International Society for Genetic Eye Diseases & Retinoblastoma

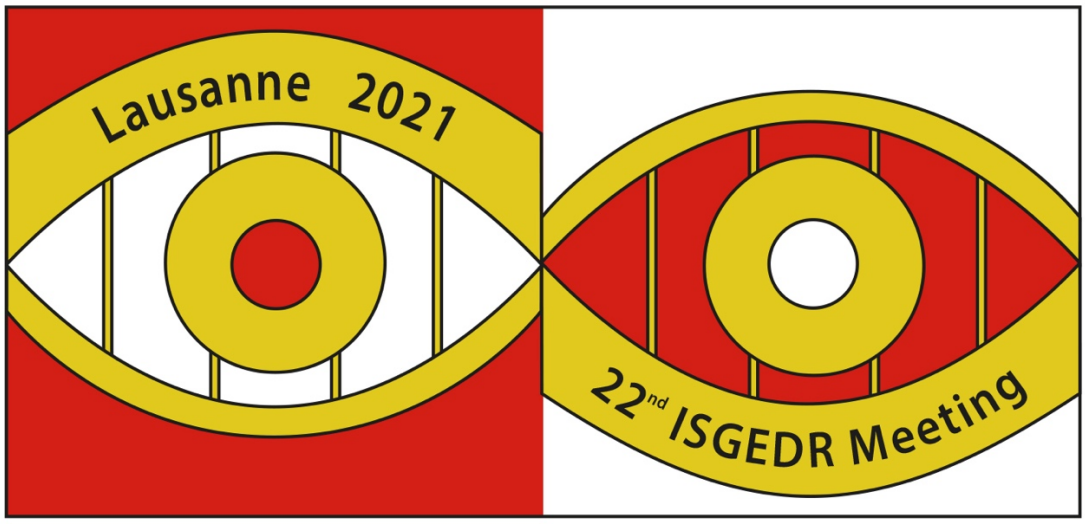
ISGEDR 2021 Meeting

Program

Olympic Museum
Lausanne, Switzerland
2 - 4 September 2021

Local Host & ISGEDR President:
Professor Francis MUNIER
Hôpital Ophtalmique Jules-Gonin, Lausanne, Switzerland

Held in hybrid format and jointly with the
European Retinoblastoma Group (EURbG)
Société de Génétique Ophtalmologique Francophone (SGOF)
&
European Reference Network for Rare Eye Diseases (ERN-EYE)



Venue: Musée Olympique

Quai d'Ouchy 1, 1006 Lausanne, Switzerland

Program Committee (alphabetical):

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Guillermo CHANTADA, Barcelona, Spain & Buenos Aires, Argentina
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Meeting Organizers (alphabetical):

Paola CAPUTO, Lausanne, Switzerland
Geoff CROSS, Harrogate, UK
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Francis MUNIER, Lausanne, Switzerland
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Continuous Medical Education (CME) Credits:

The Swiss Society of Ophthalmology (SSO) has awarded 18 CME credits for the entire ISGEDR meeting; 7 CME credits for Thursday, 2 Sep 2021; 6 CME credits for Friday and 5 CME credits for Saturday. You will be asked to fill in an evaluation form online, after which you can claim your CMEs. Practical instructions on how to do that will be made available during the ISGEDR 2021 Meeting.

Comments:

A unique abstract code for each presentation was generated based on chronological submission dates. The code can be used to find an abstract in the ISGEDR 2021 Meeting Abstract Book.

All times stated are Lausanne times (Central European Time - CET).

The meeting language is English, except for the SGOF 2021 Satellite Meeting, which will be held in French.

Sponsors:

The meeting organizers are exceptionally grateful to the following sponsors for their financial support for the ISGEDR 2021 Hybrid Meeting. The challenges in these difficult times would have been very difficult to overcome without their strong and enthusiastic support.

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Thursday, September 2nd, 2021
ISGEDR 2021
Ocular Genetics Sessions

08.00-9.00 hrs WELCOME COFFEE

MORNING SESSIONS **Auditorium**

8:30-8:45 hrs WELCOME Francis MUNIER, Isabelle S AUDO, Annette MOLL & Hélène DOLFUSS

8:45-10:00 hrs Ophthalmic Genetics 1 Phenotypes & Genotypes - Inherited Retinal Disorders

Moderators (in person) Isabelle MEUNIER & Irene H MAUMENEE

8:45-8:53 *Abstract OcGen6* Isabelle MEUNIER, Montpellier *(in person)*
Retinitis Punctata Albescens and *RLBP1*-Associated Phenotypes: Phenotype-Genotype Correlation and Natural History with the Aim of Gene Therapy

8:53-9:00 *Abstract OcGen7* Alina V DUMITRESCU, Iowa City, IA *(in person)*
Congenital Stationary Night Blindness Presenting without Night Blindness in Children. What Leads to Diagnostic Delays?

9:00-9:07 *Abstract OcGen78* Gabriel HALLALI, Paris *(in person)*
***NR2E3*-Related Dystrophies: Retrospective Case Series of 50 Patients. Genetic Findings, Detailed Phenotype and Electrophysiological Analysis**

9:07-9:14 *Abstract OcGen34* Julie DE ZAEYTIJD, Ghent *(in person)*
Isolated Maculopathy and Moderate Rod–Cone Dystrophy Represent the Milder End of the *RDH12*-Related Retinal Dystrophy Spectrum

9:14-9.21 *Abstract OcGen99* Irene H MAUMENEE, New York, NY *(in person)*
Retrospective Analysis of Ocular Findings in Patients with Biallelic Mutations in *CRB1*

9:21-9:28 *Abstract OcGen101* Rachel HEATH JEFFERY, Perth *(virtual)*
Sibling Concordance in Lesion Growth Rates in Stargardt Disease Using Ultra-Widefield Fundus Autofluorescence

9:28-9:35 *Abstract OcGen57* Jana SAJOVIC, Ljubljana *(virtual)*
Progression of Stargardt Disease as Determined by Electroretinography and Autofluorescence over a Median Period of 17 Years

9:35-9:42 *Abstract OcGen24* Ine STRUBBE, Ghent *(in person)*
Autosomal Recessive Bestrophinopathy in a Ghent Cohort

9:42-9:49 *Abstract OcGen77* Francesco TESTA, Naples *(virtual)*
Genotype-Phenotype Correlation in a Large Italian Cohort of Patients with Autosomal-Dominant Retinitis Pigmentosa due to Variants in *RHO* gene: a Longitudinal Natural History Study

9:49-9:54 *Abstract OcGen36* Nika VRABIC, Ljubljana *(in person)*
Progression Of Outer Retinal Layer Structural Changes in *CNGA3* - and *CNGB3* - Associated Retinopathy

9:54-10:00 Discussion

Ophthalmic Genetics 1 Associated Posters with Online Rapid-Fire Presentations

Abstract OcGen68 Kevin MAIROT, Montpellier *(in person)*
Clinical and Genetic Characteristics of 38 Consecutive Patients with *CRB1*-associated Retinal Dystrophies

Abstract OcGen48 Nicholas BELLO, Philadelphia, PA *(virtual)*
Two Cases of *CRB1*-Related Retinal Dystrophy Associated with Retinal Masses

Abstract OcGen81 Valentina DI IORIO, Naples *(virtual)*
A Multicentric Longitudinal Natural History Study of Usher Syndrome due to Mutations in the *MYO7A* gene: Preliminary Results in the Italian Cohort

Abstract OcuGen1 Monika GRUDZINSKA PECHHACKER, Stockholm *(in person)*
Natural history study of visual function in patients with BBS1- and BBS10-related retinal degeneration

Abstract OcGen56 Asimina MATAFTSI, Thessaloniki *(in person)*
Very High Myopia Associated with *RPGR* Gene Variant

Abstract OcGen80 Ana FAKIN, Ljubljana *(virtual)*
Genetic and Clinical Characteristics of Slovenian Patients with *RPGR*-Associated Retinal Disorders

Abstract OcGen69 Marco NASSISI, Paris *(in person)*
Genotype-Phenotype Correlation and Disease Modeling in *RPGR*-Related Retinal Dystrophies

Abstract OcGen60 Julien NAVARRO, Paris *(in person)*
Deep Analysis of WGS Data, Strategy and Algorithms

Abstract OcGen74 Andrea VINCENT, Auckland *(virtual)*
Natural History of Retinal Degeneration Associated with a Founder Mutation in the *PDE6B* Gene

Abstract OcGen83 Christina GERTH-KAHLERT, Zürich *(in person)*
Phenotype Variability Associated with Mutations in the *POC1B* Gene

Abstract OcGen89 Lev PRASOV, Ann Arbor, MI *(virtual)*
Phenotypic and Molecular Characterization of Ocular and Systemic Features of *DDX58*-Related Singleton-Merten Syndrome

Abstract OcGen102 Anita AGARWAL, Nashville, TN *(virtual)*
Insights from Imaging *PRPH2* Phenotypes

Abstract OcGen35 Tjaša KRAŠOVEC / Ana FAKIN, Ljubljana *(virtual)*
Association Between Phenotype and Serum Concentration of Vitamin A in Patients with p.G90D Variant in the *RHO* Gene

Abstract OcGen49 Hosannah EVIE, St. Louis, MO *(virtual)*
Manifestation of Retinal Dystrophy in a child with *TCIRG1*-related Autosomal Recessive Osteopetrosis

Abstract OcGen76 Anaïs GRIMAUD, Paris *(in person)*
Identification of Dormant Cones in Subjects with Rod-Cone Dystrophy

Abstract OcGen61 Luiza NEVES, Paris *(in person)*
USH2A Variants Spectrum in a French Cohort

Abstract OcGen4 Monika GRUDZINSKA PECHHACKER, Stockholm *(in person)*
SCLT1-Related Disease as a Rare Cause of Cone Dystrophy with Subtle Systemic Associations Resembling Ciliopathy

Abstract OcGen27 Monika CHYLOVA, Prague *(in person)*
Spectrum of Mutations in 11 Choroideremia Czech Patients

Abstract OcGen28 Kiyoko GOCHO, Chiba *(in person)*
High-resolution Retinal Imaging Analysis of Patients with Autosomal Dominant Retinitis Pigmentosa Caused by *HK1* Mutation

Abstract OcGen32 Giacomo Maria BACCI, Florence *(in person)*
Optic Nerve Involvement in *CACNA1F* Phenotype: Observations in a Multicentric Case Series

Abstract OcGen11 Marie VAJTER, Prague *(in person)*
Achromatopsia in the Czech Republic: Mutational Spectrum and Clinical Findings

Abstract OcGen41 Nadine NSIANGANI LUSAMBO, Kinshasha *(virtual)*
Phenotype and Genotype of Inherited Retinal Disorders in Democratic Republic of Congo (DRC)

Abstract OcGen39 Evangelia S PANAGIOTOU, Thessaloniki *(virtual)*
Studying the Genetic Basis of Inherited Retinal Diseases (IRDs) in Greece: Preliminary Results

10:00-11:00 hrs Ophthalmic Genetics 2 Mechanisms of Disease & Preclinical Models in Ophthalmic Genetics

Moderators (in person) Pascal ESCHER & Carla SANJURJO SORIANO

10:00-10:07 *Abstract OcGen22* Yvan ARSENIJEVIC, Lausanne *(in person)*
Polycomb Repressive Complex-2 Contributes to Rod Photoreceptor Death Process in Several Forms of Retinal Degeneration

10:07-10:14 *Abstract OcGen29* Helen FREDERIKSEN, Paris *(in person)*
Shedding Light on Retina Signaling by Studying Complete Congenital Stationary Night Blindness

10:14-10:21 *Abstract OcGen51* Tasnim BEN YACOUB, Paris *(in person)*
Toward a Better Understanding of *ITM2B* Pathogenicity in a Specific Retinal Dystrophy, and its Potential Role in Mitochondria

10:21-10:28 *Abstract OcGen38* Ning CHANG, Lausanne *(in person)*
Towards Gene Therapy for *FAM161A*-Associated Retinitis Pigmentosa in a Mouse Model

10:28-10:35 *Abstract OcGen53* Carla SANJURJO SORIANO, Montpellier *(in person)*
Validation of CRISPR/Cas9-Mediated Gene Correction of the Two Most Prevalent *USH2A* Mutations in iPSC-Derived Retinal Organoids

10:35-10:42 *Abstract OcGen42* Florian UDRY, Lausanne *(in person)*
Suitability of iPS-derived Retinal Pigment Epithelium (iRPE) as a Model for Lentiviral-mediated Gene Therapy (GT)

10:42-10:49 *Abstract OcGen2* Imran YUSUF, Oxford *(virtual)*
Gene Therapy Rescues Cone and Rod Function in a Pre-Clinical Model of *CDHR1*-Associated Retinal Degeneration Through Restoration of Photoreceptor Outer Segments

10:49-10:56 *Abstract OcGen37* Pascal ESCHER, Bern *(in person)*
Mouse Models for Dominant and Recessively Inherited *NR2E3*-linked Retinal Degenerations

10:56-11:03 *Abstract OcGen106* Robert HUFNAGEL, Bethesda, MD *(virtual)*
A Tissue-specific Allelic Hierarchy Predicts Phenotypes for *USH2A*-related Disorders in the RUSH2A Study

11:03-11:10 Discussion

11:05-11:30 hrs COFFEE BREAK & POSTER VIEWING

11:30-12:30 hrs Ophthalmic Genetics 3 Treatment of Inherited Retinal Disorders

Moderators in person Birgit LORENZ & Veronika VACLAVIK

11:30-11:45 *Abstract OcGen14* Hendrik SCHOLL, Basel *(in person)*
Therapeutic Pipeline for Stargardt Disease

11:45-11:52 *Abstract OcGen40* Vasily SMIRNOV, Paris *(in person)*
High Dose Vitamin A Improves a Fundus Albipunctatus-like Phenotype Related to a Novel Null Retinol Carrier -Variant

11:52-12.02 *Abstract OcGen45* Birgit LORENZ, Bonn *(in person)*
Voretigene Neparvovec Gene Therapy to Treat *RPE65*-Mutation-Associated IRDs – Experience in German Centers

12:02-12:12 *Abstract OcGen104* Daniel CHUNG, Paris *(in person)*
Select Strategies for Gene Agnostic Approaches to Cone Photoreceptor Function Preservation

12:12-12:22 *Abstract OcGen31* Bart P LEROY, Ghent & Philadelphia, PA *(in person)*
The Phase III REFLECT Trial: Efficacy and Safety of Bilateral Gene Therapy for Leber Hereditary Optic Neuropathy (LHON)

12:22-12:30 Discussion

12:30-13:30 hrs LUNCH BREAK

AFTERNOON SESSIONS Auditorium

13.30-13.40 hrs Bart P LEROY, Ghent & Philadelphia, PA *(in person)*
Introduction of the Jules François Lecturers

13.40-14.20 hrs Jean BENNETT & Albert M MAGUIRE, Philadelphia, PA *(virtual)*
FRANÇOIS LECTURE: Using Learnings from *RPE65* Gene Therapy to Advance Treatments for Other Forms of Congenital Blindness

14.20-16.00 hrs Symposium: Gene Therapy (Nuclear Genes)

Moderators (in person): Hwei Wuen CHAN, Daniel C CHUNG & Mark PENNESI

14.20 - 14.30 Dominik FISCHER, Oxford & Tübingen *(virtual)*
Luxturna in Clinical Practice: The PERCEIVE Study

14.30 - 14.40 José A SAHEL, Paris & Pittsburgh, PA *(virtual)*
Optogenetics: The Generic Future?

14.40 - 14.50 Tomas S ALEMAN, Philadelphia, PA *(virtual)*
Untangling Phenotypes and Treating Severe Retinal Degenerations

14.50 - 15.00 Paul SIEVING, Davis, CA *(virtual)*
Intravitreal AAV-based Gene Therapies: XLRS

15.00 – 15.10 Bart P LEROY, Ghent & Philadelphia, PA *(in person)*
RNA Interference Treatment for *CEP290*-IRD

15.10 - 15.20 Isabelle S AUDO, Paris *(in person)*
RNA Interference Treatment for *USH2A*-IRD

15.20 – 15.30 Alberto AURICCHIO, Naples *(virtual)*
Expanding AAV Transfer Capacity in the Retina

15.30 – 15.40 Steven S TSANG, New York, NY *(virtual)*
CRISPR/Cas9 for Inherited Retinal Disease

15.40 – 15.50 **Discussion**

15.50 – 16.20 hrs COFFEE BREAK & POSTER VIEWING

16:20-18:05 hrs Ophthalmic Genetics 4 Grand Rounds ERN-EYE Transnational (GREET) by the European Reference Network for Rare Eye Disease (ERN-EYE)

Moderators (in person) Isabelle S AUDO, Bart P LEROY & Hélène DOLLFUS

16:20-16:26 *Abstract OcGen85* Andreea-Alexandra IONESCU, Strasbourg *(virtual)*
A Maculopathy in a Patient Diagnosed with a Rare Plurimalformative Overgrowth Syndrome

16:26-16:32 *Abstract OcGen46* Hwei Wuen CHAN, Ghent & Singapore *(in person)*
Clinical Manifestations of the Non-Syndromic Subtype of *MFSD8*-associated Disease

16:32-16:38 *Abstract OcGen32* Sabine DEFOORT-DHELLEMMES, Lille *(in person)*
Early diagnosis of *CDG1*-associated Retinopathy

16:38-16:44 *Abstract OcGen16* Ian MACDONALD, Alberta *(in person)*
A Novel Mutation in *PIKfyve* Causes a Multi-System Autosomal Dominant Disorder with Iris Transillumination Defects

16:44-16:50 *Abstract OcGen71* Mariana DE PALMA, São Paolo *(virtual)*
Atypical X-linked Retinoschisis

16:50-16:56 *Abstract OcGen25* Alessia AMATO, Milan *(in person)*
Regressive Flecked Retina Phenotype in a Case of Initially Misdiagnosed Senior-Loken Syndrome

16:56-17:02 *Abstract OcGen9* Juliana SALLUM, São Paolo *(virtual)*
Analysis of a Next-Generation Retinopathy Panel Detects Chromosome 1 Segmental Uniparental Isodisomy in a Patient with *RPE65*-related Leber Congenital Amaurosis

17:02-17:08 *Abstract OcGen87* Asma HASSAIRI, Tunis *(virtual)*
Whole Exome Sequencing Identified Novel Homozygous *ALMS1* Mutations in a Tunisian Family with Alström Syndrome

17:08-17:14 *Abstract OcGen100* Rebecca SCHUR, Cleveland, OH *(virtual)*
Atypical Electrophysiological Profile of a Missense Mutation in *BEST1*

17:14-17:20 *Abstract OcGen44* Filip VAN DEN BROECK, Ghent *(in person)*
The Usher Syndrome 1B Spectrum Includes Mild Cases

17:20-17:26 *Abstract OcGen94* Giancarlo IAROSSI, Rome *(in person)*
Blue Cone Monochromatism with Foveal Hypoplasia Caused by the Concomitant Effect of Variants in *OPN1LW*/*OPN1MW* and *GPR143* genes

17:26-17:32 *Abstract OcGen55* Alessia AMATO, Milan *(in person)*
Clinically Diagnosed Dominant Cystoid Macular Dystrophy Presenting with a Peculiar Phenotype

17:32-17:38 *Abstract OcGen67* Sarah VAN DER ENDE, Halifax *(virtual)*
A Syndrome of Hearing Loss and FEVR in a Girl with Biallelic *FZD4* variants

17:38-17:44 *Abstract OcGen3* Basamat AL MOALLEM, Riyad *(virtual)*
Novel Biallelic *AHR* Splice Site Mutation Causes Isolated Foveal Hypoplasia in Saudi Patient

17:44-17:50 *Abstract OcGen50* Arif O KHAN, Abu Dhabi *(virtual)*
Retinal Arteriolar Macroaneurysms with Supravalvular Pulmonic Stenosis in the United Arab Emirates

17:50-17:56 *Abstract OcGen5* Martina JARC-VIDMAR, Ljubljana *(in person)*
Macular Dystrophies with Secondary CNV – Diagnosis and Treatment

17:56-18:02 *Abstract OcGen88* Emna OTHMANI, Tunis *(virtual)*
OCT-Angiography Assessing Quiescent and Active Choroidal Neovascularization in Retinitis Pigmentosa Associated with a *PRPH2* Pathogenic Variant

18:02-18:08 *Abstract OcGen8* Virginia A MIRALDI UTZ, Cincinnati, OH *(virtual)*
Long-Term Results of Planned Preterm Delivery and Treatment of a Child with Norrie Disease

18:08-18:15 Discussion

Ophthalmic Genetics 4 GREET-Associated Posters with Online Rapid-Fire Presentations

Abstract OcGen48 Chloé WERNER, Nantes *(in person)*
A Novel *RHO* Mutation: Atypical Phenotype

Abstract OcGen95 Irina BALIKOVA, Leuven *(in person)*
Congenital Stationary Night Blindness in a Patient with Mild Learning Disability due to a Compound Heterozygous Microdeletion of 15q13 and a Missense Mutation in *TRPM1*

Abstract OcuGen93 Aleksandra PETROVIC, Lausanne *(in person)*
A Novel *TGFBI* Gene Mutation Associated with a Deep Stromal Phenotype and Late-Onset Lattice Dystrophy in a French Family

Abstract OcGen84 Marine SOURDRIL, Nantes *(in person)*
Jalili Syndrome: a Novel Mutation - Phenotypic Expression

Abstract OcGen15 Akiko HIKOYA, Hamamatsu *(virtual)*
A Case of Early-Onset Retinal Dystrophy Associated with *NEK1* Gene Mutations in Siblings

Abstract OcGen19 Savleen KAUR, Chandigarh *(virtual)*
A Novel *GUCY2D* Missense Mutation in a Patient with Leber Congenital Amaurosis

Abstract OcGen79 Raffaella BRUNETTI-PIERRI, Naples *(virtual)*
A Patient with a Mild Clinical Presentation of Joubert Syndrome due to Biallelic *MKS1* Truncating Mutations

18.15 – 20.15hrs SWISS WINE TASTING
at Musée Olympique

FREE EVENING - *Please be informed that in Switzerland, restaurants do not serve food after 21.00hrs*

Friday 3 September 2021
ISGEDR 2021
Retinoblastoma Sessions

08.00-9.00 hrs WELCOME COFFEE

MORNING SESSIONS **Auditorium**

9:15-10:00 hrs Retinoblastoma 1 Diagnosis, Clinical Outcome & Second Cancers

Moderators in person: Christina STATHOPOULOS & Annette MOLL

9:15-9:22 *Abstract Rb23* Christiaan DE BLOEME, Amsterdam *(in person)*
Imaging Phenotype of Retinoblastoma with MYCN Amplification without RB1 Mutation: a Multicenter Case Control Study

9:22-9:29 *Abstract Rb21* Isabelle AERTS, Paris *(virtual)*
Results of a Second Multicenter Prospective French National Study on the Postoperative Treatment of Unilateral Retinoblastoma after Primary Enucleation

9:29-9.36 *Abstract Rb43* Christina STATHOPOULOS, Lausanne *(in person)*
Long Term Efficacy and Safety of Intravitreal Chemotherapy for Vitreous Seeding in Retinoblastoma

9:36-9:43 *Abstract 34* Francis MUNIER, Lausanne *(in person)*
Long-Term Efficacy, Safety and Toxicity of Intracameral Chemotherapy for Spontaneous Primary and Secondary Aqueous Seeding in Retinoblastoma

9:43-9:50 *Abstract Rb11* Gabriela VILLANUEVA, Buenos Aires *(virtual)*
Subsequent Malignant Neoplasms in the Pediatric Age in Retinoblastoma Survivors in Argentina

9:50-10:00 Discussion

Retinoblastoma 1 Associated Posters with Online Rapid-Fire Presentations

Abstract Rb6 Alicia LÓPEZ DE EGUILETA, Barcelona *(virtual)*
Application of Ruthenium-106 Brachytherapy Plaque for Residual or Recurrent Retinoblastoma after Intra-arterial Chemotherapy

Abstract Rb10 Mona MOHAMMAD, Amman *(virtual)*
Intravitreal Melphalan Chemotherapy for Vitreous Seeds in Retinoblastoma

Abstract Rb13 Nadiia BOBROVA, Odesa *(virtual)*
Different Melphalan Intravitreal Dose in Combined Retinoblastoma Polychemotherapy

Abstract Rb26 Tatiana USHAKOVA, Moscow *(virtual)*
The Role of Three-component Selective Intra-arterial Chemotherapy in the Failure of Standard Organ-Preserving Treatment in Children with Intraocular Retinoblastoma

Abstract Rb14 Bastien MARTI, Lausanne *(virtual)*
Non-ocular Vascular Aspects and Complications of Intraarterial Chemotherapy in Children with Retinoblastoma: a Ten-year Experience

Abstract Rb29 Petra KETTELER, Essen *(virtual)*
Comparison of Efficacy and Side-effects of Two Different Eye-preserving Systemic Chemotherapy Regimens in Children with Retinoblastoma

Abstract Rb32 Denis VOLODIN, Moscow *(virtual)*
Transpupillary Thermotherapy in Children with Retinoblastoma: Long-Term Results

Abstract Rb25 Parag SHAH, Coimbatore *(virtual)*
Safety and Efficacy of Pars Plana Vitrectomy in Management of Rhegmatogenous Retinal Detachments with Posterior Breaks Occurring urding Management of Active Retinoblastoma

Abstract Rb35 Rejin KEBUDI, Istanbul *(virtual)*
Advances in Treatment of Bilateral Retinoblastoma: 26-year Experience

Abstract Rb37 Aaditeya JHAVERI, Toronto *(virtual)*
Updating and Evaluating Adherence to Standard Operating Procedures in the Retinoblastoma Pathway of Care

Abstract Rb7 Nara BADALOVA, Amsterdam *(virtual)*
Evaluation of Screening Protocol Used for Children at Risk for Familial Retinoblastoma: Dutch Retrospective Population Based Cohort Study

Abstract Rb36 Alexandre MATET, Paris *(in person)*
Advanced Unilateral Retinoblastoma: Proposal for European Standard Clinical Practice Guidelines (ERN PaedCan / EuRbG Working Group 2)

10:00-11:00 hrs Retinoblastoma 2 Biology and Dissemination

Moderators in person: Alexandre MOULIN & Josephine DORSMAN

10:00-10:20 Invited Lecture Guillermo CHANTADA, Barcelona & Buenos Aires *(virtual)*
Description and Clinical Relevance of Two Molecular Retinoblastoma Subtypes

10:20-10:27 *Abstract Rb22* Santiago ZUGBI, Buenos Aires *(virtual)*
Preclinical Platform of Disseminated Retinoblastoma

10:27-10:34 *Abstract Rb4* Ashwin REDDY, London *(virtual)*
MYCN Amplification Levels in Primary Retinoblastoma Tumors Analyzed by Multiple Ligation-Dependent Probe Amplification

10:34-10:41 *Abstract Rb42* Rod BREMNER, Toronto *(virtual)*
Pan-Cancer Insights for New Retinoblastoma Therapies

10:41-10:48 *Abstract Rb41* Adeline BERGER, Lausanne *(in person)*
H3K27Ac Profiling to Elucidate the Epigenetic Identity of Retinoblastoma

10:48-10:55 *Abstract Rb39* Nathalia GRIGOROVSKI, Rio de Janeiro *(virtual)*
Multimodality Treatment Including Intra-Arterial Chemotherapy for Disseminated Retinoblastoma

10:55-11:00 Discussion

Retinoblastoma 2 Associated Posters with Online Rapid-Fire Presentations

Abstract Rb38 Alexandre MOULIN, Lausanne *(in person)*
Distant Retrobulbar Optic Nerve Infiltration in Treated Retinoblastoma: a Case Report

Abstract Rb27 Tatiana USHAKOVA, Moscow *(virtual)*
Extended Surgical Interventions in Children with Retinoblastoma Invasion into the Optic Nerve

Abstract Rb12 Nadiia BOBROVA, Odesa *(virtual)*
Enucleation Using High-Frequency Electric Welding in High-Risk Retinoblastoma

11:00-11:30 COFFEE BREAK & POSTER VIEWING

11:30-12:30 hrs Retinoblastoma 3 Preclinical and Early Clinical Research

Moderators (in person) *Manoj PARULEKAR & Yvan ARSENIJEVIC*

11:30-11:38 *Abstract Rb15* Paula SCHAIQUEVICH, Tandil *(virtual)*
Ocular and Systemic Safety of Repeated Intravitreal High Doses of Topotecan in Rabbits: Implications in Retinoblastoma Treatment

11:38-11:46 *Abstract Rb16* Po-Jen TSENG, Lausanne *(in person)*
High-Throughput Screening of Drug Libraries Identifies a New Synergistic Drug Combination for the Treatment of Retinoblastoma

11:46-11:54 *Abstract Rb19* Irina SINENKO, Lausanne *(in person)*
Three-Dimensional In Vitro Retinoblastoma Model to Evaluate Drugs in Combination with Focal Therapy

11:54-12:02 *Abstract Rb33* Rosario ASCHERO, Buenos Aires *(virtual)*
An Immunohistochemistry Panel as a Tool for Identifying Retinoblastoma Molecular Subtypes

12:02-12:10 *Abstract Rb24* Brenda GALLIE, Toronto *(virtual)*
Phase I Retinoblastoma Trial of Sustained-Release Episceral Topotecan

12:10-12:18 *Abstract Rb40* Isabella JANUSONIS, Toronto *(virtual)*
Treatments Required for Retinoblastoma Following Experimental Chemoplaque versus Standard Care

12:20-12:30 Discussion

12:30-13:30 LUNCH BREAK

AFTERNOON SESSIONS Auditorium

13.30-13.40 hrs Francis L MUNIER, Lausanne *(in person)*
Introduction of the Robert M Ellsworth Lecturer

13.40-14.20 hrs David H ABRAMSON, New York, NY *(virtual)*
ELLSWORTH LECTURE: Cell Free DNA in Retinoblastoma: What We Know and What We Don't Know in 2021

14:30-15:30 hrs Retinoblastoma 4 Mini-Symposium on Liquid Biopsies

Moderators (in person) *Dietmar LOHMANN & Charlotte DOMMERING*

14:30-14:38 *Abstract Rb1* Irene JIMÉNEZ, Paris *(virtual)*
Molecular Diagnosis of Retinoblastoma by Circulating Tumor DNA Analysis

14:38-14:45 *Abstract Rb18* Nicole BARWINSKI, Essen *(in person)*
cf-DNA and EVs as Sources for Biomarkers for Early Detection of Second Primary Malignancies in Patients with Heritable Retinoblastoma

14:45:14:52 *Abstract Rb30* Lisa GOLMARD, Paris *(virtual)*
Highly Sensitive Detection Method of Retinoblastoma Genetic Predisposition and Biomarkers

14:52-15:00 *Abstract Rb2* Jesse BERRY, Los Angeles, CA *(virtual)*
The Aqueous Humor Liquid Biopsy for Diagnosis, Prognosis and Treatment monitoring of Retinoblastoma

15:00-15:07 *Abstract Rb20* Luiz Fernando TEIXEIRA, São Paulo *(virtual)*
Investigating the Aqueous Humor and Tumor Tissue of Naïve Retinoblastoma Eyes in a Next-Generation Sequencing Panel for Pediatric Malignancies

15:07-15:14 *Abstract Rb9* Angela GALARDI, Rome *(virtual)*
Proteomics of Aqueous Humor for Biomarkers Discovery in Retinoblastoma

15:14-15:30 Discussion

15.30-16.00 hrs Invited Lecture Botond ROSKA, Basel *(in person)*
Retinal Cell Types as a Basis for Ophthalmology in the 21st Century

16.00 – 16.30hrs COFFEE BREAK & POSTER VIEWING

16:30-17:30 hrs Retinoblastoma 5 Retinoblastoma around the World

Moderator (in person) *Maja BECK-POPOVIC & Antonino ROMANZO*

16:30-16:50 Invited lecture Sandra LUNA *(in person)*
WHO Global Initiative for Childhood Cancer: Retinoblastoma Priority Program and Collaborations

16:50-17:00 Invited lecture Didi FABIAN *on behalf of the Global Rb Study Group* *(virtual)*
Retinoblastoma Survival: A Prospective analysis of 4064 Patients from 260 Countries

17:00-17:05 *Abstract Rb5* Laurence DESJARDINS, Paris *(virtual)*
AMCC Program in Sub-Saharan Africa

17:05-17:10 *Abstract Rb31* Helen DIMARAS, Toronto, Canada *(virtual)*
Characterization of International Partnerships in Global Retinoblastoma Care and Research: A Network Analysis

17:10-17:15 *Abstract Rb8* Yacoub YOUSEF, Amman *(virtual)*
How Telemedicine and Centralized Care Changed the Natural History of Retinoblastoma in Jordan: Analysis of 478 Patients

17:15-17:20 *Abstract Rb17* Marcus DE JONG, Amsterdam *(in person)*
MRI of Retinoblastoma with Orbital Cellulitis and Impact of Inflammation on Interpretation of Metastatic Risk Factors

17:20-17:25 *Abstract Rb3* Annette MOLL, Amsterdam *(in person)*
At What Age Could Screening for Familial be Discontinued? A Systematic Review

17:25-17:30 Discussion

Retinoblastoma 5 Associated Poster with Online Rapid-Fire Presentation

Abstract Rb28 Geoffrey WABULEMBO, Vienna & Kampala *(in person)*
Hurdles in Retinoblastoma Management in Uganda

18.45 – 22.30hrs CRUISE & GALA DINNER (join the landing stage at 18.15hrs; boat leaves at 18.45hrs sharp)

Saturday 4 September 2021
ISGEDR 2021
Ocular Genetics Sessions

08.00-9.00 hrs WELCOME COFFEE

MORNING SESSIONS **Auditorium**

9:00-10:20 hrs Ophthalmic Genetics 5 Consortia & Strategies in Ophthalmic Genetics

Moderators (in person) Mark PENNESI & Alex LEVIN

9:00-9:08 *Abstract OcGen13* Mark PENNESI, Portland, OR *(in person)*
Accelerating Research in Rare Inherited Retinal Disorders (IRDs) Through an International Consortium

9:08-9:16 *Abstract OcGen30* Emily KRAUSS, Philadelphia, PA *(virtual)*
Experiences of Genetic Testing Among Individuals with Retinitis Pigmentosa

9:16-9.24 *Abstract OcGen54* Orla GALVIN, Dublin *(virtual)*
Inherited Retinal Degeneration Genetic Testing Landscape Study Europe and Australia

9:24-9:32 *Abstract OcGen12* Kari BRANHAM, Ann Arbor *(virtual)*
FFB Consortium Gene Poll: An International Assessment of IRD Gene Distribution and IRD Clinical Practices

9:32-9:40 *Abstract OcGen26* Fulya YAYLACIOGLU TUNCAY, Ankara *(virtual)*
Practice Patterns and Needs Assessment of Ophthalmologists for Inherited Eye Disorders: Do We Need the Subspecialty of Ophthalmic Genetics?

9:40-9:48 *Abstract OcGen17* Alex LEVIN, Rochester, NY *(in person)*
Can the Low-Income Countries Have Ocular Gene Therapy?

9:48-09.56 *Abstract OcGen10* Giacomo Maria BACCI, Florence *(in person)*
***RPE65*-related IRDs and Gene Therapy: a Multicentre Narrative Medicine Approach to Investigate Patients', Caregivers', and Physicians' Expectations**

09:56-10.04 *Abstract OcGen72* Nieraj JAIN, Atlanta, GA (*virtual)*
Telemedicine-Based Approach to Caring for Patients with Inherited Retinal Diseases: Patient Satisfaction and Diagnostic Testing Completion Rates

10.04-10:20 Discussion

Ophthalmic Genetics 5 Associated Posters with Online Rapid-Fire Presentations

Abstract OcGen33 Caroline IBERG, Strasbourg *(virtual)*
Patient Pathways and Actors in Rare Eye Diseases: the French Example, French Rare Sensory Disease Network SENSGENE

Abstract OcGen70 Stacy PARTIN, Atlanta, GA *(virtual)*
Readability, Suitability and Quality of Online Health Information for Retinitis Pigmentosa & Retinitis Pigmentosa Treatment Options

Abstract OcGen82 Dorothée LEROUX, Strasbourg *(virtual)*
ERN-EYE, Latest Advances of the Network to Better Serve Patients' Needs

10:20-11:00 hrs Ophthalmic Genetics 6 Anterior Segment, Ocular Surface & Development

Moderators (in person) Ken NISCHAL & Christina GERTH-KAHLERT

10:20-10:26 *Abstract OcGen59* Ken NISCHAL, Pittsburgh, PA *(in person)*
Congenital Primary Aphakia Associated with Microphthalmia with Linear Skin Defects Syndrome (MLS)

10:26-10:32 *Abstract OcGen63* Lev PRASOV, Ann Arbor, MI *(virtual)*
Absence/Hypoplasia of Extraocular Muscles Associated with Anterior Segment Dysgenesis

10:32-10.38 *Abstract OcGen66* Phattrawan PISUCHPEN, Bangkok *(virtual)*
Identification of a Novel *PRDM5* Mutation in a Family with Brittle Cornea Syndrome

10:38-10:44 *Abstract OcGen18* Madlen RESCHKE, Berlin *(virtual)*
Ligneous Conjunctivitis and Other Systemic Manifestations of Plasminogen Deficiency, Caused by Variants in the *PLG* Gene

10:44-10:52 *Abstract OcGen65* Hannah SCANGA, Pittsburgh, PA *(in person)*
Linear Sebaceous Nevus Syndrome due to a Somatic *KRAS* A146T Variant

10.52-11:00 Discussion

Ophthalmic Genetics 6 Associated Posters with Online Rapid-Fire Presentations

Abstract OcGen90 Britta VT BRÜWILER, Zürich *(in person)*
Genotype-Phenotype Spectrum in Patients with Novel Variants in the *ADAMTSL4* Gene

Abstract OcGen20 James PLOTNIK, Omaha, NE *(virtual)*
Progressive Bilateral Nuclear Cataracts Associated with Cerebellar-Facial-Dental Syndrome: a Case Report, Literature Review and Identification of a New Genetic Variant

Abstract OcGen23 Olivia ZIN, São Paulo *(virtual)*
Novel Mutation in *CRYBB3* Causing Pediatric Cataract and Microphthalmia

Abstract OcGen64 Ken NISCHAL, Pittsburgh, PA *(in person)*
Congenital Corneal Opacification in 22q Deletion Syndrome

Abstract OcGen96 Fabiola CERONI, Bologna & Oxford *(virtual)*
The Retinoic Acid Pathway and Developmental Eye Anomalies: Expanding the Genetic and Phenotypic Spectra

Abstract OcGen52 Jana MORAVÍKOVÁ, Prague *(in person)*
Confirmation of n.37C>T in *MIR204* as a Cause of Retinal Dystrophy, Iris Coloboma and Cataract

11:00-11:30 COFFEE BREAK & POSTER VIEWING

11:30-12:30 hrs Ophthalmic Genetics 7 Optic Neuropathies & Strabismus

Moderator (in person) Rola BA-ABBAD & H Viet TRAN

11:30-11:36 *Abstract OcGen21* Lea KOVAČ, Ljubljana *(virtual)*
Reversible Bilateral Vision Loss Mimicking LHON in a Patient with Exacerbation of Cystic Fibrosis and *OPA3* Gene Mutation

11:36-11:42 *Abstract OcGen103* Rola BA-ABBAD, Riyadh *(in person)*
Identifying New Candidate Genes in Patients with Hereditary Optic Neuropathy

11:42-11.48 *Abstract OcGen98* Katharina VALENTIN, Graz *(virtual)*
Is it LHON?

11:48-11:54 *Abstract OcGen91* Louise F PORTER, Strasbourg *(in person)*
A Novel Homozygous Variant in *HMBS* is Associated with Optic Atrophy and Pyramidal Features

11:54-12:00 *Abstract OcGen97* Sanja PETROVIC PAJIC, Ljubljana & Belgrade *(in person)*
Long-Term Follow-Up of Two Patients with LHON Caused by *DNAJC30* Mutation

12:00-12:06 *Abstract OcGen75* Benjamin Manuel IBANEZ, Makati City *(virtual)*
Autonomic Manifestations in Congenital Cranial Dysinnervation Syndromes

12:06-12:12 *Abstract OcGen62* Eva ROOMETS, Tallinn & Helsinki *(virtual)*
Carbonic Anhydrase II Deficiency: A Rare Cause of Infantile Optic Nerve Atrophy

12:12-12:18 *Abstract OcGen47* Zia CHAUDHURI, Delhi *(virtual)*
Phenotype Analysis of 75 Informative Families of Indian Origin with Primary Concomitant Horizontal Strabismus

12:18-12:24 *Abstract OcGen58* Hannah SCANGA, Pittsburgh, PA *(in person)*
Expanding the Phenotype of *DNM1L*-Related Autosomal Dominant Optic Atrophy (OPA5): Report of a Novel Variant with Early-onset Atrophy and Significant Intrafamilial Variability

12:24-12:30 Discussion

Ophthalmic Genetics 7 Associated Posters with Online Rapid-Fire Presentations

Abstract OcGen43 Fatma KIVRAK PFIFFNER, Zürich *(virtual)*
Homozygosity for a Novel *DOCK7* Variant Due to Segmental Uniparental Isodisomy of Chromosome 1 in a Child with Infantile Epileptic Encephalopathy and Cortical Vision Impairment

Abstract OcGen86 Béatrice BOCQUET, Montpellier *(in person)*
Relative Frequencies of Inherited Retinal Dystrophies and Optic Neuropathies in Southern France: From the Clinic to Genetics

Abstract OcGen73 Sarah HULL, Auckland *(virtual)*
Congenital Posterior Pole Tumour in Disseminated Infantile Myofibromatosis due to a Rearrangement in *PDGF*

12:30-13:30 LUNCH BREAK

AFTERNOON SESSIONS **Auditorium**

13.30-13.40 hrs Elias I TRABOULSI, Cleveland, OH *(virtual)*
Introduction of the Adolphe Franceschetti Lecturer

13.40-14.20 hrs Ian MACDONALD, Alberta *(in person)*
FRANCESCHETTI LECTURE: Choroideremia – Lessons from Research and Clinical Trials

14.20-15.20 hrs Symposium: Mitochondrial Genetics & Optic Neuropathies

Moderators (in person) Guy LENAERS & Andrea SODI

14.20 - 14.35hrs Patrick YU-WAI-MAN, London & Cambridge *(virtual)*
New Insights into Leber Hereditary Optic Neuropathy

14.35-14.50 Catherine VIGNAL-CLERMONT, Paris *(virtual)*
Gene Therapy for Leber Hereditary Optic Neuropathy

14.50-15.05 Guy LENAERS, Angers *(in person)*
New insights into Dominant Optic Atrophy

15.05-15.20 Marcela VOTRUBA, Cardiff *(virtual)*
Dominant Optic Atrophy - Opportunities for Advanced Therapies

15.20 - 15.30hrs AU REVOIR - END **Francis MUNIER, Lausanne** ***(in person)***

Thursday 2 September 2021

SATELLITE MEETING

European Retinoblastoma Group (EURbG) 2021 Meeting

MEMBERS ONLY **Salle Olympie**

11.30 hrs **Introduction**
by the EURbG chairs Maja BECK-POPOVIC & Annette MOLL
Welcome
EURbG update
EURbG papers policy
EURbG data & data commons
Collaborations

12.00-16.00 hrs ***Feedback from WGs: 15 min presentation and 15 minutes discussion***
Study results should not be presented on Thursday, please present those on Friday in the ISGEDR Rb sessions
Topics:
What happened last year?
How many WG members?
New WG leader?
Future plans

12.00 hrs **WG1 Conservative Therapy**
Chairs Francis MUNIER & Maja BECK-POPOVIC, Switzerland

12.30-13.30 hrs **LUNCH**

13.30 hrs **WG2 Non-Conservative Therapy**
Chair François DOZ, France

14.00 hrs **WG 3 Late Effects, Quality**
Chair Annette MOLL, The Netherlands

14.30 hrs **WG 4 Constitutional Mutation Analysis and Genomics, Basic Research, Tissue Banking**
Chairs Dietmar LOHMANN & Petra KETTELER, Germany

15.00 hrs **WP7 Imaging/ERIC group**
Chair Pim DE GRAAF, The Netherlands

15.30 hrs **WG5 Dissemination of the Project, Networking, Education and Web Resources**
Chairs Tomas KEPAK, Czech Republic, Rejin KEBUDI, Turkey, Manoj PARULEKAR & Helen JENKINSON, UK

WG6 Parental Groups
Chairs Catherine BOTHOREL & Erika LEIMAN, France

New working group leaders

16.00 hrs **END OF EURbG Meeting & COFFEE BREAK**

Friday 3 September 2021

SATELLITE MEETING

Réunion de la Société de la Génétique Ophtalmologique Francophone (SGOF) 2021

MEMBERS ONLY ***Meeting held in French*** **Salle Olympie**

09.15 hrs **Introduction**
Par la Présidente de la SGOF Isabelle S AUDO

09.30 hrs **Présentations de Cas Cliniques**
modérées par Isabelle S AUDO, Isabelle MEUNIER & Xavier ZANLONGHI

12.00 hrs **Réunion d'Affaires de la SGOF**

12.30-13.30 hrs **DÉJEUNER**

13.30 hrs **Présentations de Cas Cliniques**
modérées par Isabelle AUDO, Isabelle MEUNIER & Xavier ZANLONGHI

16.00hrs **FIN DE LA RÉUNION DE LA SGOF & PAUSE CAFÉ**