

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.

Presentations

Presenters and presentations may change.

Time	Topic	Speaker
1 PM	Phenotyping inherited ocular diseases	Ramiro Maldonado, MD. Duke University

Time	Topic	Speaker
	<p>Inherited ocular diseases are a leading cause of blindness. The diagnostic landscape for these conditions has undergone a remarkable transformation, propelled by technological advancements and scientific breakthroughs. In the contemporary era, we enjoy the capabilities of cutting-edge multimodal imaging, intricate electrophysiology techniques, and precise genetic testing methodologies. These advancements collectively empower clinicians and researchers to unravel the diagnosis of these challenging conditions. As we embark on this first presentation, our focus will be dedicated to showing the audience on the latest diagnostic tools seamlessly integrated into the modern inherited retinal disease clinic, offering a glimpse into the forefront of ocular healthcare innovation.</p>	
1:30 PM	Gene 101. Basics of molecular diagnostics	Robert B. Hufnagel, MD, PhD, National Eye Institute
	<p>Genetics of rare eye diseases, clinical molecular diagnostics, bioinformatics tools, and variant classification will be discussed.</p>	
2 PM	Genetic counseling in Ophthalmology	Kari E. Branham, MS, CGC University of Michigan
	<p>Genetic counseling is an essential aspect of the clinical care and management of patients affected with inherited eye diseases. During this portion of the education course, we will review essential elements of a genetic counseling session and discuss the value genetic counseling and genetic testing can provide for both patients and their health care providers. Moreover, we will discuss different models for the integration of genetic counselors into the care of patients with inherited eye disease. Case examples will be presented to illustrate these points.</p>	
2:30 PM	Interpreting genetic test results for ocular disorders: The basics of molecular diagnosis through application of results	Kristy Lee, MS, CGC. University of North Carolina (UNC)
	<p>Genetic testing options for ocular disorders are expanding and becoming more widely used in clinical care. However, genetic testing results are not always easy to interpret or informative. This presentation will provide an overview on how gene variants are evaluated for pathogenicity, as well as highlight some of the challenges and pitfalls with interpreting genetic testing results and offer guidance on sharing results with patients.</p>	
3 PM	Break	
3:15 PM	ClinGen gene: Disease and variant curation	Gavin Arno, PhD, BSc, UCL Institute of Ophthalmology
	<p>The genomic revolution and widespread availability of next generation sequencing for rare diseases has driven genetic discovery for more than a decade and now dominates molecular diagnostic practice. With these advances in our ability to detect rare variants across the entire genome comes increased uncertainty of the impact of many variants and genotypes. Standardization of variant interpretation</p>	

Time	Topic	Speaker
	<p>(ACMG/AMP guidelines) often leads to uncertainty (variants of uncertain significance) and the importance for many genes in disease is unclear. The overall aim of the ClinGen Gene Curation Expert Panels (GCEP) and the Variant Curation Expert Panels (VCEP) is to provide gene and variant level expert guidance for interpretation of genetic results for clinicians, clinical scientists, molecular geneticists and counsellors. Here, we will explore the process of gene and variant curation with case studies to highlight some of the challenges.</p>	
3:45 PM	Public databases and Sponsored testing programs	Emily Place, MS, LCGC MEEI-Harvard
	<p>Sharing of genetic data is critical to further our understanding of mendelian disease and improve interpretation of genetic results. This talk will discuss data sharing in the different yet equally important areas of sponsored genetic testing programs and public variant databases. We will discuss aspects of each area, highlight benefits and limitation of each and discuss useful ways to share data while protecting the identity of patients.</p>	
4:15 PM	Expecting the unexpected: incidental and secondary findings in genetic testing	Linda Reis, MS, CGC Medical College of Wisconsin
	<p>The use of next generation sequencing, through panel, exome, and/or genome testing is often the most efficient method to identify causative variants in genetically heterogeneous ocular disorders. However, broad testing for ocular conditions can reveal unexpected results. Conversely, exome sequencing for other indications may identify an incidental diagnosis of an unrecognized ocular condition. This presentation will review the range of incidental and secondary findings or unexpected diagnoses that may be returned from clinical testing.</p>	
4:45 PM	Panel: Open Q&A with presenters	
5 PM	Adjourn	