Meeting of the International Society for Genetic Eye Diseases & Retinoblastoma ISGEDR

Halifax, Nova Scotia, Canada
August 6-8, 2015
Welcome to Halifax and to Your Meeting …

Dear ISGEDR and other members of the Ophthalmic and Genetics communities,

We have the honor of welcoming you to the ISGEDR 2015 conference in Halifax, Nova Scotia, Canada. August is one of the best months to visit Nova Scotia and Halifax is a friendly seacoast city that has retained much of its historical charm. One can easily walk from the conference venue located at the edge of the downtown area to many historical sites, hotels, restaurants, shops and, of course, the lively waterfront.

The conference is held at Dalhousie University in one of the Faculty of Medicine’s main auditoriums in the Sir Charles Tupper Medical Building (5850 College Street). We invite you to the Opening Reception at the Canadian Museum of Immigration at Pier 21, and the Gala Dinner at the Maritime Museum of the Atlantic. Both venues are located on the waterfront, and each offering a different perspective on Nova Scotia’s rich maritime heritage locally and internationally. The museum’s access will be reserved for the ISGEDR members during each event, and catering will be available on-site.

We look forward to what promises to be a very exciting conference, and we hope that you will have a memorable visit with us.

Johane Robitaille, M.D.  
Meeting Chair, and Head of Local Organizing Committee

David A. Mackey, M.D.  
President, ISGEDR

Elias I. Traboulsi, M.D.  
Executive Vice-President, ISGEDR

Brenda Gallie, M.D.  
Member, Scientific Program Committee  
Kate Paton, M.D.  
Member, Scientific Program Committee

Ian MacDonald, M.D.  
Member, Scientific Program Committee  
Arlene Drack, M.D.  
Member, Scientific Program Committee

Alex Levin, M.D.  
Member, Scientific Program Committee  
Meghan Marino, M.S., L.G.C.  
Member, Genetic Counseling Committee
The International Society for Genetic Eye Diseases
ISGEDR

Mission Statement

To bring together individuals interested in the field of genetic diseases of the eye and in Retinoblastoma
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To provide a forum for researchers in the field of genetic diseases of the eye to share information
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To promote international collaborations in the study of genetic diseases of the eye and in Retinoblastoma
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To disseminate scientific knowledge through international conferences and through its official publication, Ophthalmic Genetics
The International Society for Genetic Eye Disease & Retinoblastoma wishes to thank the following sponsors who have generously supported the 2015 ISGEDR Meeting

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Sincere Thanks to

Sandy Wong
ISGEDR Coordinator,
Cole Eye Institute
Cleveland Clinic
Cleveland, USA

Sandi Leaf
ISGEDR 2015 LOC
Department of Pharmacology
Dalhousie University
Halifax, Canada

Nadeem Traboulsi
CME Department
Cleveland Clinic
Cleveland, USA

And the following Haligonian volunteers:
From the Orthoptics Clinical Vision Science Program, Dalhousie University
Jessica Wood
Jenny Faries
Rebecca Fels
Lianne Esmores

Amr Zaki
Resident, Department of Ophthalmology and Visual Sciences
Dalhousie University
Halifax, Nova Scotia

Janet Brownell
Administrative Assistant, Finances
Department of Ophthalmology and Visual Sciences
Dalhousie University
Halifax, Nova Scotia
Named Lecture Speakers for ISGEDR 2015

**Franceschetti Lecture:** Elise Héon, *Toronto, Canada*

“Lessons Learned From Ocular Genetics: The Power of Phenotyping”

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**Elise Héon, MD, FRCS(C)**

*Professor, Department of Ophthalmology and Vision Sciences, University of Toronto*

Director of the Ocular Genetics Program

Mira Godard Chair in Vision Research

Associate Surgeon-in-Chief for Research

The Hospital for Sick Children

Toronto, Canada

former Chief of Ophthalmology (2003-2013)

Dr. Héon is a Clinician-Scientist in the field of Ocular Genetics, Director of the Eye Genetics Program and Senior Associate Scientist at The Hospital for Sick Children Research Institute in the program of Genetics and Genomics Biology. Dr. Héon carries both clinical and basic research projects. Her laboratory, supported by peer-reviewed grants, is involved in the genetic analysis of inherited eye disorders mostly retinal dystrophies.

Her clinical work focuses on the management of hereditary eye diseases, which include hereditary cancer (retinoblastoma) and other non-cancerous blinding conditions such as retinitis pigmentosa. Dr. Héon’s role as Director of the Ocular Genetics program is to ensure that patients are provided with state of the art global care. This program is unique in Canada and part of only a few in the world. In addition to her numerous administrative activities, Dr. Héon has a busy teaching schedule dedicated to undergraduate, graduate and post graduate students as well as clinical and research Fellows. Through her research Dr. Heon works to better understand disease characteristics and mechanisms with the goal of improving outcome and quality of life of patients.
François Lecture: Richard Weleber, Portland, USA
“Clinical Trials for Inherited Retinal Diseases: New Endpoints, Analyses”

Richard G Weleber, MD, FACMG
Professor, Department of Ophthalmology and Department of Molecular and Medical Genetics,
Director of the Oregon Retinal Degeneration Center
Casey Eye Institute of Oregon Health & Science University
3375 SW Terwilliger Blvd, Portland, Oregon, USA

Dr. Weleber is a Clinician-Scientist in the field of Ocular Genetics, retinal degenerations, visual electrophysiology, visual field methodology, and design and conduct of clinical trials. He founded the first visual electrophysiology service at OHSU in 1974 and was Director of the Visual Function Service for 39 years. He took two fellowships in Genetics—one at OHSU and the other at University of Colorado Medical Center. Since 1984, he has been the Director of the Oregon Retinal Degeneration Center at the Casey Eye Institute. He is board certified in both Ophthalmology and Clinical Genetics and was one of the Founding Members of the Ophthalmic Genetics Study Club, the International Society for Genetic Eye Disease, and the American College of Medical Genetics. Over the past 20 years, he has trained nine ophthalmologists in Ophthalmic Genetics, all of whom, following the Fellowship, accepted appointments at academic universities in the United States, England, and British Columbia.

With 40 years’ experience in the study of inherited diseases of the retina, Dr. Weleber has evaluated, provided consultation, and cared for over 10,000 patients with a variety of ophthalmic genetic diseases. In his clinic he regularly identifies, screens and oversees the genetic testing of patients with retinal dystrophies, including retinitis pigmentosa and allied disorders. He has an international reputation in retinal electrophysiology and the evaluation of patients with retinal disease. He has a total of 197 peer-reviewed publications and numerous book chapters. Other major interests include improving the methodology for the evaluation of kinetic and static visual fields, volumetric measures of visual fields from 3D modeling of the Hill of Vision, and the development of clinical trial endpoints. He has received numerous honors including the Franceschetti Award/Lecture from the ISGEDR and the Adachi Award/Lecture from the ISCEV.
Ellsworth Lecture: Junyang Zhao (China)
“Retinoblastoma in China: Past, Present and Future”

Junyang Zhao, MD
Senior Staff of Ophthalmology Department
Beijing Children's Hospital
Capital Medical University
Beijing, China

Dr. Junyang Zhao earned his MD from the Peking University at 1992 and completed his resident training at Beijing Tongren Hospital from 1992 to 1997. He became an attending ophthalmologist from then and received Master of Science Degree at 2004 form Capital Medical University. Dr. Junyang Zhao became the board member of Chinese Strabismus and Pediatric Ophthalmology Association from 2013. Dr. Junyang Zhao started focus on retinoblastoma from 2006 and treated more than 2000 patients by now. He set up a collaboration containing more than 20 hospitals all over China. He received around 300 new retinoblastoma cases each year and greatly improved the survival rate, saved many eyes for these patients. He moved to Beijing Children's Hospital from December 2014 and still on retinoblastoma.
Thursday August 6th

8:00 Registration and breakfast

9:00 Welcome
Drs. Robitaille and Mackey

9:15 Official opening
Faculty of Medicine Dalhousie University

9:30 Ophthalmic Genetics Session 1 (Free papers)
Moderator: Graeme Black

9:30 IMPG1 AND IMPG2 CAUSE MACULAR VITELLIFORM DYSTROPHIES. CHRISTIAN HAMEL, GAEL MANES, BEATRICE BOCQUET, ALMUNEDA AVILA-FERNANDEZ, SANDRO BANFI. Montpellier, France

9:42 PRESERVED VISUAL FUNCTION IN RETINAL DYSTROPHY DUE TO HYPOMORPHIC RPE65 MUTATIONS. ANTHONY MOORE, SARAH HULL, RAJARSHI MUKHERJEE, ANTHONY G ROBSON, MICHEL MICHAELIDES, GRAHAM E HOLDER, ANDREW R WEBSTER. San Francisco, California.

9:54 GENOTYPE-PHENOTYPE CORRELATIONS IN CARRIERS OF PRPF31 MUTATIONS. BIRGIT LORENZ, MONIKA ANDRASSI-DARIDA, WADIM BOWL, CHRISTOPHER FRIEDBURG, HANNO J. BOLZ. Giessen, Germany

10:06 THE NATURAL HISTORY OF THE PROGRESSION OF ATROPHY SECONDARY TO STARGARDT DISEASE STUDIES (PROGSTAR): YEARLY PROGRESSION RATE OF ATROPHIC LESIONS IN THE RETROSPECTIVE STUDY. RUPERT STRAUSS, ALEX HO, BEATRIZ MUNOZ, MOHAMED AHMED, SRINIVAS SADDA. Baltimore, Maryland

10:18 THE IMPACT OF FOUNDER EFFECT ON GENOTYPIC-PHENOTYPIC CORRELATIONS IN THE FIRST COMPREHENSIVE SCREENING OF AUTOSOMAL DOMINANT RETINITIS PIGMENTOSA (ADRP) GENES IN THE FRENCH-CANADIAN POPULATION. RAZEK GEORGES COUSSA, CHRISTINA CHAKAROVA, FARES ANTAKI, AYESHA KHAN, IRMA LOPEZ, HUANAN REN, NAUSHIN WASEEM, KUNKA KAMENAROVA, SHOMI S BHATTACHARYA, ROBERT K KOENEKOOP. Montreal, Canada.

10:30 PNPLA6-OPTHATHIES: AN EMERGING SPECTRUM OF CONGENITAL, CHILDHOOD, AND ADULT NEURODEGENERATIVE CONDITIONS. ROBERT B. HUFNAGEL, SARAH HULL, ROBERT A. SISK, GAVIN ARNO, CORINNE STOETZEL. Cincinnati, Ohio.

10:42 AUTOSOMAL RECESSIVE MICROCEPHALY WITH CHORIORETINOPATHY (MCMR) AND MUTATIONS IN TUBGCP4, ENCODING A MEMBER OF THE GAMMA-TUBULIN RING COMPLEX
GAMMA TURC. HELENE DOLLFUS, SOPHIE SCHEIDECKER, CHRISTELLE ETARD, LAURENCE HAREN, SARAH HULL. Strasbourg, Germany.

10:54 NOVEL MUTATIONS IN KIF11 GENE IN JAPANESE PATIENTS WITH FEVR AND MLCRD. HIROYUKI KONDO, ITSUKA MATSUSHITA, EIICHI UCHIO, SHUNJI KUSAKA. Kitakyusyu Japan

11:06 THE EXTENDED MOLECULAR HETEROGENEITY OF FAMILIAL EXUDATIVE VITREORETINOPATHY AND MICROCEPHALY. SARAH HULL, GAVIN ARNO, PIA OSTERGAARD, LOUISE BICKNELL, CAROL-ANNE MARTIN. London, United Kingdom

11:18 PHENOTYPIC SPECTRUM AND MOLECULAR DIAGNOSIS OF FAMILIAL EXUDATIVE VITREORETINOPATHY (FEVR) AND RELATED CONDITIONS IN AN INTERNATIONAL DATABASE. JOHANE ROBITAILLE, KARIN WALLACE, JILL BEIS, ROXANNE GILLET, MARISSA LEBLANC, DANIEL GASTON, MATHEW NIGHTINGALE, MICHAEL P MACKY, CHRISTINE MCGILLIVRAY, CHRISTOPHER MCMASTER, KAREN BEDARD. Halifax, Nova Scotia Canada

11:30 Break/Poster Viewing

12:00 Franceschetti Lecture: Elise Héon, Toronto, Canada Lessons learned from Ocular Genetics: the Power of Phenotyping
Introduction by Elias I. Traboulsi

12:45 Lunch/Poster Viewing

13:45 Retinoblastoma Session 1 (Free papers)
Moderator: Bruce Crooks

13:45 GOOGLING DR GOOGLE: LEUKOCORIA, RETINOBLASTOMA AND THE WORLD WIDE WEB. SANDRA STAFFIERI, ALEX HEWITT, LISA KEARNS, DAVID MACKY. Melbourne, Australia

13:57 BUILDING A LEARNING HEALTH SYSTEM FOR RETINOBLASTOMA. BRENDA GALLIE, TRAN TRUONG, JUSTIN LIU, YULIYA GAVRYLYUK. Toronto, Ontario

14:09 WHAT DIFFERENCE DOES NEXT GENERATION SEQUENCING MAKE TO RETINOBLASTOMA GENETIC TESTING? HILARY RACHER, SHARLENE KOROSCIL, CRYSTAL D’SILVA, XUE WU, CHAO ZHANG, XIAOFANG HUANG, FRANNY JEWETT, YANG SHAO, BRENDA GALLIE. Toronto, Ontario

14:21 MANAGEMENT AND OUTCOMES OF RETINOBLASTOMA IN THE REPUBLIC OF IRELAND. WE FONG SIAH, BERNADETTE LANIGAN, BRENDA GALLIE, MICHAEL O’KEEFE. Republic of Ireland
14:45  Ophthalmic Genetics Session 2 (Free papers)
     Moderator: David Mackey

14:45  NOVEL FOXC1 MUTATIONS WITHIN THE DNA-BINDING FORKHEAD DOMAIN RESULT IN A CONGENITAL OCULAR PHENOTYPES OF AXENFELD-RIEGER SYNDROME. HANNAH L. SCANGA, BHARESH CHAUHAN, ANAGHA MEdSINGE, SUNEETA MADAN-KHETARPAL, KEN K. NISCHAL.  Pittsburgh, PA.

14:57  FOXC1 MUTATION WITH SIGNIFICANT POSTNATAL IRIS GROWTH AND MODULATION. MERINA THOMAS, BEHRAD Y MILANI, IRENE H MAUMENEE, JAVANEH ABBASIAN. Chicago, Illinois

15:09  FERRITIN LIGHT CHAIN GENE MUTATION IN A LARGE AUSTRALIAN FAMILY WITH HEREDITARY HYPERFERRITINEMIA-CATARACT SYNDROME. SEYHAN YAZAR, MARIA FRANCHINA, KATHRYN BURDON, DAVID A MACKEy. Nedlands WA, Australia

15:21  CONGENITAL EYE MOVEMENT DISORDERS: IMPROVING PHENOTYPIC DESCRIPTIONS AFTER GENETIC ANALYSIS- ANOTHER LOOK AT MOEBIUS SYNDROME. DARREN OYSTRECK, SARAH MACKINNION, CAROLINE ANDREWS, WAI-MAN CHAN, DAVID HUNTER. HALIFAX, NOVA SCOTIA, CANADA

15:33  RETURN OF RESULTS FOR IMPUTED MYOCILIN MUTATIONS IN GWAS STUDIES. DAVID A MACKEY, PUYA GHARAHKHANI, KATHRYN P BURDON, ALEX W HEWITT, MATTHEW H LAW. Perth, Australia

15:45  Break/Poster Viewing

16:15  Symposium Ethics and Consenting
     Moderator: Johane Robitaille

16:15  ETHICAL CONSIDERATIONS OF INNOVATIVE THERAPY IN CHILDREN WITH UNILATERAL RETINOBLASTOMA. CHRYSSA MCALISTER, BRENDA L GALLIE.  Toronto, Ontario Canada

16:45  PLANNING TO OFFER TARGET AND INCIDENTAL GENOMIC RESULTS. CONRAD FERNANDEZ  Halifax Nova Scotia, Canada

17:15  OCULAR GENETICS: WHO SHOULD DO IT? ALEX LEVIN Philadelphia, USA

18:00  Bus pick up to Pier 21

18:30-21:00  Opening Reception - Pier 21
7:30  **Ophthalmic Genetics Session 3 (Free papers)**
      Moderator: Godfrey Heathcote

7:30 **MYSTERY CASE.** ANN Hoskin-Mott. *Halifax, Nova Scotia, Canada*

7:42 **A NOVEL MUTATION IN PRPH2/RDS GENE AND ITS SPECTRUM OF PHENOTYPIC MANIFESTATIONS IN A SYRIAN FAMILY WITH AUTOSOMAL DOMINANT RETINITIS PIGMENTOSA (ADRP).** SUSAN WAKIL, RAZEK GEORGES COUSSA, CHRISTINA CHAKAROVA, VINCENT SUN, AYESHA KHAN, IRMA LOPEZ, HUANAN REN, KUNKA KAMENAROVA, SHOMI S. BHATTACHARYYA, ROBERT K. KOENEOOP. *Quebec, Canada*

7:54 **A CASE OF JALILI SYNDROME.** MARK PENNESI, AKSHAY THOMAS, ZHONGQI GE, RUI CHE. *Portland, Oregon.*

8:06 **PHENOTYPIC VARIATION IN AFFECTED MEMBERS OF A FAMILY HARBORING AN NDP GENE MUTATION.** IRINA DE LA HUERTA, JACQUE L. DUNCAN, ANNE SLAVOTINEK, ANTHONY T. MOORE, ALEJANDRA G. DE ALBA CAMPOMANES. *San Francisco, California*

8:18 **RECOGNIZING LYONIZATION IN FEMALE HETEROZYGOTES OF X-LINKED OCULAR DISEASE IN THE ERA OF MOLECULAR DIAGNOSTICS.** BART P. LEROY, ELKE O. KREPS, ELFRIDE DE BAERE, JULIE DE ZAEYTJJD. *GHENT, BELGIUM*

8:30 **François Lecture:** Richard Weleber, *Portland, USA*
      “Clinical Trials for Inherited Retinal Diseases: New Endpoints, Analyses for Efficacy, and Model for Progression”
      Introduction by Bart Leroy

9:15 **Break/Poster Viewing**

9:45 **Symposium on Congenital Cataracts**
      Moderator: Irene Maumenee

9:45 **PHENOTYPE-GENOTYPE CORRELATIONS IN PEDIATRIC CATARACT.** ARIF O KHAN, *Riyadh, Saudi Arabia*

10:15 **MOLECULAR TESTING FOR CONGENITAL CATARACT.** GRAEME CM BLACK, RACHEL GILLESPIE, IAN CHRISTOPHER LLOYD, JILL CLAYTON-SMITH, JANE ASHWORTH. *Manchester, United Kingdom*

11:15 Ophthalmic Genetics Session 4 (Free papers)
Moderator: Birgit Lorenz


11:39 Brothers with Congenital Ocular Motor Apraxia, Juvenile Nephronophthisis and Mild Cerebellar Defects. Stephanie Chan, Jaspreet Rayat, Yves Sauvá, Ian M. Macdonald. Alberta, Canada


12:05 Matthew Herder (Canada)
Orphan drug policies: Lessons learned?

12:30 ISGEDR Business Meeting

13:00 Lunch/Poster Viewing

14:00 Symposium Retinoblastoma Imaging in RB - Driving Clinical Innovations
Moderator: Brenda Gallie

14:00 Francis Munier (Switzerland)

14:25 Junyang Zhao (China)
EUA and the retina camera application on retinoblastoma diagnosis

14:50 Cynthia Vandenhoven (Canada)

14:00  Ophthalmic Genetic Counselor's Breakout Session
       Sponsored by Foundation Fighting Blindness, USA
       Moderator: Meghan Marino
14:00  Introduction. Meghan Marino
14:02  Ophthalmic GC Collaborations and Community Outreach. Karmen Truzpek
14:22  Billing Challenges, Barriers to Services, And Solutions. Dianna Wheaton And Kari Branham
14:42  Counseling/Testing Standards & NSGC Practice Guidelines. Meghan Marino
15:02  Ethical Dilemmas. Jill Beis
15:17  Closing Remarks And Future Plans. Meghan Marino

16:00 - 19:00 Trip to Peggy’s Cove - Bus pick up at 16:00 from Conference Venue and 19:00 from Peggy’s Cove (return 20:00 in Halifax)

FREE EVENING IN HALIFAX
Saturday August 8\textsuperscript{th}

8:00  \textbf{Symposium: Developmental Genes and Eye Malformations}  
\textit{Symposium in memory of Mette Warburg}  
Moderator: Elias I. Traboulsi

8:00  METTE WARBURG. ELIAS I. TRABOULSI. Cleveland, Ohio

8:10  \textbf{ADAMTS PROTEINS, IMPORTANT PARTICIPANTS IN OCULAR MORPHOGENESIS AND GENETIC DISORDERS.} SUNEEL APTE. Cleveland, Ohio

8:32  \textbf{CONSERVED GENETIC PATHWAYS IN MICROPHTHALMIA, ANOPHTHALMIA AND COLOBOMA.} ELENA V. SEMINA, BRETT DEML, ARIANA KARIMINEJAD, RAZIEH H. R. BORUJERDI, SANAA MUHEISEN, LINDA M. REIS. Milwaukee, USA

8:54  \textbf{RECESSIVE MICROPHTHALMIA SYNDROMES.} NICOLA RAGGE. Oxford, UK

9:16  \textbf{THE TRANSCRIPTION FACTOR FOXC1 DIRECTLY REGULATES PROSTAGLANDIN RECEPTORS: A POTENTIAL EXPLANATION FOR RECALCITRANT GLAUCOMA IN AXENFELD-RIEGER SYNDROME.} MICHAEL WALTER, TIM FOOTZ, LANCE DOUCETTE. Alberta, Edmonton, Canada

9:38  \textbf{NEW POSTNATAL THERAPEUTIC OPTIONS FOR CONGENITAL OCULAR MALFORMATIONS.} CHERYL GREGORY-EVANS, XIA WANG, KISHOR WASAN, KEVIN GREGORY-EVANS. Vancouver, BC

10:00  \textbf{Break/Poster Viewing}
10:30    Ellsworth Lecture: Junyang Zhao (China)
Retinoblastoma in China: Past, Present and Future
Introduction by Brenda Gallie

11:45    Ophthalmic Genetics Session 5 (Free papers)
Moderator: Bart Leroy
11:45    ANTISENSE OLIGONUCLEOTIDE SELECTIVELY TARGETING THE P23H VARIANT OF
RHODOPSIN FOR THE TREATMENT OF P23H RHODOPSIN-MEDIATED ADRP.  MICHAEL L.
MCCALEB, ALI JAZAYERI, RAECHEL PERALTA, SHULING GUO, BRETT P. MONIA, SUE F. MURRAY.
Carlsbad  CA

11:57    AN EX VIVO GENE THERAPY APPROACH IN THE TREATMENT OF X-LINKED
RETINOSCHISIS. KEVIN GREGORY-EVANS, EMRAN BASHAR, ISHAQ VIRINGIPURAMPEER,
ANDREW METCALFE.  Vancouver, BC, Canada.

12:09    VERSICAN ENHANCES VIRAL-MEDIATED GENE THERAPY BY ACTIVATING JAK/STAT
SIGNALING.  RICHARD HURWITZ, PATRICIA AKINFENWA, WESLEY BOND, CRISTHIAN ILDEFONSO,
MARY HURWITZ.  Houston, Texas.

13:21    GENOME EDITING WITH CRISPR/CAS9 IN ZEBRAFISH: AN EVOLVING TOOL TO MODEL
RARE GENETIC RETINAL DISEASES FOR DRUG DISCOVERY.  JASON BERMAN, SERGEY
PRYKHOZHIJ, SHELBY STEELE PHD, VINOTHKUMAR RAJAN, TENILLE FLEISHMAKER.  Ghent,
Belgium

14:33    GENE THERAPY RESEARCH REPORTING TRENDS IN THE USA, UK AND CANADA AND
IMPLICATIONS FOR CLINICAL COMMUNICATION.  STEPHANIE KOWAL, SHELLY BENJAMINY, IAN
MACDONALD, TANIA BUBELA.  Edmonton, Canada

12:45    Lunch/Poster Viewing

13:45    Retinoblastoma Session:
The Debate: Cell of Origin in RB - David Cobrinik vs. Brenda Gallie
Moderator: Kevin Gregory-Evans
14:30 Symposium Gene Therapy
Moderators: Ian MacDonald and Arlene Drack
14:30 VALIDATION AND CLINICAL APPLIATION OF A NOVEL MOBILITY TEST TO ASSESS FUNCTIONAL VISION IN PATIENTS WITH INHERITED RETINAL DEGENERATION. DANIEL CHUNG, SARAH MCCAGUE, JENNIFER WELLMAN, ZI-FAN YU, SATHA THILL. Philadelphia, PA

14:50 CRITICAL ANALYSIS OF TESTS SEPARATING ROD AND CONE VISUAL PATHWAYS AS OUTCOME PARAMETERS IN GENE THERAPEUTIC TRIALS OF HEREDITARY RETINAL DISEASES. BIRGIT LORENZ. Giessen, Germany

15:10 MANAGING EXPECTATIONS IN GENE THERAPY TRIALS AND LESSONS LEARNED ALONG THE WAY. ARLENE DRACK. Iowa City, Iowa

15:30 PARTICIPANT SELECTION FOR A PHASE 1 CLINICAL TRIAL OF AAV2-REP IN CHOROIDEREMIA. IAN M MACDONALD, IOANNIS S DIMOPOULOS, STEPHANIE CHAN, GARY GOLDSAND, RIZWAN SOMANI. Edmonton, AB, Canada

15:50 UPDATE ON RETINAL GENE THERAPY TRIALS AT THE CASEY EYE INSTITUTE. MARK PENNESI, RICHARD G. WELEBER, ANDREAS K. LAUER, PAUL YANG, JOSE A. SAHEL. Portland, Oregon

16:10 CURRENT THINKING ABOUT LONGEVITY OF THE EFFECT OF SUBRETINAL GENE THERAPY FOR RPE65-RELATED LCA. BART P LEROY. Ghent Belgium

16:30 Poster Viewing

17:00 Retinal Degenerative Diseases - Patient and Family Session
Sponsored by Foundation Fighting Blindness, Canada
Panel of speakers on a variety of topics including basics of genetics, role of genetic testing and new technologies, gene therapy and gene-based therapies

19:00 Gala Dinner, Maritime Museum Reception and Visit
Tania Barragan Arevalo (Mexico)
Novel ACTG1 mutation in a Mexican patient with Baraitser-Winter syndrome

Miriam Ehrenberg (Israel)
NCF2-related chronic granulomatous disease and retinitis pigmentosa

Mary Hurwitz (USA)
Decreased expression of SKAP2 increases proliferation of retinoblastoma cells

Timothy Corson (USA)
Residual disease monitoring in a retinoblastoma patient by PCR of a novel deletion breakpoint

Mohamed H. Abdel-Rahman (USA)
Comprehensive review of the BAP1 tumor predisposition syndrome with report of three new cases

Justin Pyne (Canada)
Visual outcomes in carriers of familial exudative vitreoretinopathy (FEVR) gene mutations

Michael Ngo (Canada)
FZD4 haploinsufficiency delays recovery of retinopathy of prematurity in the ocular ischemic retinopathy (OIR) mouse model

Dianna Hughbanks-Wheaton (USA)
Retinal targeted-capture next generation sequencing and CLIA confirmation in a representative range of patients with inherited retinal degeneration: a pilot of the Texas 1000 project.

Hoda Rajabi (Canada)
Validation of a diagnostic panel comprised of ABCA4 mutations causing Stargardt disease in the Newfoundland and Labrador (NL) population

Rajani Battu (India)
Status of genetic eye research in India: An analysis of the publications in eye genetics in the last decade and the challenges associated

Heather MacDonald (Canada)
Phenotypic characterization of males with suspected X-linked juvenile retinoschisis is highly sensitive to detect RS1 mutations

Sandra Staffieri (Australia)
Clinical and molecular characterization of females affected by X-linked retinoschisis
Hannah Scanga (USA)
X-linked juvenile retinoschisis with manifestations in female carriers demonstrated by ophthalmic imaging

Choi Mun Chan (Singapore)
A family with maternally inherited diabetes and deafness (MIDD) from Singapore

Kari Branham (USA)
Integrating Genetic Testing into a Retinal Dystrophy Clinic
ACCREDITATION

As an accredited provider, Dalhousie University, CPD, designates this continuing professional development activity for up to 20.75.00 credit hours as an accredited group learning Section 1 activity as defined by the Maintenance of Certification Program of the Royal College of Physicians and Surgeons of Canada.

Thursday – 6.75  Friday – 5.75  Saturday – 8.25

Through an agreement between the Royal College of Physicians and Surgeons of Canada and the American Medical Association, physicians may convert Royal College MOC credits to AMA PRA Category 1 Credits™. Information on the process to convert Royal College MOC credit to AMA credit can be found at www.ama-assn.org/go/internationalcme.

Live educational activities, occurring in Canada, recognized by the Royal College of Physicians and Surgeons of Canada as Accredited Group Learning Activities (Section 1) are deemed by the European Union of Medical Specialists (UEMS) eligible for ECMEC®.

In keeping with CMA Guidelines, program content and selection of speakers are the responsibility of the planning committee. Support is directed toward the costs of the course and not to individual speakers.
**ISGEDR Travel Awards**

Razek Coussa, Medical Student, McGill University, *Montreal, Canada*
Susan Wakil, Ophthalmology Resident, McGill University, *Montreal, Canada*
Miriam Ehrenberg, M.D., Pediatric Ophthalmologist, *Tel Aviv, Israel*
Rajani Battu, M.D., Pediatric Retina Specialist, *Bangalore, India*

**Spark Therapeutics Genetic Counselor Travel Award**

Hannah Scanga, Genetic Counselor, Pittsburgh, USA