ISGEDR 2025 Oslo
held jointly with EURBG

Program

Thursday, 11 September
National Museum Oslo

Start	End	Session	Moderator	Presenter	Paper title	Presentation no.
08:30	09:00	REGISTRATION				
9:00	10:15	SESSION 1: Opening Ceremony				
9:00	9:05	Welcome	Josephine Prener Holtan			
9:05	9:15	Opening Ceremony	Bart Leroy	Francis Munier		N/A
9:15	10:15	Introduction & Franceschetti Lecture	Elias Traboulsi	Graeme Black	"The Guide is definitive. Reality is frequently inaccurate." - Douglas Adams, *The Hitchhiker's Guide to the Galaxy*	1-101
10:15	11:02	SESSION 2: Award & Travel Grant Recipients				
10:15	10:17	Introduction	Brian P Brooks		Elias I Traboulsi Distinguished Trainee Research Award	N/A
10:17	10:26	Elias I Traboulsi Distinguished Trainee Research Award	Elias Traboulsi & Josephine Prener Holtan	Leyla Yavuz Saricay	Refractive Errors in Patients with Bardet Biedl Syndrome	1-102
10:26	10:35	ISGEDR 2025 Travel Grant		Caroline Atef Tawfik	Clinical and Genetic Characterization of *IMPG2*-Associated Retinopathy in an Egyptian Cohort	1-103
10:35	10:44			Goura Chattannavar	Homozygous R383H variant in *IDUA* gene causing Pericentric Retinitis Pigmentosa in attenuated Mucopolysaccharidosis Type I	1-104
10:44	10:53			Piet Noë	Retinoblastoma treatment at a charitable eye hospital in Rwanda	1-105
10:53	11:02	Ophthalmic Genetics I		Mette Bertelsen	A nationwide overview of the clinical and genetic landscape of inherited ocular disorders in Denmark	1-106
11:02	11:30	BREAK				
11:30	12:30	SESSION 3: Retinal Disorders & Ocular Development				
11:30	11:40	Ophthalmic Genetics I	Bart Leroy & Evangelia Panagiotou	Hélène Dollfus	Consensus-Based Revision of Diagnostic Criteria and Management for Bardet-Biedl Syndrome: Recommendations from European Reference Networks	1-107
11:40	11:50			Arif O. Khan	A homozygous *NRL* variant (c.339C>G; p.Try113*) underlies enhanced-S-cone syndrome in the United Arab Emirates and is associated with an electronegative electroretinogram	1-108
11:50	12:00			Beau J. Fenner	How Does the Diagnosis of Inherited Retinal Disease Influence Reproductive Decision-Making in Singapore?	1-109
12:00	12:10			Dorothy Wang	Clinical and Genetic Characterization of Inherited Retinal Diseases in a Diverse North American Cohort of Over 1,800 Cases	1-110
12:10	12:20			Xènia Ferrer-Cortès	New insights on the genetic basis underlying hereditary high myopia in a cohort of more than 100 Spanish families	1-111
12:20	12:30			Nicola K Ragge	Developmental eye anomalies: new genes, mechanisms and approaches to improve genetic diagnoses	1-112
12:30	13:30	LUNCH Meet the sponsors				

Start	End	Session	Moderator	Presenter	Paper title	Presentation no.
13:30	14:30	SESSION 4: Case Reports Retinal Disorders				
13:30	13:36	Case reports	Elias Traboulsi & Caroline Atef Tawfik	Brian P. Brooks	Variants in *NR6A1* cause a novel Oculo Vertebral Renal (OVR) syndrome	I-113
13:36	13:42			Ulrika Kjellström	A case series: The description of a family with autosomal dominant *KCNJ13* associated retinal dystrophy and a novel phenotype	I-114
13:42	13:48			Ragnhild Wivestad Jansson	A novel deletion in 3'UTR of the retina-specific isoform of *RPGR* leads to reduced expression of *RPGR* in a family with X-linked retinitis pigmentosa.	I-115
13:48	13:54			Erlend C. S. Landsend	Substantial difference in ocular phenotype and follow-up of two patients with *KIF11*-mutation	I-116
13:54	14:00			Dong Hyun Jo	Clinical Characterization of *NMNAT1*-Associated Leber Congenital Amaurosis in a Korean Cohort	I-117
14:00	14:06			Laura Mauring	Cone-Rod Dystrophy due to a Homozygous *KIZ* Variant	I-118
14:06	14:12			Miriam Ehrenberg	Patchy perifoveal chorioretinal atrophy related to a novel gene: *C19ORF44*	I-119
14:12	14:18			Evangelia Panagiotou	A rare case of *RAB28*-related retinal disease due to complete uniparental isodisomy of chromosome 4	I-120
14:18	14:24			Suma P. Shankar	Micro RNA 204 (*MIR204*) as a novel cause of persistent pupillary membrane, high myopia, coloboma and lens opacity: a case study	I-121
14:24	14:30			WITHDRAWN		I-122
14:30	15:30	SESSION 5: Case Reports Miscellaneous & Retinoblastoma				
14:30	14:36	Case reports	Arif O. Khan & Alessia Amato	Lucianna De Luca	*ACO2*-Related Retinal Disease: Expanding the Phenotypic Spectrum Beyond Optic Neuropathy	I-123
14:36	14:42			Michelle Lingao	First case report of Wolfram Syndrome type 1 in the Philippines: Diagnostic challenges and discovery of a novel Exon 3 *WFS1* heterozygous nonsense mutation	I-124
14:42	14:48			Alena Egense	*NBAS*-related short stature, optic nerve atrophy, and Pelger-Huet anomaly (SOPH) presenting as early-onset optic atrophy	I-125
14:48	14:54			Karthikeyan Arcot Sadagopan	Microspherophakia, inguinal hernia, tortuous retinal vessels & facial dysmorphism as the presenting features in Six Indian Asian Children with Traboulsi syndrome and report of a novel pathogenic variant in the *ASPH* gene.	I-126
14:54	15:00	Case reports	Arif O. Khan & Alessia Amato	Annamari Immonen	Phenotypically Mild Congenital Stromal Corneal Dystrophy in a Finnish Family Caused by a Novel Missense Variant c.1036T>C in the Decorin Gene	I-127
15:00	15:06			Ana Ćurić	Giant Cyst of Dermis-Fat Graft Following Retinoblastoma Treatment Complicated with MRSA	I-128
15:06	15:12			Khitam Fakhir	An unexpected response of orbital retinoblastoma to inconsistent systemic chemotherapy alone. A case report	I-129
15:12	15:18			Hiroyoshi Hattori	Three Case Reports of Retinoblastoma with Optic Nerve and CNS Invasion Treated with Multimodal Therapy Including Intrathecal and High-Dose Chemotherapy	I-130
15:18	15:24			Małgorzata Danowska	Presumed bilateral ciliary body medulloepithelioma in a child with DICER1 syndrome and pleuropulmonary blastoma	I-131
15:24	15:30			Ashwin Mallipatna	Case Report: Axenfeld Rieger Syndrome in Patients with 13q Deletion Syndrome	I-132
15:30	15:30	Close	Josephine Prener Holtan			N/A
15:30	17:00	Social at the Museum				
17:00	19:00	Free time				
19:00	22:00	Mingle at Barcode Street Food, Bjørvika (own expense)				

Auditorium 1

Please note that parallel sessions are being conducted throughout the day!

Start	End	Session	Moderator	Presenter	Paper title	Presentation no.
8:30	9:00	REGISTRATION Poster set up				
9:00	10:00	SESSION 1: Ellsworth Lecture				
9:00	10:00	Ellsworth Lecture	Francis Munier	Paula Schaiquevich	Two decades of translational research in retinoblastoma	2-101
10:00	11:02	SESSION 2: Retinoblastoma Diagnosis & Awareness				
10:00	10:20	Judith Kingston Lecture	Ashwin Reddy	Maja Beck-Popovic	Retinoblastoma: challenges of a rare disease	2-102
10:20	10:27	Retinoblastoma 1	Doris Hadjistilianou & Helen Dimaras	Liesbeth Gardoen	Guidelines on imaging of retinoblastoma : a 2025 update on behalf of the European Retinoblastoma Imaging Collaboration and the European Retinoblastoma Group	2-103
10:27	10:34			Carme Julià	Leukocheck : a mobile app for the early detection of retinoblastoma	2-104
10:34	10:41			Helen Dimaras	Uncertain heritable risk in retinoblastoma : Classification of genetic testing results for precision risk stratification	2-105
10:41	10:48			Ashwin Mallipatna	Construction and validation of 3D-printed eyeball models in red reflex training of medical students	2-106
10:48	10:55			Doris Hadjistilianou	Bilateral retinoblastoma and inner ear common cavity malformation : A novel association ?	2-107
10:55	11:02			Rebecca Clark	Novel, deep intronic *RB1* variant exhibiting incomplete penetrance and a parent-of-origin effect	2-108
11:02	11:30	BREAK Poster Session - Retinoblastoma Sessions 1 to 4 and Ophthalmic Genetics Session 1				
11:30	12:30	SESSION 3: Ophthalmic Genetics - General Category				
11:30	11:40	Ophthalmic Genetics 2	Nicola K Ragge & Line Kessel	Alex V. Levin	The Role of Orbis in Enhancing Worldwide Education in Ocular Genetics	2-109
11:40	11:48			Hannah L. Scanga	Quantifying the Clinical Utility of Genetic Testing for Inherited Retinal Dystrophies to Inform Genetic Counseling	2-110
11:48	11:56			Sandra Staffieri	There's more to Leber Hereditary Optic Neuropathy than vision impairment: listening to everyone's voice.	2-111
11:56	12:04			David A Mackey	Detection of low-penetrance Leber Hereditary Optic Neuropathy (LHON) mtDNA variants in the general population.	2-112
12:04	12:12			Faeeqah Almhmoudi	Posterior Microphthalmia related to *PRSS56* Variants in a Saudi Cohort: A Longitudinal Study of Visual Function	2-113
12:12	12:20			Petra Ketteler	Genetic predisposition to melanoma of the choroid and ciliary body	2-114
12:20	12:28			Mohamed H. Abdel-Rahman	Genetic Predisposition to uveal melanoma	2-115
12:30	13:30	LUNCH Meet the sponsors 12:45 ISGEDR Assembly (Room 1)				

Start	End	Session	Moderator	Presenter	Paper title	
13:30	14:30	SESSION 4: Mini-symposium - Therapies for IRDs I				
13:30	13:45	Mini-symposium: Therapeutic innovations in IRD's: Gene-specific	Mark Pennesi	Bart Leroy	Overview of therapeutic innovations in IRDs: Gene-specific	2-116
13:45	14:30	Panel Discussion		Panelists: Bart Leroy Aniz Girach Zuhul Butuner Jayashree Sahni Tomas Aleman Martin Smedstad Meghan J DeBenedictis		N/A
14:30	15:30	SESSION 5: Mini-symposium - Therapies for IRDs II				
14:30	14:45	Mini-symposium: Therapeutic innovations in IRD's: Gene-agnostic	Mark Pennesi	Isabelle Audo	Overview of therapeutic innovations in IRDs: Gene-agnostic	2-117
14:45	15:30	Panel Discussion		Panelists: Katherine High Daniel Chung Peter Francis Kali Stasi Isabelle Audo Bart Leroy Martin Smedstad Meghan J DeBenedictis		N/A
15:30	16:00	BREAK Poster Session - Retinoblastoma 1 to 4 and Ophthalmic Genetics 1				
16:00	17:30	SESSION 6: IRD - Gene Therapy, Natural history & Outcome Measures				
16:00	16:10	Ophthalmic Genetics 3	Ulrika Kjellström & Jasmina Cehajic-Kapetanovic	Ragnheiður Bragadóttir	Special Lecture: Gene Therapy: From Experimental Research to Clinical Application	2-118
16:10	16:18			Jasmina Cehajic-Kapetanovic	Special lecture: Gene therapy delivery	2-119
16:18	16:26			Brittni Scruggs	Retinal Gene Therapy Using Epiretinal AAV-Containing Fibrin Hydrogel Implants	2-120
16:26	16:34			Leonardo Colombo	Characterizing *PRPF31* -related Retinal Dystrophy: Clinical Insights from Baseline Data in a Natural History Study	2-121
16:34	16:42			Kiyoko Gocho	Progression Assessment with OCT and Adaptive Optics: A Subset Analysis of the KEYS study	2-122
16:42	16:50			Kirk Stephenson	Quantitative Choroidal Analysis in Molecularly Characterized Retinitis Pigmentosa	2-123
16:50	16:58			Peter Francis	Efficacy Testing Facility for Low Vision Patients, the Vision Research and Assessment Institute "VRAI"	2-124
16:58	17:06			Veronika Vaclavik	*RP2* -Associated Rod Cone Dystrophy in Females Carriers: A Spectrum of Severity and follow up of 10 years	2-125
17:06	17:14			Brooke Koritala	Eye-Related Quality of Life in Children with Inherited Retinal Diseases Using PedEyeQ	2-126
17:14	17:22			Said El Shamieh	Copy-number variation significantly contributes to the pathogenicity of inherited retinal diseases in Lebanon	2-127
17:22	17:30	Close	Josephine Prener Holtan			N/A
17:30	22:00	Dinner at Sporten *Buses will transport attendees from the University of Oslo to the venue and back to the city center.*				

Auditorium 2

Please note that parallel sessions are being conducted throughout the day!

Start	End	Session	Moderator	Presenter	Paper title	Presentation no.
8:30	9:00	REGISTRATION				
9:00	10:00	No sessions				
10:00	12:30	SESSION 1: Electrophysiology Workshop				
10:00	10:15	Electrophysiology Workshop endorsed by ISCEV (+ break-our room 2)	Josephine Prener Holtan	Bart Leroy	Introduction to electrophysiology techniques (generalised response; ffERG, flash VEP, FST)	2-201
10:15	10:30			Carlos E. Mendoza-Santiesteban	Introduction to electrophysiology techniques (localized response; mfERG, Pattern ERG, pattern VEP)	2-202
10:30	10:50			Arlene Drack	Introductory cases pediatrics	2-203
10:50	11:00			Juliana Maria Ferraz Sallum	Introductory cases adults	2-204
11:00	12:00			Diagnosys/ Roland Consult/LKC	Practical demonstration	2-205
			Arlene Drack	N/A	WITHDRAWN	2-206
12:00	12:10			Bart Leroy	Challenging case 1	2-207
12:10	12:20			Carlos E. Mendoza-Santiesteban	Challenging case 2	2-208
12:20	12:30			Alina V Dumitrescu	Challenging case 3	2-209
12:30	13:30	LUNCH Meet the sponsors 12:45 ISGEDR Assembly (Room1)				
13:30	14:24	SESSION 2: Retinoblastoma Treatment				
13:30	13:39	Retinoblastoma 2	Petra Ketteler & Ashwin Reddy	David H. Abramson (VIRTUAL)	High dose Topotecan (90-180µG) for recurrent retinal and subretinal retinoblastoma	2-210
13:39	13:48			Sam Gurney	High dose intravitreal topotecan; a new therapy for retinal disease	2-211
13:48	13:57			Francis Munier	High dose intravitreal Topotecan monotherapy for aqueous seeding, subretinal seeding and retinal relapse	2-212
13:57	14:06			Joe Abbott	Intra-arterial chemotherapy: comparing techniques, does safety come at the cost of efficacy?	2-213
14:06	14:15			Ida Russo	Early results of the national AIEOP RTB018 protocol	2-214
14:15	14:24			Christina Stathopoulos	Risk factors for rhegmatogenous retinal detachment after intraarterial chemotherapy in endophytic retinoblastoma	2-215
14:24	15:30	SESSION 3: Retinoblastoma Metastatic Disease & Quality of Life				
14:24	14:32	Retinoblastoma 3	Livia Lumbroso-Le Rouic & Ida Russo	Hind M Alkatan	Adjuvant chemotherapy in treating retinoblastoma: Validation and controversies	2-216
14:32	14:40			Ida Russo	Long-Term (>5-Year) remission and survival after immunotherapy with anti-GD2 monoclonal antibody dinutuximab Beta following autologous hematopoietic stem cell transplantation in a child with metastatic retinoblastoma	2-217
14:40	14:48			Yan Honggai	Retinoblastoma patients died without high pathology Risk Factors	2-218
14:48	14:56			Ashwin Reddy	EURBG survey on mortality at Retinoblastoma Centres in Europe 2012-2022 : Part 1 metastatic disease from retinoblastoma	2-219
14:56	15:04			Livia Lumbroso-Le Rouic	EURBG survey on mortality at retinoblastoma centres in Europe 2012-2022 : Part 2 non metastatic deaths	2-220
15:04	15:12		Annette Moll & Marlies Hummelen	Emma Hughes	Supporting the psychosocial needs throughout the patient journey – The role of the play specialist.	2-221
15:12	15:20			Annette Moll	Visual outcome of retinoblastoma patients in the Netherlands : a cohort study	2-222
15:20	15:28			Anne Vestli	Retinoblastoma in Norway : Health-related quality of life and late effects	2-223

15:30	16:00	BREAK Poster Session - Retinoblastoma Sessions 1 to 4 and Ophthalmic Genetics Session 1			
16:00	17:12	SESSION 4: Retinoblastoma Liquid Biopsy, Biomarkers & Research			
16:00	16:08	Retinoblastoma 4	Amy Gerrish & Manoj Parulekar	Liya Xu (VIRTUAL)	Extracellular Vesicles in the Aqueous Humor: A Novel Avenue for Retinoblastoma Research and Clinical Management	2-225
16:08	16:16			Claudia Román-Montañana	Clinical variables associated with cell-free DNA yield obtained by anterior chamber tap in retinoblastoma	2-226
16:16	16:24			Alexandre Matet	Aqueous humor cfDNA exploration in retinoblastoma can be superior to tumor DNA analysis: a case series	2-227
16:24	16:32			Amy Gerrish	Genetic diagnosis of retinoblastoma via post chemotherapy anterior chamber tap	2-228
				N/A	WITHDRAWN	2-229
16:32	16:40			Tatsiana Ryl	Collaborative study to identify Biomarkers to adjust treatment intensity for children with Retinoblastoma (CoBioRB) - A pilot study for the multicentre clinical trial EURBG2	2-230
16:40	16:48			Lisa Golmard	Prognostic value of molecular markers in unilateral retinoblastoma treated by first-line enucleation	2-231
16:48	16:56			Luigi Mazzeo	Mapping Retinoblastoma Heterogeneity with Spatially Resolved Transcriptomics	2-232
16:56	17:04			Sima Das	Circulating cell-free tumor DNA-Based Liquid Biopsy of Cerebrospinal Fluid (CSF) in retinoblastoma	2-233
17:04	17:12			Sima Das	Spatial Transcriptomics to Identify Differentially Expressed Genes in high-risk factor-positive Retinoblastoma versus Non-Aggressive Disease	2-234
17:12	17:20	Close	Josephine Prener Holtan			N/A
17:30	22:00	Dinner at Sporten *Buses will transport attendees from the University of Oslo to the venue and back to the city center.*				

Auditorium 1

Please note that parallel sessions are being conducted throughout the day!

Start	End	Session	Moderator	Presenter	Paper title	Presentation no.
8:30	9:00	REGISTRATION				
9:00	10:30	SESSION 1: Mini-symposium - Corneal Genetics; Diagnosis and Management				
9:00	10:30	Mini-symposium: Corneal Genetics; Diagnosis and Management	Francis Munier & Graeme Black	Alice Davidson	Novel genes and disease mechanisms cornea dystrophies	3-101
				Andrea L. Vincent	Innovations in corneal dystrophy treatments	3-102
				Dimitri Roels	Syndromic diseases with corneal manifestations	3-103
				N/A	WITHDRAWN	3-104
				Liv Drolsum	Current clinical management of corneal dystrophies	3-105
10:30	10:58	SESSION 2: Anterior Segment				
10:30	10:37	Ophthalmic Genetics 4	Dimitri Roels & Meghan J DeBenedictis	Brenda L. Bohnsack	Purpose: To investigate genetics and outcomes in Axenfeld-Rieger Syndrome (ARS)	3-106
10:37	10:44			Andy Drackley	Updates on the ClinGen Pediatric Cataract VCEP's CYP27A1 Guidelines Development	3-107
10:44	10:51			Lev Prasov	*RIG1* variants cause a treatable glaucoma in humans and mice	3-108
10:51	10:58			Meghal Gagrani	Phenotypic and genotypic characterization of Posterior polymorphous corneal dystrophy in children : a 13 year clinical review	3-109
11:00	11:30	BREAK Poster Session - Ophthalmic Genetics 2 to 7				
11:30	12:30	SESSION 3: Francois Lecture				
11:30	12:30	Francois Lecture	Bart Leroy	Irene H. Maumenee	Six decades of ophthalmic genetics	3-110
12:30	13:30	LUNCH Meet the sponsors				
13:30	14:50	SESSION 4: Treatment for IRDs				
13:30	13:40	Ophthalmic Genetics 5	Isabelle Audo & Leonardo Colombo	Elias Traboulsi	Oral Gildeuretinol Slows Disease Progression in Early Stargardt Disease: Updates from the TEASE-3 Study	3-111
13:40	13:50			Arlene Drack	The effect of light exposure on schisis in X-linked retinoschisis	3-112
13:50	14:00			Tomas S. Aleman	Recovery of Cone-Mediated Vision in Severe Ciliopathies after Gene Therapy: One Year Results of a Phase I/II Trial for LCA5-LCA	3-113
14:00	14:10			Valentina Di Iorio	LUCE-1: a phase 1/2, dose escalation, safety and efficacy study of administration of AAVB-081 gene therapy in Usher Syndrome type 1B (USH1B)	3-114
14:10	14:20			Alina V Dumitrescu	Intravitreal Enzyme Replacement Therapy for Children with Neuronal Ceroid Lipofuscinosis Type 2, The University of Iowa experience	3-115
14:20	14:30			Josephine Prener Holtan	A Phase 1B Multiple Ascending Dose Study of VP-001; a peptide conjugate of oligonucleotide designed to treat *PRPF31* -related Retinitis Pigmentosa	3-116
14:30	14:40			Mark Pennesi	Subretinal gene therapy laru-zova for X-linked retinitis pigmentosa (XLRP): Phase 2 DAWN Trial, preliminary month 9+ results	3-117
14:40	14:50			Bart Leroy	RNA-based therapy in Leber congenital amaurosis type 10 (LCA10): key lessons learned from the randomized double-masked, sham-controlled Phase 3 study of sepofarsen	3-118

Start	End	Session	Moderator	Presenter	Paper title	Presentation no.
14:50	15:30	SESSION 5: Retinal Dystrophies & Electrophysiology				
14:50	14:58	Ophthalmic Genetics 6	Ragnheidur Bragadottir & Beau Fenner	Rachel Huckfeldt	Expanding the genetic understanding of retinal dystrophies and sensorineural hearing loss	3-119
14:58	15:06			Virginie MM Buhler	Genetic landscape of inherited retinal diseases in Switzerland: identification of prevalent pathogenic heterozygous variant causing autosomal dominant IRDs	3-120
15:06	15:14			Andrea L. Vincent	Genetic Drift and Founder Effects in *CYP4V2* and *PRPH2* -associated Inherited Retinal Disease Among Pasifika in Oceania	3-121
15:14	15:22			Lotta Gränse	A case series: Multifocal Electroretinography (mfERG) for evaluation of macular function in children under four	3-122
15:22	15:30			Alessio Amato	Negative Electroretinograms Beyond Congenital Stationary Night Blindness: Broadening the Differential Diagnosis of Post-Phototransduction Inherited Retinal Diseases	3-123
15:30	15:38			Juliana Maria Ferraz Sallum	Microperimetry, a valuable tool for assessing changes in visual function following voretigene neparvovec-rzyl gene therapy	3-123-2
15:30	16:00	BREAK Poster Session - Ophthalmic Genetics Session 2 to 7				
16:00	17:30	SESSION 6: IRDs - Mechanisms & Treatments				
16:00	16:10	Ophthalmic Genetics 7	Arlene Drack & Rola Ba-Abbad	Magnar Bjørås	Special lecture: Disease modelling and preclinical testing in retinal organoids derived from Childhood dementia *CLN3* patients	3-124
16:10	16:18			Arlene Drack	Subretinal gene therapy rescues cones >rods in a mouse model of Bardet-Biedl Syndrome Type 10	3-125
16:18	16:26			Carlotta Librasi	A novel RNA therapeutic platform for Retinitis Pigmentosa 11	3-126
16:26	16:34			Maya Helms	A murine model of microcephaly and chorioretinopathy reveals the importance of *TUBGCP4* in vascular development	3-127
16:34	16:42			Paula Gaudó	Deep characterization of mutant and CRISPR-corrected *PDE6A* and *PDE6C* retinal organoids derived from IRD patients	3-128
16:42	16:50			Pascal Escher	To identify molecular mechanisms underlying *NR2E3* -linked retinal degenerations in murine knock-in models	3-129
16:50	16:58			Rola Ba-Abbad	Clinical and Biochemical Characterization of Specific *GUCY2D* Alleles Associated With a Rare Form of Night Blindness	3-130
16:58	17:06			Josephine Prener Holtan	AAVB-039: A dual AAV8.ABCA4 gene therapy for patients with Stargardt's disease (STGD1)	3-131
17:06	17:30	Closing Ceremony	Bart Leroy, Francis Munier, Arlene Drack & Josephine Prener Holtan			N/A

Auditorium 2

Please note that parallel sessions are being conducted throughout the day!

Start	End	Session	Moderator	Presenter	Paper title	Presentation no.
8:30	9:00	REGISTRATION				
9:00	11:00	SESSION 1: Retinoblastoma Workshop				
9:00	9:20	Retinoblastoma Workshop	Christina Stathopoulos & Lisa Golmard	Eleanor Hay	The role of genetic counselling in retinoblastoma	3-201
9:20	9:40			Manoj Parulekar	Clinical presentation and Diagnosis	3-202
9:40	10:00			Christina Stathopoulous	Treatment modalities of intraocular retinoblastoma	3-203
10:00	10:20			Amy Gerrish	The applications of cell-free DNA analysis.	3-204
10:20	10:40			François Radvanyi	Molecular risk factors for retinoblastoma	3-205
10:40	11:00			Open discussion		N/A
11:00	11:30	BREAK Poster Session - Ophthalmic Genetics 2 to 7				
11:30	12:30	No sessions				
12:30	13:30	LUNCH Meet the sponsors Launch of third edition of "Genetic Diseases of the Eye", Edited by Elias Traboulsi, Viginia Miraldi Utz and Arif O. Khan, Published by Oxford University Press.				
13:30	14:30	IRD Patient Session				
13:30	14:00	Patient Session	Martin Smedstad	Sigrid Bratlie	Introduction to genetic treatments - Norwegian	3-206
14:00	14:30			Magnar Bjørås	Patient-derived retinal organoids - Norwegian	3-207
14:30	15:00			Daniel Chung	Gene-agnostic treatments - English	3-208
15:00	15:30			Tomas Aleman	Clinical trial design and outcome measures - English	3-209
15:30	16:00	BREAK Poster Session - Ophthalmic Genetics 2 to 7				
16:00	17:00	IRD Patient Session				
16:00	17:00	Patient Session	Martin Smedstad	Elias Traboulsi	Keynote lecture: Current research status and remaining challenges for treatment of inherited retinal disorders - English	3-210
				Bart Leroy		3-211
				Meghan DeBenedictis		3-212

Room 1

Please note that parallel sessions are being conducted throughout the day!

Start	End	Session
9:00	12:30	No sessions
12:30	12:50	LUNCH Meet the sponsors Launch of third edition of "Genetic Diseases of the Eye", Edited by Elias Traboulsi, Viginia Miraldi Utz and Arif Khan, Published by Oxford University Press.
12:50	15:30	EURBG Business Meeting
15:30	16:00	BREAK Poster Session - Ophthalmic Genetics Session 2 to 7
16:00	17:00	EURBG Business Meeting
17:00	17:30	No sessions

Friday, 12 September

Poster no.	Presenter	Poster title
Retinoblastoma		
Associated to Retinoblastoma 1- Retinoblastoma Diagnosis & Awareness		
1	Lucy Njambi	Setting up genetic testing services for retinoblastoma in Kenya : a SWOT analysis report
2	Joe Abbott	Familial retinoblastoma; distribution of new tumours and recurrent disease in the context of genetic
Associated to Retinoblastoma 2 - Retinoblastoma Treatment		
3	Ashwin Mallipatna	Tylectomy (pars plana endoresection) for retinoblastoma : indications, outcomes, and surgical technique
4	Elena-Cristina Nitulescu	What has changed in the treatment of retinoblastoma in Romania
5	Krzysztof Cieślik	Late recurrences of retinoblastoma
6	Yan Honggai	Analysis of the clinical characteristics of patients with retinoblastoma undergoing binocular enucleation
Associated to Retinoblastoma 3 - Retinoblastoma Metastatic Disease & Quality of Life		
7	Sima Das	Reclassifying Extraocular Retinoblastoma Based on Imaging: Prognostic Value of Orbital Spread Patterns
8	Maureen McCalla	The retinoblastoma nurse specialist in Europe - a vital link for effective communication and raising awareness with retinoblastoma families
Associated to Retinoblastoma 4 - Retinoblastoma Liquid Biopsy, Biomarkers & Research		
9	Jesus Hernandez Monge	Analysis of pathogenic variants in retinoblastoma reveals a potential gain of function mutation
10	Gabriel Alonso Perez	Study of the stability of Rb and its modulation caused by *MDM2*
11	Andrés Rodríguez	Retinoblastoma biomarkers in tears
12	Yuri Melissa Romero Chaves	Regulatory Mechanisms in Retinoblastoma
Ophthalmic Genetics		
Associated to Ophthalmic Genetics 1 - Retinal Disorders & Ocular Development		
13	Alejandra Antacle	ROSAH syndrome
14	Alex V. Levin	To characterize the ophthalmologic and genetic features of nine patients with *CLN3* disease (a form of neuronal ceroid lipofuscinosis or Batten disease) evaluated over a five-year period at a tertiary eye care
15	Alex V. Levin	Identifying Factors that Influence Wellness in Adult Patients with Non-syndromic Retinitis Pigmentosa
16	Arif O. Khan	*MERTK* -related retinopathy in the United Arab Emirates
17	Beau J. Fenner	Prioritizing High-Burden East Asian Variants for Targeted Therapy in Retinitis Pigmentosa
18	N/A	WITHDRAN
19	Diego Ignacio Paredes Jalil	*MPDZ* related Macular Dystrophy: a case report
20	Elena Avram	OFTALMOGENETICA.RO – Romanian online medical platform and registry for patients with inherited eye diseases
21	Elena Avram	*RPGR* Retinal dystrophy – clinical cases and a new genetic variant in romania
22	Erlend Sæther	Clinical and genetic characterization of *BEST1* - mediated retinal dystrophies in the Norwegian population
23	Hannah L. Scanga	Unilateral Retinal Dysfunction: A Retinal Riddle
24	Laura Echandi	Stargardt's like phenotyope by an unexpected gene
25	Milam A. Brantley	Benefits of an Interdisciplinary Inherited Retinal Disease Clinic
26	Miriam Ehrenberg	Beyond the Textbooks; Choroidal Neovascularization in Two teenagers with a Rare Retinal Dystrophy
27	Pascal Escher	Enhanced S-Cone sensitivity Syndrome (Goldmann-Favre-Syndrome): A case series
28	Raphael Gumafelix	Purpose: To report a case of Stargardt disease (STGD1) associated with an intronic mutation in the *ABCA4* gene.
29	Raphael Gumafelix	First Genetically Confirmed Case of Autosomal Recessive Bestrophinopahy (ARB) in the Philippines: A Sibling Case Study
30	Rola Ba-Abbad	Usher Syndrome: Dual Pathogenic Variants in a Single Family Leading to Different Phenotypes
31	Suneth Lindamulage	Genetically Characterized Autosomal Recessive Bestrophinopathy from Sri Lanka: A Three-Case Series Including a Patient with Two Novel Compound Heterozygous *BEST1* Variants
32	Virginie MM Buhler	Genotype/Phenotype Correlations in Patients with *ELOVL4* -linked Stargardt disease
33	Virginie MM Buhler	Molecular and Clinical Characterisation of *PRPH2* -p.(Arg172Trp)-associated Macular Dystrophy
34	Xènia Ferrer-Cortès	Major genes in inherited retinal dystrophies: ten years of next generation sequencing
35	Ariadni Gavriilidou	Use of Patient-Reported Outcome Measures (PROMs) in interventional clinical trials and observational studies regarding Inherited Retinal Diseases (IRDs) -A scoping review.
36	Louise Fischer Christensen	Are We Missing the Rest? A Scoping Review of Sleep, Fatigue, and Circadian Regulation in Retinitis Pigmentosa

Saturday, 13 September

Poster no.	Presenter	Poster title
Ophthalmic Genetics		
Associated to Ophthalmic Genetics 2 - General Category		
37	Alex V. Levin	Access to Care for Ocular Genetic Disease
38	Allison Weisman	Ocular Albinism Type 1 in Three Hispanic Families Associated with a Recurrent *GPR143* Hemizygous
39	Bronwyn Bateman	Evaluation of patients with genetic eye disease: Who should evaluate, the ethics of DNA testing and conflict of Interest?
40	N/A	WITHDRAWN
41	Christina Gerth-Kahlert	Evidence of digenic (TYR and OCA2) and tri-allelic inheritance of oculocutaneous albinism in two families
42	Diego Ignacio Paredes Jalil	Ocular manifestations of Gorlin-Goltz syndrome: a case report
43	Diego Ignacio Paredes Jalil	Establishing a low vision aids service in an Academic Institution
44	Emily Levine	A clinical review of neuro-ophthalmic developmental disorders resulting from defects in the γ-tubulin ring complex
45	Eva Roomets	Syndromic macular hypoplasia – two cases
46	Fulya Yaylacıoğlu Tuncay	Genotype and phenotype characteristics of patients with familial exudative vitreoretinopathy: First report on a Turkish cohort.
47	Goura Chattannavar	Sudden bilateral vision loss in a child with *LYRM7*-related Leukoencephalopathy, differentiating from Acute Disseminated Encephalomyelopathy
48	Goura Chattannavar	Novel Ocular Phenotype in *TMEM53*-Associated Craniotubular Dysplasia
49	Goura Chattannavar	A Year in Ocular Genetics at a single centre: Clinical Insights and Key Learnings
50	Hannah L. Scanga	When One Diagnosis Becomes Two: Lessons from the Diagnostic Workup of Albinism in a Single Family
51	Hannah L. Scanga	*PRPF31*-related Retinitis Pigmentosa and Cystoid Macular Edema: A Cohort Study
52	Hannah L. Scanga	Detection of Bilateral Cataracts in Male Fetuses at Routine 20 Week Ultrasound: Always Examine the Mothers!
53	Jennifer Ling	The Effect of Oral Belzutifan on Retinal Hemangioblastomas in Von-Hippel-Lindau Syndrome
54	Karthikeyan Arcot Sadagopan	Abnormalities in extraocular muscles and their insertions is one of the most consistent yet least reported ocular abnormalities in albinism; a developmental hypothesis and its clinical implications.
55	Katja Rončević	Expanding the *IDH3A* genotype: macular pseudocoloboma in the absence of a null variant
56	Konsta Kesti	A Novel Heterozygous c.1103C>G Variant in *FZD5* in a Finnish Family with Coloboma and Microphthalmia
57	Molly Welsh	Risk of Optic Neuropathy in Pediatric Patients with Molecularly Confirmed External Ophthalmoplegia including CFEOM
58	Pauliina Repo	*BAP1* Germline Variants in Finnish Patients with Uveal Melanoma
59	Mohamed H. Abdel-Rahman	*BAP1*-Tumor Predisposition Syndrome: The Ohio State University Experience
60	Marilena Cojocaru	Leber hereditary optic neuropathy (LHON) – a neurodegenerative disorder of the optic nerve with mitochondrial and autosomal recessive inheritance
Associated to Ophthalmic Genetics 3 - Gene Therapy, Natural history & Outcome Measures		
61	Alessia Amato	Exploring the Role of Two-Color Dark-Adapted Perimetry in Inherited Retinal Diseases Beyond Retinitis Pigmentosa
62	Brittni Scruggs	Retinal Viral Gene Therapy: Impact of Route of Administration on Serious Adverse Events
63	Julie Van Puyvelde	Deep Retinal Phenotyping in a Case of Best Vitelliform Macular Dystrophy using Adaptive Optics Imaging
64	Marcela Perez	Clinical and multimodal imaging characterization of a Stargardt Cohort in Chile
65	Nicolas Chateau	Imaging protocol for retinal phenotyping at the microscopic level in retinitis pigmentosa
66	Pascal Escher	High-Resolution OCT findings in patients with Sorsby Fundus Dystrophy: A case series
67	Pascal Escher	Multimodal longitudinal analysis of Swiss patients harboring the pathogenic *PRPH2* c.422A>G p.(Tyr141Cys) variant
Associated to Ophthalmic Genetics 4 - Anterior Segment		
68	Jennifer Rossen	Gene Curations for Crystallin Genes and Pediatric Cataracts
Associated to Ophthalmic Genetics 5 - Treatment for IRDs		
69	Leen Hertens	Short-term outcomes of Belgian paediatric patients with mild autosomal recessive *RPE65*-associated retinal dystrophy treated with voretigene neparvovec
70	Marcela Ciccioli	Prognosis of *ABCA4* retinopathies patients (and other IRDs) when they use filters and supplements.
71	Marcela Ciccioli	Demographic perspective of IRDs in Argentina: national database, candidat patients selection for clinical trials and advanced therapies
72	Michelle Alabek	Clinical Trial Knowledge Among Adults with Inherited Retinal Dystrophy (IRD)

Associated to Ophthalmic Genetics 6 - Retinal Dystrophies & Electrophysiology		
73	Jennifer Ling	Novel Variants in Congenital Stationary Night Blindness
74	Johane Robitaille	Molecular Genetic Testing for Inherited Retinal Dystrophies in Maritime Canada
75	Molly Welsh	Clinical Value of PERG in Pediatric Genetic Eye Disease
76	Sarah J. Garnai	Genotypic spectrum of nanophthalmos and high hyperopia in the United States and Brazil
77	Sigrid Aslaksen	Functional Characterization of *ABCA4* Missense Variants Aids Variant Interpretation and Phenotype Prediction in Patients With *ABCA4*-Retinal Dystrophies
78	Maria Ludovica Ruggeri	Development of a machine learning enabling OCT-based algorithm to assign regenerative therapy modality in patients with hereditary retinal disorders
79	Arjana Alihodzic	Exploring the association between *OPN1LW* exon-3 skipping haplotypes and refractive error in a population with low myopia prevalence
80	Farjina Jannat Juli	Exploring the relationship between early-onset high myopia (eoHM) mutations and single nucleotide polymorphism (SNPs) in the *ARR3* gene
Associated to Ophthalmic Genetics 7 - IRDs - Mechanisms & Treatments		
81	Fabiola Ceroni	Wnt ligand secretion mediator (WLS) and structural eye anomalies: a novel case and zebrafish models support the critical role of WLS in eye development and its interactions with other eye genes
82	Jørn-Ove Schjølberg	Modeling *PRPF3* I Retinitis Pigmentosa 11 in Patient-Derived Retinal Organoids: A Gene-Therapy Model
83	Vlasta Hadalin	Towards exploring the role of *RPGR* isoforms in rods and cones in human retinal organoids