APPLYING GENETICS TO EYE CARE TODAY

FRIDAY, DECEMBER 6, 2019
The University Club, 1 West 54th Street, New York, NY
Welcome to Columbia Ophthalmology’s fourth annual Precision Ophthalmology™ meeting, “Applying Genetics to Eye Care Today.” We are excited to present this cutting-edge meeting focused on the role of genetics in ophthalmology. The information presented today will help guide the incorporation of DNA analysis and gene therapy into the practice of ophthalmology. Our goal is to enhance your understanding of the critical role genetic principles play in the field of ophthalmology through our discussion of clinical presentations, basic research, specific types of genetic tests, ongoing clinical trials and gene therapy. We hope that this information will lead to greater use of genetic testing by clinicians in a sound, effective, and ethical way. Through the collaborative efforts of the clinicians and scientists presenting at this meeting, we hope to provide a lively, engaging, and valuable educational experience for everyone in attendance.

CHAIR, DEPARTMENT OF OPHTHALMOLOGY

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Professor of Pathology and Cell Biology
THE ARTHUR GERARD DEVOE, MD LECTURE

William W. Hauswirth, PhD
University of Florida

“Retinal Gene Therapy for ABCA4 Stargardt Disease”

THE JOHN H. DUNNINGTON, MD MEMORIAL FUND LECTURE

Marius Üffing, PhD
University of Tübingen

“A Systems Level Investigation on AMD: Insights from the European Eye-Risk Project”

THE DONALD AND BARBARA JONAS LECTURE

Reed E. Pyeritz, MD, PhD
University of Pennsylvania

“The Paradox of Genomic Testing: Increasing Uncertainty in Interpreting and Applying the Results”

THE FLORENCE EPSTEIN TEICHER LECTURE

Simon John, PhD
Columbia University

“Experimental Characterization of Glaucoma to Suggest Novel Treatment Strategies”
LEARNING OBJECTIVES
Upon completion of this activity, attendees will be better able to:

• List the common diseases where genetic testing is appropriate
• Order the appropriate tests for these diseases and arrange for genetic counseling
• Determine if a patient’s genetic testing results make them eligible for a certain gene therapy treatment or clinical trial
• Explain to a patient how gene therapy works

ACCREDITATION STATEMENT
The Columbia University Vagelos College of Physicians and Surgeons is accredited by the Accreditation Council for Continuing Medical Education to provide continuing medical education for physicians.

AMA CREDIT DESIGNATION STATEMENT
The Columbia University Vagelos College of Physicians and Surgeons designates this live activity for a maximum of 7.0 AMA PRA Category 1 Credits™. Physicians should claim only the credit commensurate with the extent of their participation in the activity.
FRIDAY, DECEMBER 6TH, 2019

7:15–8:00 a.m.  Registration and Breakfast

8:00–10:15 a.m.  SESSION I: Genetics Workshop

MODERATED BY G. A. (Jack) Cioffi, MD

8:00–8:05 a.m.  Opening Remarks
G. A. (Jack) Cioffi, MD

8:05–8:17 a.m.  Subspecialties within Genetics
Irene H. Maumenee, MD

8:18–8:30 a.m.  Genomic Medicine at Columbia University
Vimla S. Aggarwal, MBBS, FACMG

8:31–8:43 a.m.  A Primer for Ophthalmologists: Understanding the Basics
Aliaa H. Abdelhakim, MD, PhD

8:44–8:56 a.m.  From Research Discovery to Diagnostic Testing
Rando Allikmets, PhD

8:57–9:09 a.m.  The Need for Precise Phenotyping in the Lab and Clinic
Janet R. Sparrow, PhD

9:10–9:30 a.m.  The John H. Dunnington, MD Memorial Fund Lecture
A Systems Level Investigation on AMD: Insights from the European Eye-Risk Project
Marius Üffing, PhD, Institute for Ophthalmic Research, Tübingen, Germany

9:30–9:45 a.m.  PANEL DISCUSSION

PANEL MEMBERS: Aliaa H. Abdelhakim MD, PhD, Vimla S. Aggarwal, MBBS, FACMG, Rando Allikmets, PhD, Irene H. Maumenee, MD, Janet R. Sparrow, PhD, Marius Üffing, PhD

9:45–10:15 a.m.  CASE PRESENTATIONS

9:45–9:55 a.m.  Case 1: JOAG MYOC Variant
CLINICAL PRESENTER: Salman Dar, MD
DISCUSSANT/MODERATOR: Jeffrey Liebmann, MD

9:55–10:05 a.m.  Case 2: Marfan Syndrome
CLINICAL PRESENTER: Michelle Maeng, MD
DISCUSSANT/MODERATOR: Reed E. Pyeritz, MD, PhD

10:05–10:15 a.m.  Case 3: Stickler Syndrome
CLINICAL PRESENTER: Mark Breazzano, MD
DISCUSSANT/MODERATOR: Tongalp Tezel, MD

10:15–10:40 a.m.  Break
SESSION II: Applied Genetics I
MODERATED BY Jeffrey Liebmann, MD

10:40-10:52 a.m.  Panel? WES? WGS? Which DNA test do I choose?
Stephen H. Tsang, MD, PhD

10:53-11:05 a.m.  Reading a Sequencing Report
Joy Tanaka, PhD

11:06-11:18 a.m.  The Three Cs: Counseling, Consenting, Coverage
Megan Soucy, MS, CGC

11:19-11:39 a.m.  The Florence Epstein Teicher Lecture
Experimental Characterization of Glaucomas to Suggest Novel Treatment Strategies
Simon John, PhD, Columbia University Irving Medical Center, New York, NY

11:40-11:55 a.m.  PANEL DISCUSSION
PANEL MEMBERS: Simon John, PhD, Megan Soucy, MS, CGC, Leejee H. Suh, MD, Joy Tanaka, PhD, Stephen H. Tsang, MD, PhD, Lawrence A. Yannuzzi, MD

11:55 a.m.-12:25 p.m.  CASE PRESENTATIONS

11:55 a.m.-12:05 p.m.  Case 4: Exfoliation Syndrome
CLINICAL PRESENTER: Sabine Khan, MD
DISCUSSANT/MODERATOR: C. Gustavo De Moraes, MD, PhD, MPH

12:05-12:15 p.m.  Case 5: Leber Congenital Amaurosis CRB1 Variant
CLINICAL PRESENTER: Jose R. Lima de Carvalho Junior, MD
DISCUSSANT/MODERATOR: Irene H. Maumenee, MD

12:15-12:25 p.m.  Case 6: Aniridia
CLINICAL PRESENTER: Portia E. Sirinek, MD
DISCUSSANT/MODERATOR: Xin Zhang, PhD

12:25-1:15 p.m.  Lunch

SESSION III: Applied Genetics II
MODERATED BY Brian Marr, MD

1:15-1:27 p.m.  Genome Surgery
Stephen H. Tsang, MD, PhD

1:28-1:40 p.m.  Gene Therapy Trial for the CEP290 Variant of LCA
Steven E. Brooks, MD

1:40-2:00 p.m.  The Arthur Gerard DeVoe, MD Lecture
Retinal Gene Therapy for ABCA4 Stargardt Disease
William W. Hauswirth, PhD, University of Florida, Gainesville, FL

2:00-2:15 p.m.  PANEL DISCUSSION
PANEL MEMBERS: Rando Allikmets, PhD, Steven E. Brooks, MD, G. A. (Jack) Cioffi, MD, William W. Hauswirth, PhD, Irene H. Maumenee, MD, Tongalp Tezel, MD, Stephen H. Tsang, MD, PhD
2:15–2:46 p.m.  CASE PRESENTATIONS
2:16–2:26 p.m.  Case 7: Age-related Macular Degeneration
CLINICAL PRESENTER: Marlene D. Wang, MD
DISCUSSANT/MODERATOR: Stanley Chang, MD
2:26–2:36 p.m.  Case 8: Stargardt Disease
CLINICAL PRESENTER: Vlad Diaconita, MD
DISCUSSANT/MODERATOR: Rando Allikmets, PhD
2:36–2:46 p.m.  Case 9: Axenfeld-Rieger Syndrome
CLINICAL PRESENTER: Sylvia Casas de Leon, MD
DISCUSSANT/MODERATOR: Danielle Trief, MD, MSc
2:46–3:10 p.m.  Break

3:10–4:30 p.m.  SESSION IV: Ethical Considerations and Applied Genetics
MODERATED BY Irene H. Maumenee, MD
3:10–3:30 p.m.  The Donald and Barbara Jonas Lecture
The Paradox of Genomic Testing: Increasing Uncertainty in Interpreting and Applying the Results
Reed E. Pyeritz, MD, PhD, University of Pennsylvania, Philadelphia, PA
3:30–4:00 p.m.  CASE PRESENTATIONS
3:30–3:40 p.m.  Case 10: Congenital Cataract
CLINICAL PRESENTER: Carolina Adams, MD
DISCUSSANT/MODERATOR: Steven A. Kane, MD, PhD
3:40–3:50 p.m.  Case 11: Retinitis Pigmentosa
CLINICAL PRESENTER: Jose R. Lima de Carvalho Junior, MD
DISCUSSANT/MODERATOR: Stephen H. Tsang, MD, PhD
3:50–4:00 p.m.  Case 12: Retinoblastoma
CLINICAL PRESENTER: Vlad Diaconita, MD
DISCUSSANT/MODERATOR: Brian Marr, MD

4:00–4:20 p.m.  PANEL DISCUSSION
Panel Members: Lorraine Frazier, PhD, RN, FAAN,
Simon John, PhD, Jeffrey Liebmann, MD, Reed E. Pyeritz, MD, PhD,
Leejee H. Suh, MD
4:20–4:30 p.m.  Closing Remarks
G. A. (Jack) Cioffi, MD
4:30 p.m.  Reception
CME: FACULTY DISCLOSURES

DISCLOSURES

Before the program, all faculty will disclose the existence of any financial interest and/or other relationship(s) (e.g. employee, consultant, speaker’s bureau, grant recipient, research support, stock ownership or any other special relationship) they might have with a) the manufacturer(s) of any commercial product(s) to be discussed during their presentation and/or b) any commercial contributor to this activity. When unlabeled uses are discussed, these will also be indicated.

Vimla S. Aggarwal, MBBS, FACMG
None

Aliaa H. Abdelhakim, MD, PhD
None

Carolina Adams, MD
None

Rando Allikmets, PhD
None

Mark Breazzano, MD
None

Steven E. Brooks, MD*
None

Sylvia Casas de Leon, MD
None

Stanley Chang, MD
Consulting Fees
Genentech, Inc.
Travel Reimbursement
Alcon Vision, LLC

G. A. (Jack) Cioffi, MD
None

Salman Dar, MD
None

C. Gustavo De Moraes, MD, PhD, MPH
None

Vlad Diaconita, MD
None

Lorraine Frazier, PhD, RN, FAAN
None

William W. Hauswirth, PhD
Consulting Fees & Ownership Interest
Applied Genetic Technologies Corporation
Contracted Research
Applied Genetic Technologies Corporation, IVERIC Bio, Inc., Decibel Therapeutics, Inc. & Adverum Biotechnologies

Simon John, PhD
None

Steven A. Kane, MD, PhD
None

Sabine Khan, MD
None

Jeffrey Liebmann, MD
None

Jose R. Lima de Carvalho Junior, MD
None

Michelle Maeng, MD
None

Brian Marr, MD
Consulting Fees
Aura Biosciences, Inc., Castle Biosciences, Inc. & Immunocore, Ltd.

Irene H. Maumenee, MD
None

Reed Pyeritz, MD, PhD
Royalty
McGraw-Hill Education & Elsevier B.V.

Portia E. Sirinek, MD
None

Megan Soucy, MS, CGC
None

Janet Sparrow, PhD
None

Leejee H. Suh, MD
None

Joy Tanaka, PhD
None

Tongalp Tezel, MD
None

Danielle Trief, MD, MSc
None

Stephen H. Tsang, MD, PhD
None

Marius Üffing, PhD
Consulting Fees
F. Hoffmann-La Roche, Ltd.

Marlene D. Wang, MD
None

Lawrence A. Yannuzzi, MD
None

Xin Zhang, PhD
None

*Indicates that the speaker intends to discuss unlabeled uses of a commercial product, or an investigational use of a product not yet approved for this purpose. The speaker will disclose this information during his/her presentation.
LECTURESHIPS

The Arthur Gerard DeVoe, MD Lectureship at Columbia University was established in 1999 to honor Arthur Gerard DeVoe, MD for his extraordinary contributions to ophthalmology and vision science and to recognize his leadership of the Department of Ophthalmology at the Edward S. Harkness Eye Institute. Supported through the generosity of Joan Gilson, a former patient of Dr. DeVoe, the lectureship attracts international leaders in ophthalmology who present recent and important findings in their field.

The John H. Dunnington, MD Memorial Fund was established by Dr. Dunnington's nephew, Walter Dunnington, in 1984. The Lectureship was introduced by the Executive Committee of the Department of Ophthalmology to perpetuate this outstanding physician's contributions in the field of ophthalmology and to the development of the Edward S. Harkness Eye Institute.

The Donald and Barbara Jonas Lecture is supported through the Jonas Children’s Vision Care Initiative, a first-of-its-kind integrated effort to prevent and treat blindness and other serious eye disorders in children that draws upon Columbia Ophthalmology's many strengths, including clinical care from infancy throughout life, genetic testing of individuals with eye disease for diagnosis and treatment, the resources of a wide array of laboratories, and the University’s many initiatives in an interdisciplinary field called translational research.

The Florence Epstein Teicher Lectureship was established in 2005 by Mr. Milton S. Teicher in memory of his wife, Mrs. Florence Epstein Teicher. She looked upon life as a learning process and devoted her philanthropic energies to cultural and humanitarian causes. Mr. Teicher created this lectureship with the goal of disseminating new knowledge to physicians who are treating patients with blinding diseases such as open angle glaucoma, a condition from which Mrs. Teicher suffered.
Marius Üffing, PhD, earned his PhD in Biology from the Albert-Ludwig-University Freiburg in Germany. He is the Director of the Institute for Ophthalmic Research and Co-Chair of the Centre for Ophthalmology at the University Medical Center, in Tübingen, one of the largest centers for ophthalmology in Europe. Dr. Üffing directs the Medical Proteome Center, a core facility of the medical faculty in Tübingen, and acts as a co-director of QBiC, the Quantitative Biology Center of the University of Tübingen. Dr. Üffing has a long-standing history in medical research as well as a background in protein science and molecular genetics. In 1988, he was appointed Research Associate at Columbia University, Vagelos College of Physicians and Surgeons, following that role, he worked as a group leader in pharmaceutical industry at Goedecke-Parke-Davis and at the University Medical Centre Munich for fifteen years. In 2008, Dr. Üffing became the Director of the Independent Research Unit/Division of Protein Science at the National Research Centre for Environment and Health in Munich.

Between 2015 and 2019 Dr. Üffing has coordinated the EU Horizon2020 Systems Medicine Project EYE-RISK (www.eyerisk.eu) focused on AMD. He coordinates the eye-examination module of the German National Cohort, a population-based observational study including 200,000 individuals (www.nationale-kohorte.de).

Marius Üffing, PhD has published more than 240 original articles.

William W. Hauswirth, PhD, is the Rybaczki-Bukkard Professor of Ophthalmology at the University of Florida. Dr. Hauswirth received his BS in Chemistry from Stanford University, and his PhD in Chemistry-Biochemistry from the Oregon State University. His work focuses on a long-term research interest in using viral vectors for the delivery and testing of potentially therapeutic genes for many different forms of inherited retinal disease in animal models, including Achromatopsia, X-linked and autosomal dominant RP, Leber Hereditary Optic Neuropathy, Best Macular Dystrophy, Usher 1B, LCA, Stargardt and MERTK disease. Nine are in early phase human clinical trials or on clinical trial trajectories. Recent honors include speaking in 2018 at the Vatican upon a Papal invitation and sharing the Champalimaud Vision Research award in 2019. He has received visiting professorships from the universities of Oxford and Edinburgh, UK, and Pavia, Italy, as well as the Pasteur Institute, Paris, France. Overall, Dr. Hauswirth has published more than 400 research papers. His current research is supported by two NIH R24 grants and an NIH R01 grant, both as a co-PI, and by five biotech company collaborative grants, all as the Principal Investigator.
Reed E. Pyeritz, MD, PhD, a medical geneticist, focuses his research in three areas: Mendelian disorders of the cardiovascular system; the concept of uncertainty in clinical implications of medical genetics; and ethical, legal, and social implications of human genetics. He is continuing his studies, begun over 30 years ago, of Marfan syndrome and related conditions, diseases in which the aorta, and occasionally major arterial branches, gradually enlarge and dissect, leading to early demise if untreated. Current efforts address the identification of additional genes that predispose to arteriopathy, and improving methods for diagnosing and treating arterio-venous malformations, especially in hereditary hemorrhagic telangiectasia (HHT).

Simon John, PhD, is Professor of Ophthalmic Sciences at Columbia University Irving Medical Center. His lab works to elucidate the molecular basis of glaucoma with the aim of developing new therapeutic strategies. They apply a multi-disciplinary approach combining molecular, genetic, and physiologic methods to identify genes and molecular mechanisms that underlie this neurodegenerative disease. In collaboration with engineers, they are also developing new mini-electronic devices to monitor physiologic pressure and to monitor and manipulate neural activity with the goals of transforming research capabilities and improving patient care.
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