fetus: Clinical diagnosis allows targeted genetic testing **M. Messner**
2	P3-66: Next-Generation sequencing identifies a novel de novo pathogenic variant in the DYRK1A gene causing syndromic intellectual disability and microcephaly **D. Berner**
2	P3-67: Prenatal case report: Modified Whole Exome Sequencing revealed a homozygous mutation in a RNA-coding gene causing MOPD1 **A. Pohle**
2	P3-68: *De novo* missense variant in *PBX1* in a patient with 46,XY gonadal dysgenesis **S. Ledig**
3	P3-69: Joubert syndrome in two siblings – novel truncating mutation **S. Weidensee**
3	P3-70: KaRhab – A registry for cardiac rhabdomyomas **V.-M. Herrmann**

3	P3-71: *NXN*-related Robinow-syndrome – expansion of the clinical and genetic spectrum **E. Schreiner**
3	P3-72: An inherited inversion detected by exome sequencing causes ATP6V0A2 related cutis laxa **L. Segebrecht**
3	P3-73: Mutations in Follistatin-like 1 (*FSTL1*) may be associated with a connective tissue disorder **O. Klaas**
4	P3-74: How to take a short cut – from genetics to specific metabolic diagnostics **J. Beuschlein**
4	P3-75: The clinical utility of noninvasive prenatal testing (NIPT) for detection of fetal unbalanced translocations **M. Shoukier**
4	P3-76: Mutational spectrum of suspected Charcot-Marie-Tooth disease in 399 patients **V. Klaschka**
4	P3-77: The phenotype in adulthood of rare genetic diseases: Nicolaides-Baraitser, Coffin-Siris, Costello and cardiofaciocutaneous syndrome **A. Schmetz**
5	P3-78: Molecular autopsy – proclamation for an undervalued procedure with preventive power for relatives at risk **A.-C. Berking**
5	P3-79: REPORT OF 23 NEW INDIVIDUALS WITH *PHIP*-ASSOCIATED CHUNG-JANSEN SYNDROME **A. Kampmeier**
5	P3-80: Delineation of the DLL1-associated phenotype **L. A. Koch**
5	P3-81: Approach to Cohort-Wide Re-Analysis of Exome Data in 1000 Individuals with Neurodevelopmental Disorders **T. Bartolomaeus**
Complex Diseases, Population Evolutionary Genetics and Genetic Epidemiology	
6	P4-1: A transgenerational mutational signature from ionizing radiation exposure **F. Brand**
6	P4-2: Using the ends of the polygenic risk score spectrum for schizophrenia to better capture anxiety-related brain structures – A UK Biobank study **L. Sindermann**
6	P4-3: Genome-wide association study of therapy response in alopecia areata **F. B. Basmanav Ünalan**
6	P4-4: Identification of pathobiologically relevant cell types for male-pattern hair loss **S. Henne**
6	P4-5: Analyses of bipolar disorder polygenic risk in iPSC-derived neural progenitors from genetically characterized patients and controls **K. Hünten**
6	P4-6: Identification of marker chromosome mosaicism in a newborn with VACTERL association **J. Ćomić**
7	P4-7: Analysis of non-syndromic cleft lip with or without cleft palate GWAS candidate genes in single-cell expression data from human embryonic development **A. Siewert**
7	P4-8: Use of soft-clustering algorithms to reduce the genetic complexity of chronic kidney disease **A. Eoli**
7	P4-9: Gene-based association analysis of transcription factors involved in differential regulation of bipolar susceptibility genes during neurodevelopment **P. Thirunavukkarasu**
7	P4-10: The role of rare genetic variants enrichment in epilepsies of presumed genetic etiology **D. Le Duc**
7	P4-11: Epigenetic age acceleration in major psychiatric disorders **F. David**
Slot 2: 11:30–12:00 Uhr	
Cytogenetics and CNVs	

1	P5-1: Mosaic Chromosomal Alterations in non-neoplastic cells of Children with Burkitt Lymphoma **A. Fischer**
1	P5-2: When should we perform prenatal testing for uniparental disomies and imprinting disorders? **T. Eggermann**
1	P5-3: Complex triplication rearrangement (DUP-TRP-DUP) in a patient with severe neurodevelopmental disorder **D. Popp**
1	P5-4: Prenatally detected whole gene deletion of *SF3B4* associated with Nager syndrome **M. Kuschmann**
1	P5-5: Rare prenatal case with trisomy 22 and triploidy mosaicism – need for cytogenetic expertise **A.-C. Teichmann**
2	P5-6: Case report of two patients carrying a partial trisomy 16q **C. Wilmsen**
2	P5-7: Class I and class II analphoid supernumerary marker chromosomes: Pitfalls in prenatal diagnostic and parental transmission **Y. Stratis**
2	P5-8: De novo reciprocal translocation of the long arms of chromosome 5 and chromosome 9 in a patient with features of Bosch-Boonstra-Schaaf syndrome **A. C. Schnause**
2	P5-9: Comparison of Bionano optical genome mapping with conventional cytogenetic tests - region-specific analyses **M. Blankenburg**
3	P5-10: Two cases of Xq28 duplications with uncommon pathomechanisms **M. Apelt**
3	P5-12: Mild manifestations of trisomy 13 **A. T. Abad Perez**
3	P5-13: double ring chromosome 2 mosaicism due to large interstitial deletion 2q32.1q36.1 in a boy with muscular hypotonia **F. Roessler**
3	P5-14: Cas9-mediated nanopore sequencing enables precise characterization of structural variants in *CCM* genes **D. Skowronek**
Monogenic Disease – from Gene Identification to Molecular Mechanism	
4	P6-1: Altered Notch signaling in Dowling-Degos disease - a transcriptomic insight into disease pathogenesis **S. Kumar**
4	P6-2: Somatic mosaicism in *STAG2*-associated cohesinopathies: Expansion of the genotypic and phenotypic spectrum **J. Schmidt**
4	P6-3: Variants in *FGF10*: a cause of isolated neonatal lung developmental disorder? **S. v. Hardenberg**
4	P6-4: *In vitro* modelling of Sorsby fundus dystrophy **K. Plößl**
4	P6-5: Loss of function variants affecting the STAGA complex component *SUPT7L* cause a developmental disorder with generalized lipodystrophy **B. Fischer-Zirnsak**
5	P6-6: Gene Therapy for Best vitelliform macular dystrophy based on Haplotype-Specific CRISPR/Cas Editing **A. Milenkovic**
5	P6-7: Detailed analysis of alpl/Tnap function in zebrafish and establishment of a new *in vivo* model constituting the rare disease hypophosphatasia **D. Liedtke**
5	P6-8: Functional analysis of potential splice site variants in craniosynostosis causing genes **A. Borst**
5	P6-9: Non-5q-SMAs: a conundrum with lessons for 5q-SMA or an overdiagnosed clinical picture? **M. Karakaya**
5	P6-10: Unmasking splicing effects of Otoferlin missense variants **Z. Zhang**
6	P6-11: Functional analysis of a *MALRD1* variant associated with increased bile acid synthesis and hepatic cholestasis **V. Reichenbach**

6	P6-12: Functional analysis of novel ZRS enhancer variants associated with preaxial polysyndactyly **R. Hark**
6	P6-13: Two cases with MBD5-associated neurodevelopmental disorder expand the known disease spectrum and highlight role of WGS in detecting non-coding CNVs **J. Fischer**
6	P6-14: Multigene panel sequencing in highly consanguineous families and Isolated patients with congenital forms of skeletal dysplasias **N. Kakar**
6	P6-15: Whole genome sequencing uncovers a 8,8 kb deletion between *L1CAM* and *AVPR2* in a boy with Hirschsprung disease and nephrogenic diabetes insipidus **I. Bader**
7	P6-16: Detection of circular RNAs in a mouse model for intellectual disability **L. R. Jensen**
7	P6-17: Novel variant in Kozak sequence of *KCNC3* might affect ion channel expression **M. C. Reis**
7	P6-18: Affection of the catalytic triad of UFSP2 leads to a severe phenotype of spondyloepimetaphyseal dysplasia type Di Rocco **L. Mattern**
7	P6-19: A *TMC8* splice variant causes epidermodysplasia verruciformis in a Pakistani family **X. Xiong**
7	P6-20: Functional characterization of variants of unknown significance in the Alagille Syndrome-associated genes *JAG1* and *NOTCH2* ***N. Buhl***
7	P6-21: Mutational Analyses in Egyptian and Pakistani Patients with Skeletal Phenotypes **N. Wasif**
Slot 3: 12:00–12:30 Uhr	
Monogenic Disease – from Gene Identification to Molecular Mechanism	
1	P6-22: *NUDT2* loss-of-function is associated with a recessive neurodevelopmental disease with mental retardation, movement disorder, and peripheral neuropathy **J. C. Hennings**
1	P6-23: Comprehensive genetic and clinical characterization of 200 patients with idiopathic Small Fiber Neuropathy **A. Lischka**
1	P6-24: Role of homozygous variants in generalised pustular psoriasis and two other rare pustular diseases **M. D. Hayatu**
1	P6-25: Possible association of bi-allelic variants in DHX16 with epilepsy and recurrent subdural hematoma **A. Caliebe**
1	P6-26: Identification of a large inversion disrupting the dystrophin gene in a 24-year-old with Duchenne muscular dystrophy by optical genome mapping **L. S. Erbe**
2	P6-27: Further evidence for an in-frame deletion in the *HP* gene to cause hepatopathy due to ectopic endoplasmic reticulum storage **A. Stalke**
2	P6-28: Description of a homozygous deletion in *COL7A1* in three non-related families **K. Echle**
2	P6-29: Mouse models to study the pathophysiology of GMPPB-associated disorders **M. Schuring**
2	P6-31: Homozygous disruption of *PPM1D* induces defective DNA repair and genomic instability in a patient with syndromic microcephaly **G. Yigit**
3	P6-32: *MECR* is essential for coordinated energy transformation **I. Evangelakos**
3	P6-33: Modelling 16p11.2 Microduplication syndrome: transcriptome analysis of human iPSC-derived neural cells highlights defects in early neural progenitor **M. Bertin**
3	P6-34: A homozygous variant in the *VAPA* gene leads to disturbed lipid metabolism and organellar contact sites **L. Hipp**
3	P6-35: Disease-specific biomarkers of pathogenic *HRAS* variants in skin **T. Nauth**

Technology and Bioinformatics	
4	P7-1: Whole-exome sequencing as a method for identification of uniparental disomies **J. Moch**
4	P7-2: Lipidomics as a functional analysis tool of genetic variants in (un-)known hereditary metabolic disease **C. Schlein**
4	P7-3: aRgus: multilayer visualization of non-synonymous SNVs & pathogenicity score modeling for enhanced vulnerability assessment **H. Brennenstuhl**
4	P7-4: Development of a quantitative splice assay for the characterization of variants of uncertain significance using long-read sequencing **A.-L. Katzke**
4	P7-5: Single-cell RNA phenotyping of a mouse model for hypothyroidism reveals a pivotal role of thyroid hormone receptor alpha for hypothalamic development **V. Sreenivasan**
5	P7-6: Decipher patient needs in the process of genetic counseling to individualize the personal assistant GEN-SUPPORT **N. Ammon**
5	P7-7: Automated DNA extraction for detection of tandem repeat expansions with nanopore sequencing **H. Faust**
5	P7-8: Navigating the uncharted landscape of RNA modifications by leveraging Oxford Nanopore Technology's direct sequencing and machine learning algorithms **A. Wierczeiko**
5	P7-9: Inferring gene-level expression levels from mid-to-low coverage cell-free DNA shotgun sequencing **K. Köhler**
5	P7-10: Detection of small structural variants in patients with high clinical suspicion **A. Moscu-Gregor**
5	P7-11: Information integration to predict chromatin folding using neural networks **S. Sys**
6	P7-12: NanopoReaTA: user-friendly tool for nanopore-seq real-time transcriptional analysis **S. Pastore**
6	P7-13: Third Generation Sequencing of a Skin Lesion Swab from a Monkeypox Infection **C. Hewel**
6	P7-14: Clinvar-This: ClinVar API Submission Made Easy **M. Holtgrewe**
6	P7-15: Long read sequencing identifies novel pathogenic intronic variants in Gitelman syndrome **K. Neveling**
6	P7-16: Pathogenicity prediction of missense variants using AlphaFold2-derived features **A. Schmidt**
7	P8-1: Prospective evaluation of NGS based sequencing in epilepsy patients: results of 7 clinical genetic laboratories **M. Witzel**
7	P8-2: All-in-one AAV vectors for CRISPR/SpCas9 genome editing **F. Biasella**
7	P8-3: POLG assoc. mitochondriopathies - Prel. results of a recruiting patient fibroblast study and human neurons show reduced mtDNA and therapeutic options **C. Çakmak**