

Genetic testing interpretation from basic to advanced

Course organizers

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Course description

Interpreting genetic testing results is a complex skill that requires an understanding of basic genetic principles, molecular techniques in genetic testing, and clinical implications of genes, variants, and phenotypes. Rapid advancements in genetics and genomics have created expertise gaps among healthcare providers and researchers for interpreting genetic testing for clinical purpose-however, understanding the distinct types of genetic testing and how to discern between pathogenic and benign variation variants is key in disease diagnosis, treatment, and prevention. Attend this course if you are struggling with the interpretation of genetic testing results. Learn from experts in the field who will cover genetic testing and genetic counseling issues, as well as clinical information about syndromic and non-syndromic inherited ocular diseases. Attendees will walk away with practical tips for the genetic diagnosis of inherited ocular diseases, and a comprehension of aspects ranging from ophthalmological evaluation to variant curation, including genetic counseling, research consortia, and public databases.

Learning objectives

Attendees will leave this session with the ability to:

- Recognize the most common syndromic and non-syndromic inherited ocular diseases.
- Describe the molecular basis of genetic testing.
- Interpret positive, inconclusive and negative genetic testing results.
- Identify challenges health care providers face in the ophthalmic genetics field.
- Discuss new concepts to utilize public databases for gene and variant curation.

Physician accreditation statement

The Association for Research in Vision and Ophthalmology (ARVO) is accredited by the Accreditation Council for Continuing Medical Education (ACCME) to provide continuing medical education for physicians.

Physician credit designation

ARVO designates this live activity for a maximum of 3.75 *AMA PRA Category 1 Credits™*. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

International Attendees: The American Medical Association (AMA) has determined that physicians not licensed in the U.S. who participate in CME activities are eligible for *AMA PRA Category 1 Credits™*.

Statement of Need

Interpreting genetic testing results is a complex skill that requires an understanding of basic genetic principles, molecular techniques in genetic testing, and clinical implications of genes, variants, and phenotypes. Even after taking educational courses, clinicians of all specialties struggle with interpreting genetic testing results (Emily Edelman, National Society of Genetic Counselors 2019 annual meeting).

Target Audience

The target audience includes ophthalmologists, genetic counselors, geneticists, researchers, and trainees (residents, fellows, postdocs) seeking to interpret and/or discuss the results of genetic testing for eye and vision related conditions.

European Union of Medical Specialists (UEMS) CME/CPD requirements

European physicians can convert CME credit from the course to meet European Union of Medical Specialists (UEMS) CME/CPD requirements.

The American Medical Association has an agreement of mutual recognition of CME credits with the UEMS, the accreditation body for European countries. Physicians interested in converting *AMA PRA Category 1 Credit™* to UEMS-European Accreditation Council for Continuing Medical Education CME credits (ECMECs) should contact the UEMS at mutualrecognition@uems.eu.

Instructions to receive credit and/or certificates

Learners interested in claiming *AMA PRA Category 1 Credits™* or obtaining a certificate of participation for participating in this course must pay the CME credits and Certificates fee as part of their Annual Meeting Registration. Payment is collected during the Annual Meeting registration process by adding “CME credits and Certificates” as a session. Learners who do not register for CME before the conclusion of the meeting will not be able to claim and request credit. Learners registered as guests are not eligible to receive CME credit.

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CME credits and certificate fee	By March 8	After April 26
ARVO MIT/Nonmember-in-Training	\$ 79	\$ 79
ARVO Member/Exhibitor (paid full-meeting access badge)	\$ 99	\$ 119
Nonmember/Exhibitor (complimentary full meeting access badge only)	\$ 99	\$ 119

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*Some accrediting agencies may award equivalent Continuing Education (CE) credit when presented with a certificate of participation from an activity that offered *AMA PRA Category 1 Credit™*. Check with your accrediting agency for information.

Certificate of attendance

Certificates of attendance are available free to all attendees. These certificates state that one has attended the course but does not offer credit tracking. These certificates are available for self-service download from the Annual Meeting website.

Contact Us

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The following activity managers reported no relevant financial relationships.

Robert Hufnagel, MD, PhD, (Course organizer), Cathy Conley (Activity Manager) Melissa Marick, MA (Activity Manager)

The following clinical content reviewers reported no relevant financial relationships.

Kimberly Zanon, BSN, RN *The following course organizers, activity planners and managers reported relevant financial relationships.*

Full name	Role	Disclosure
Kristy Lee, MS, CGC	Course organizer	Janssen Pharmaceuticals (F)
Ramiro Maldonado, MD	Course organizer	ProQR Therapeutics (C); PYC therapeutics (C)

Priya Chaudhary, PhD	PDEC	Alcon Research Institute (F)
Azza El-Remessy, PhD, PharmD, BC-ADM	PDEC	Nour Therapeutics, LLC (O)
Edmund Tsui, MD	PDEC	Kowa Company Ltd (C, F); Cylite Pty Ltd (F); EyePoint Pharmaceuticals (C)
Thomas Ach, MD, FEBO	CME	Apellis Pharmaceutical (R); Roche (R); Novartis (R, F); Bayer (R); Johnson & Johnson (R); Macregen (I) (ended Feb. 2022)
Ron Adelman, MD, MPH, MBA	CME	Identifeye (C); Johnson & Johnson/Jenssen: (C)
Parisa Emami, MD, MPH	CME	Eyepoint Pharmaceuticals (C); Bausch and Lomb: (C)
Sally Ong, MD	CME	Eyepoint Pharmaceuticals (C); Patent Cooperation Treaty (PCT) Application No. PCT/US2023/024209 - P
Laura Lanford	Activity manager	Johnson and Johnson (I); Pfizer (I)

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