**Thursday**

| 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 |
| 09.00 h | Performing And Communicating Molecular Diagnostics |
| Chair: Hanno Bolz, David Mackey |
| 09.00 h Bolz, Hanno Jörn, Frankfurt, Germany |
| 09.20 h Mackey, David, Subiaco, Australia |
| 09.40 h Capasso, Jenina, Philadelphia, USA |
| 10.00 h Arno, Gavin, London, United Kingdom |
| 10.10 h Debenedectis, Meghan, Cleveland, USA |
| 10.20 h Hufnagel, Robert, Bethesda, USA |
| 10.30 h Kellner, Ulrich, Siegburg, Germany |
| 10.40 h Wells, Kirsty, Helsinki, Finland |
| T01: NGS: Diagnostic opportunities and challenges of gene panel, whole-exome and whole-genome sequencing |
| T02: Predictive testing in glaucoma |
| T03: We need more genetic counselors! |

| 10.00 h | O01: Interrogation of the 100,000 genomes project ophthalmic disease cohort reveals novel genes, new associations and previously undetectable mutations |
| 10.10 h | O02: The value of CNV analysis for inherited retinal diseases |
| 10.20 h | O03: Modeling gene constraint in disease populations for clinical molecular diagnostics |
| 10.30 h | O04: DNA testing in a series of 944 patients with inherited retinal dystrophies from a single german reference center |
| 10.40 h | 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 |
| Pantrangi, Madhulatha, Marshfield, USA |
| Chan, Choi Mun, Singapore, Singapore |

**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


12.40 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

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 inherited 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 neaparvovec 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 neaparvovec 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-ryzl

Panel Discussion

10.00 h – 10.30 h
Coffee Break
Poster Session

10.30 h – 11.15 h
Ellsworth Lecture
Introduction to Ellsworth Lecture

11.15 h – 13.25 h  6. Session

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

14.05 h  MacLaren, Robert, Oxford, UK
Clinical Studies in Gene Therapy II
Chair: Birgit Lorenz, Bart Leroy
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
Andrassi-Darida, Monika, Giessen, Germany
Cui, Xuan, New York, USA
Reschke, Madlen, Essen, Germany

P1: Intravitreal ranibizumab (Lucentis®) in the treatment of non-leaking macular cysts in retinal dystrophy
P17: Intravitreal ranibizumab (Lucentis®) in the treatment of non-leaking macular cysts in retinal dystrophy
P18: Laser photoagulation for hemorrhagic retinopathy in a newborn with Norrie disease
P19: Enhancing glycolytic metabolism with gene therapy and a small molecule drug attenuates neurodegeneration
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
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
T23: The Impact of the Type Of Predisposing RB1 Variants on the Incidence of Malignancies
T24: Retinoblastoma and second primary malignancies: a Dutch overview and update
O22: Understanding and predicting tumor risk in heritable retinoblastoma
O23: Study of genetic predisposition to uveal melanoma
O24: Prognostic information for mosaic and high penetrant carriers of RB1 mutations
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.
O26: Type of RB1 mutation and age at diagnosis of familial retinoblastoma screened from birth.

Associated Posters
P21: Patient-reported outcome measures for retinoblastoma: A scoping review
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

10.30 h – 11.00 h
Coffee Break
Poster Session

11.00 h – 12.30 h 9. Session
Patients in Focus
11.00 h  Dollfus, Helene, Strasbourg, France
11.15 h  Fujinami, Kaoru, Toyko, Japan
11.30 h  Dimaras, Helen, Toronto, Canada
11.45 h  Gallie, Brenda, Toronto, Canada
11.55 h  Badura, Franz, Amberg, Germany
12.05 h  Fasser, Christina, Zürich, Switzerland
12.15 h  Panel Discussion

**Chairs: Christina Fasser, Helene Dollfus**

T25: What could be the EYE-EYE role in rare eye diseases care in Europe?
T26: Clinical and genetic characteristics of East Asian patients with inherited retinal disorders
T27: Achieving meaningful patient research partnership: development of the Canadian Retinoblastoma Research Advisory Board
T28: DEPICT HEALTH "full view for life" for circle of care including families will empower research
T29: The PRO RETINA patient registry
T30: Leave no one behind – Patient's perspective

**Associated Posters**

- Escher, Pascal, Bern, Switzerland
- Nanos, Stephanie, Toronto, Canada
- Ristevski, Ivana, Toronto, Canada

P24: The Swiss Registry of Rare Eye Diseases
P25: The Canadian Retinoblastoma Research Registry
P26: The top 10 retinoblastoma research priorities in canada as determined by patients, clinicians and researchers

**12.30 h – 13.15 h**

- Bart Leroy, Ghent, Belgium
- Lorenz, Birgit, Giessen, Germany

**François-Lecture**

Introduction to the François-Lecture
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
- Balikova, Irina, Gent, Belgium
- Friedburg, Christoph, Giessen, Germany
- Pfäffli, Oliver, Zürich, Switzerland
- Ramzan, Khushnooda, Riyadh, Saudi Arabia
- Takitani, Guilherme, São Paulo - Sp, Brazil

P27: Novel mutations in MFRP and PRSS56 genes associated to posterior microphthalmos
P28: Mendelrome in patients with microphthalmia, anophthalmia and coloboma – results and challenges
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
P30: Genotype phenotype correlation in a case series of nanophthalmos
P31: Novel phenotype-genotype correlation with PEX6 gene in Saudi patients with Heimler syndrome
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

T31: Natural history of the progression of atrophy secondary to Stargardt disease (ProgStar) study
O27: Long-term follow-up, phenotypic and genetic
Netherlands

14.30 h Nguyen, Xuan-Thanh-An, Leiden, Netherlands

14.40 h Senatore, Alfonso, Durham, USA

14.50 h Testa, Francesco, Napoli, Italy

15.00 h – 15.30 h

Coffee Break
Poster Session

Glaucoma

Hosono, Katsuhiro, Hamamatsu, Japan

Lang, Elena, Zurich, Switzerland

Naruse, Sho, Kitakyushu City, Japan

Pisuchpen, Phattrawan, Philadelphia, USA

15.30 h – 16.50 h 11. Session

Gene and Cell based Therapies
Chair: Knut Stieger, Volker Busskamp

Ader, Marius, Dresden, Germany

Stieger, Knut, Giessen, Germany

Busskamp, Volker, Dresden, Germany

Müller, Brigitte, Giessen, Germany

Wimmer, Tobias, Giessen, Germany

Weller, Maria, Giessen, Germany

16.10 h

T32: Photoreceptor transplantation into the mammalian retina

T33: DNA repair mechanisms in photoreceptors

T34: Forward programming of human stem cells to photoreceptors

O31: DNA repair after ISceI mediated DSB in photoreceptors and RPE cells following AAV mediated gene transfer in vivo

O32: A bioluminescence resonance energy transfer based sensor for the precise determination of non-homologous end joining DNA repair events

Associated Posters

P42: First steps to a MMEJ genome editing approach correcting CLN3/Cln3deltaEx7/8
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