	Presenter	Titel
Thursday		**29.08.2019**
8.00 h		**Onsite registration opens**
8.30 h – 09.00 h		**Welcome**
	Prof. Dr. Birgit Lorenz	President of ISGEDR Speaker of the Section Genetics of the German Ophthalmological Society (DOG) Head and Chairman of the Department of Ophthalmology, Justus-Liebig University Giessen and University Medical Center Giessen and Marburg GmbH, Giessen Campus
	Prof. Dr. Joybrato Mukherjee	President of the Justus-Liebig University Giessen
	Prof. Dr. Wolfgang Weidner	Dean of the Medical Faculty, Justus Liebig University, Giessen
	Prof. Dr. Andreas Böning	Chief Medical Officer, University Medical Center Giessen and Marburg GmbH, Giessen Campus
09.00 h – 10.50 h	**1. Session**	**Performing And Communicating Molecular Diagnostics** *Chair: Hanno Bolz, David Mackey*
09.00 h	Bolz, Hanno Jörn, Frankfurt, Germany	T01: NGS: Diagnostic opportunities and challenges of gene panel, whole-exome and whole-genome sequencing
09.20.h	Mackey, David, Subiaco, Australia	T02: Predictive testing in glaucoma
09.40 h	Capasso, Jenina, Philadelphia, USA	T03: We need more genetic counselors!
10.00 h	Arno, Gavin, London, United Kingdom	O01: Interrogation of the 100,000 genomes project ophthalmic disease cohort reveals novel genes, new associations and previously undetectable mutations
10.10 h	Debenedictis, Meghan, Cleveland, USA	O02: The value of CNV analysis for inherited retinal diseases
10.20 h	Hufnagel, Robert, Bethesda, USA	O03: Modeling gene constraint in disease populations for clinical molecular diagnostics
10.30 h	Kellner, Ulrich, Siegburg, Germany	O04: DNA testing in a series of 944 patients with inherited retinal dystrophies from a single german reference center
10.40 h	Wells, Kirsty, Helsinki, Finland	O05: Enhancing diagnostic performance in inherited retinal diseases through advances in high resolution copy number detection and RPGR ORF15 sequencing
		Associated Posters
	Lei, Bo, Zhengzhou, Henan, China	P01: Whole-exome sequencing identifies a novel homozygous missense variant in *REEP6* in a retinitis pigmentosa patient complicated with macular hole
	Pantrangi, Madhulatha, Marshfield, USA	P02: Shaping 'PreventionGenetics' comprehensive inherited retinal disorder panel for the clinical setting and to improve diagnostic yield
	Chan, Choi Mun, Singapore, Singapore	P03: Genetic testing for macular dystrophies : The Singapore National Eye Centre experience
10.50 h – 11.20 h		**Coffee Break** **Poster Session**
11.20 h – 13.00 h	**2. Session**	**ClinicalStudies in Gene Therapy I** *Chair: Francesca Simonelli, Juliana Sallum*
11.20 h	Leroy, Bart P, Ghent, Belgium	T04: Safety & Efficacy of Antisense Oligonucleotide Therapy (QR-110) in LCA10 Patients with the c.2991+1655A>G Allele in CEP290

11.40 h	Sahel, José-Alain, Paris, France	T05: Leber Hereditary Optic Neuropathy: Gene Therapy for an Ultra-Orphan Blinding Disease
		Phenotypes – Free Papers
		Chair: Francesca Simonelli, Juliana Sallum
12.00 h	Brooks, Brian, Bethesda, USA	O06: Homozygous frameshift mutations in *FAT1* cause a syndrome characterized by colobomatous-microphthalmia, ptosis, nephropathy and syndactyly
12.10 h	Escher, Pascal, Bern, Switzerland	O07: Molecular and cellular mechanisms in *NR2E3*-linked retinal degenerations
12.20 h	Grubich Atac, David, Schlieren, Switzerland	O08: *ATOH7* loss-of-function mutations in a family with hypoplasia of the optic nerve
12.30 h	Munier, Francis, Lausanne, Switzerland	O09: Prevention of intravitreal melphalan-induced chorioretinopathy: identification of potential risk factors.
12.40 h	Ruddle, Jonathan, Parkville, Australia	O10: The genetic and clinical landscape of nanophthalmos in an Australian cohort
12.50 h	Chaudhuri, Zia, Delhi, India	O11: Pedigree analysis of familial primary concomitant horizontal strabismus in a South Asian population
		Associated Posters
	Abdel-Rahman, Mohamed, Columbus, USA	P04: Assessment of the clinical phenotype of *BAP1* germline whole gene and large deletions
	Branham, Kari, Ann Arbor, USA	P05: Suspicion for *ABCA4*-related retinal dystrophy: Clues beyond the typical Stargardt Phenotype
	Bryjova, Barbara, Giessen, Germany	P06: Misinterpretation of an OMD phenotype from a common sequence variation in *RP1L1* in a family with multiple sclerosis
	Ehrenberg, Miriam, Petach Tikva, Israel	P07: Double struggle
	Everett, Lesley, San Francisco, USA	P08: Ocular findings in two patients with vascular smooth muscle myopathy secondary to *ACTA2* mutations
	Ibanez Iv, Manuel Benjamin, Philadelphia, USA	P09: Stargardt misdiagnosis: How ocular genetics helps
	Kameya, Shuhei, Inzai, Japan	P10: A novel homozygous in-frame deletion of *GNAT1* gene cause golden discolouration of the fundus and reduced dark-adapted ERG similar to that of Oguchi disease in a Japanese family
	Majander, Anna, Helsinki, Finland	P11: Clinical characteristics of early onset retinal dystrophy in association with the *TULP1* c.148delG mutation
	Mauring, Laura, Strasbourg, France	P12: Alström syndrome with atypical retinal dystrophy and inheritance
	Prasov, Lev, Flint, USA	P13: Clinical and genetic features of Jalilli syndrome in a North American patient cohort
	Starosta, Daniela Aneta, Giessen, Germany	P14: Phenotype in five related patients with isolated optic nerve atrophy associated with a heterozygous mutation in the spastic paraplegia gene 7.
13.00 h – 14.30 h		**Lunch Break** **Poster Session**
14.30 h – 15.50 h	**3. Session**	**Stem Cells**
		Chair: Mike Karl, Eyal Banin
14.30 h	Bachmann, Björn, Cologne, Germany	T06: Treatment options for limbal stem cell deficiency in inhereted eye diseases

14.50 h	Karl, Mike, Dresden, Germany	T07: Towards modeling of neuronal and glial pathologies in retinal organoids
15.10 h	Banin, Eyal, Jerusalem, Israel	T08: Derivation of RPE cells from human embryonic stem cells (hESCs): The journey from basic research to clinical application
15.30 h	Battu, Rajani, Bangalore, India	O12: Differentiation and characterization of RPE from hiPSC and its subretinal transplantation in RCS rats
15.40 h	Galardi, Angela, Rome, Italy	O13: Proteomics profiling of retinoblastoma derived exosomes
15.50 h – 16.20 h		**Coffee Break** **Poster Session**
16.20 h – 17.05 h		**Franceschetti Medal & Lecture**
16.20 h	Albert Franceschetti, Lausanne, Switzerland	Introduction to the Franceschetti Medal & Lecture
16.35 h	Sieving, Paul, Bethesda, USA	L1: Clinical features and molecular basis of X-linked retinoschisis: From mechanism to therapy.
17.05 h – 18.35 h	**4. Session**	**Biomarkers for Substantiating Success in Treatment** *Chair: Tomas Aleman, Michael Hoffmann*
17.05 h	Aleman, Tomas, Philadelphia, USA	T09: AAV2-hCHM subretinal delivery to the macula in choroideremia: performance of outcome measures
17.25 h	Hoffmann, Michael, Magdeburg, Germany	T10: Plasticity and its limits - Cortical visual field representations in achromatopsia
17.45 h	Gocho, Kiyoko, Inzai, Japan	T11: High-resolution retinal imaging analysis in female carriers of choroideremia
18.05 h	Lima de Carvalho Jr, Jose Ronaldo, Recife, Brazil	O14: Multimodal imaging of patients with Best Vitelliform Macular Dystrophy (BVMD): a 4-year follow-up study.
18.15 h	Sallum, Juliana, Sao Paulo, Brazil	O15: Characterization of the Brazilian ARSACS phenotype: clinical, ophthalmological, neuroimaging, and genetic features of fourteen cases
18.25 h	Studer, Fouzia, Strasbourg, France	O16: Retinal implantation with Argus II artificial retina in 3 patients with Bardet-Biedl syndrome
		Associated Posters
	Tanrikulu, Özgün, Giessen, Germany	P15: Analysis of outer retinal layer alterations in patients with *RPE65* deficiency using Optical Coherence Tomography A-Scan-Analysis
19.30 h		**Gala Dinner** Restaurant Heyligenstaedt, Aulweg 41, 35392 Gießen

Friday **30.08.2019**

8.00 h		**Onsite Registration**
8.30 h – 10.00 h	**5. Session**	**Luxturna Therapy – Recent Developments** **Chair: Stephen Russell, Paul Sieving**
08.30 h	Leroy, Bart P, Ghent, Belgium	T12: voretigene neparvovec for *RPE65*-related inherited retinal dystrophies: The Philadelphia Experience
08.45 h	Drack, Arlene, Iowa City, USA	T13a: How long does gene therapy last? 4 Year followup and adult versus pediatric outcomes of phase 3 voretigene neparvovec trial in *RPE65*–associated LCA/inherited retinal

		disease T13b: Electrophysiology following subretinal treatment with voretigene neparvovec (Luxturna)
09.00 h	Simonelli, Francesca, Naples, Italy	T14: Our experience with gene therapy approaches for *RPE65* inherited retinal diseases
09.15 h	Lorenz, Birgit, Giessen, Germany	T15: Country specific problems to get started with EMA-approved gene therapy with Luxturna
09.30 h	Chung, Daniel C., Philadelphia, USA	T16: The post-authorization safety study of voretigene neparvovec-rzyl
09.45 h		**Panel Discussion**
10.00 h – 10.30 h		**Coffee Break** **Poster Session**
10.30 h – 11.15 h		**Ellsworth Lecture**
10.30 h	Francis Munier, Lausanne, Switzerland	Introduction to Ellsworth Lecture
10.45 h	Desjardins, Laurence, Rochefort En Yvelines, France	L2: Retinoblastoma around the world in 2019
11.15 h – 13.25 h	**6. Session**	**Precision Care for Children with Retinoblastoma** *Chair: Brenda Gallie, Dietmar Lohmann*
11.15 h	Di Giannatale, Angela, Rome, Italy	T17: Proteomics for biomarker identification in retinoblastoma liquid biopsy
11.35 h	Cole, Trevor, Birmingham, United Kingdom	T18: The current and future role of cell free DNA analysis in the management of retinoblastoma.
11.55 h	Gallie, Brenda, Toronto, Canada	T19: Disruptive innovations to reach precision retinoblastoma care
12.15 h	Soliman, Sameh, Toronto, Canada	T20: Optimizing focal laser photocoagulation therapy for retinoblastoma
12.35 h	Everett, Lesley, San Francisco, USA	O17: Retinoblastoma treatment in the age of intra-vitreal and intra-arterial chemotherapy: the UCSF experience
12.45 h	De Jong, Marcus, Amsterdam, Netherlands	O18: Screening for pineal trilateral retinoblastoma revisited: a meta-analysis
12.55 h	Menges, Julia, Essen, Germany	O19: A human organoid-based model for retinoblastoma
13.05 h	Tsygankov, Alexander, Moscow, Russian Federation	O20: New retinoblastoma cell culture establishment and drug resistance assessment
13.15 h	White, Jaclyn, Clayfield, Australia	O21: Delay in the diagnosis of retinoblastoma: the role of parents and practitioners
		Associated Posters
	Hattori, Hiroyoshi, Nagoya, Japan	P16: Bilateral retinoblastoma with 13q-syndrome in a patient carrying an X;13 balanced translocation without rearrangement of the RB1 gene
13.25 h – 14.05h		**Lunch Break** **Poster Session**
14.05 h – 14.45 h	**7. Session**	**Clinical Studies in Gene Therapy II** *Chair: Birgit Lorenz, Bart Leroy*
14.05 h	MacLaren, Robert, Oxford, UK	T21: Gene therapy for X-linked retinitis pigmentosa caused by mutations in RPGR
14.25 h	Russell, Stephen, Iowa City, USA	T22: Surgical challenges and outcomes with voretigene neparvovec (Luxturna)
		Treatment-associated Posters

	Alfarsi, Ammar, Muscat, Oman	P17: Intravitreal ranibizumab (Lucentis®) in the treatment of non-leaking macular cysts in retinal dystrophy
	Andrassi-Darida, Monika, Giessen, Germany	P18: Laser photocoagulation for hemorrhagic retinopathy in a newborn with Norrie disease
	Cui, Xuan, New York, USA	P19: Enhancing glycolytic metabolism with gene therapy and a small molecule drug attenuates neurodegeneration
	Reschke, Madlen, Essen, Germany	P20: Individual treatment of an infant with severe conjunctivitis lignosa (CL) and other systemic manifestations of plasminogen deficiency, caused by a compound mutation of the *PLG* gene
15.00 h		**Excursion by bus** Marburg
19.30 h		**Gala Dinner** Schloß Rauischholzhausen, Ebsdorfer Grund
Approx. 23.00 h		**Return to Giessen**

Saturday **31.08.2019**

8.00 h		**ISGEDR General assembly**
9.00 h – 10.30 h	**8. Session**	**Secondary Cancer and Survival in Retinoblastoma** *Chair: Petra Ketteler, Ashwin Reddy*
09.00 h	Ketteler, Petra, Essen, Germany	T23: The Impact of the Type Of Predisposing *RB1* Variants on the Incidence of Malignancies
09.20 h	Moll, Annette, Amsterdam, Netherlands	T24: Retinoblastoma and second primary malignancies: a Dutch overview and update
09.40 h	Lohmann, Dietmar, Essen, Germany	O22: Understanding and predicting tumor risk in heritable retinoblastoma
09.50 h	Abdel-Rahman, Mohamed, Columbus, USA	O23: Study of genetic predisposition to uveal melanoma
10.00 h	Reddy, M. Ashwin, London, United Kingdom	O24: Prognostic information for mosaic and high penetrant carriers of *RB1* mutations
10.10 h	Shah, Parag, Coimbatore, India	O25: Outcomes of *RB1* gene testing from blood samples of 113 retinoblastoma survivors and their families (398 in total) collected on a single day at Aravind Eye Hospital, Coimbatore, India.
10.20 h	van Hoefen Wijsard, Milo, Amsterdam, Netherlands	O26: Type of *RB1* mutation and age at diagnosis of familial retinoblastoma screened from birth.
		Associated Posters
	Janic, Ana, Toronto, Canada	P21: Patient-reported outcome measures for retinoblastoma: A scoping review
	Jansen, Robin, Amsterdam, Netherlands	P22: Should postlaminar optic nerve tumor invasion into the outer layers be considered a risk-factor for leptomeningeal spread of retinoblastoma? A case report and review of the literature
	Saakyan, Svetlana, Moscow, Russian Federation	P23: Long-term follow-up after retinoblastoma: secondary malignancy.
10.30 h – 11.00 h		**Coffee Break** **Poster Session**
11.00 h – 12.30 h	**9. Session**	**Patients in Focus**

		Chairs: Christina Fasser, Helene Dollfus
11.00 h	Dollfus, Helene, Strasbourg, France	T25: What could be the EYE-EYE role in rare eye diseases care in Europe?
11.15 h	Fujinami, Kaoru, Toyko, Japan	T26: Clinical and genetic characteristics of East Asian patients with inherited retinal disorders
11.30 h	Dimaras, Helen, Toronto, Canada	T27: Achieving meaningful patient research partnership: development of the Canadian Retinoblastoma Research Advisory Board
11.45 h	Gallie, Brenda, Toronto, Canada	T28: DEPICT HEALTH "full view for life" for circle of care including families will empower research
11.55 h	Badura, Franz, Amberg, Germany	T29: The PRO RETINA patient registry
12.05 h	Fasser, Christina, Zürich, Switzerland	T30: Leave no one behind –Patient's perspective
12.15 h		**Panel Discussion**
		Associated Posters
	Escher, Pascal, Bern, Switzerland	P24: The Swiss Registry of Rare Eye Diseases
	Nanos, Stephanie, Toronto, Canada	P25: The Canadian Retinoblastoma Research Registry
	Ristevski, Ivana, Toronto, Canada	P26: The top 10 retinoblastoma research priorities in canada as determined by patients, clinicians and researchers
12.30 h – 13.15 h		**François-Lecture**
12.30 h	Bart Leroy, Ghent, Belgium	Introduction to the François-Lecture
12.45 h	Lorenz, Birgit, Giessen, Germany	L3: Biomarkers in IRDs: scientifically valid – clinically relevant
13.15 h – 14.00 h		**Lunch Break**
		Poster Session
		Microphthalmia – Anophthalmia – Coloboma – Developmental Failures
	Bacci, Giacomo, Firenze, Italy	P27: Novel mutations in *MFRP* and *PRSS56* genes associated to posterior microphthalmos
	Balikova, Irina, Gent, Belgium	P28: Mendeliome in patients with microphthalmia, anophthalmia and coloboma – results and challenges
	Friedburg, Christoph, Giessen, Germany	P29: Early onset severe retinal dystrophy with irido-chorioretinal coloboma with optic disc dysplasia and macular hypoplasia in one eye due to a heterozygous *GDF6*-mutation
	Pfäffli, Oliver, Zürich, Switzerland	P30: Genotype phenotype correlation in a case series of nanophthalmos
	Ramzan, Khushnooda, Riyadh, Saudi Arabia	P31: Novel phenotype-genotype correlation with *PEX6* gene in Saudi patients with Heimler syndrome
	Takitani, Guilherme, São Paulo - Sp, Brazil	P32: Microcephaly and chorioretinopathy associated with *TUBGCP4* mutation
14.00 h – 15.00 h	**10. Session**	**Understanding Treatment Effects from Natural History Studies**
		Chair: Elias Traboulsi, Hendrik Scholl
14.00 h	Scholl, Hendrik, basel, Switzerland, Switzerland	T31: Natural history of the progression of atrophy secondary to Stargardt disease (ProgStar) study
14.20 h	Hahn, Leo, Amsterdam,	O27: Long-term follow-up, phenotypic and genetic

	Netherlands	spectrum of patients with juvenile X-linked retinoschisis in the Netherlands
14.30 h	Nguyen, Xuan-Thanh-An, Leiden, Netherlands	O28: The disease course of rhodopsin (RHO)-associated retinitis pigmentosa (RP): a follow-up study
14.40 h	Senatore, Alfonso, Durham, USA	O29: “Further evaluation of a simple perimetric approach to the differential diagnosis between blue cone monochromacy (BCM) and achromatopsia (ACHM)”
14.50 h	Testa, Francesco, Napoli, Italy	O30: Longitudinal natural history study in patients with Retinitis Pigmentosa in preparation for gene therapy clinical trials
15.00 h – 15.30 h		**Coffee Break**
		Poster Session
		Glaucoma
	Hosono, Katsuhiro, Hamamatsu, Japan	P33: A case of childhood glaucoma with a combined partial monosomy 6p25 and partial trisomy 18p11 due to an unbalanced translocation
	Lang, Elena, Zurich, Switzerland	P34: Genotypic and phenotypic characterization of childhood glaucoma
	Naruse, Sho, Kitakyushu City, Japan	P35: Development of glaucoma after early cataract surgery in case of oculo-facio-cardio-dental syndrome
	Pisuchpen, Phattrawan, Philadelphia, USA	P36: The Robison D. Harley, MD Childhood Glaucoma Research Network (CGRN) International Pediatric Glaucoma Registry
		Foveal Hypoplasia
	Chaudhuri, Zia, New Delhi, India	P37: Novel variant in SLC38A8 gene segregating with foveal hypoplasia in an autosomal recessive South Asian family
	Dumitrescu, Alina, Iowa City, USA	P38: Genotype-Phenotype Correlation in Patients with Albinism
	Kondo, Hiroyuki, Kitakyushu, Japan	P39: Correlation between genotype-phenotype in patients with autosomal dominant idiopathic foveal hypoplasia associated with *PAX6* mutations
	Kröll-Hermi, Ariane, Strasbourg, France	P40: Zebrafish, as a useful model to validate human eye candidate diseases genes.
15.30 h – 16.50 h	**11. Session**	**Gene and Cell based Therapies**
		Chair: Knut Stieger, Volker Busskamp
15.30 h	Ader, Marius, Dresden, Germany	T32: Photoreceptor transplantation into the mammalian retina
15.50 h	Stieger, Knut, Giessen, Germany	T33: DNA repair mechanisms in photoreceptors
16.10 h	Busskamp, Volker, Dresden, Germany	T34: Forward programming of human stem cells to photoreceptors
16.30 h	Müller, Brigitte, Giessen, Germany	O31: DNA repair after ISce-I mediated DSB in photoreceptors and RPE cells following AAV mediated gene transfer in vivo
16.40 h	Wimmer, Tobias, Giessen, Germany	O32: A bioluminescence resonance energy transfer based sensor for the precise determination of non-homologous end joining DNA repair events
		Associated Posters
	Weller, Maria, Giessen, Germany	P42: First steps to a MMEJ genome editing approach correcting *CLN3*/Cln3$^{deltaEx7/8}$

16.50 h	**Farewell**
17.00 h	**End of conference**