

Inherited Retinal Diseases and Electrophysiology Testing

Learning Objectives:

1. Know when to order an ERG for a suspected inherited retinal disease (IRD) patient and feel comfortable interpreting the results.
2. Articulate the major clinical characteristics of common IRDs such as RP, Stargardt, choroideremia, Usher syndrome, etc.
3. Describe how you would explain genetic risk to an IRD patient or their parent for the various inheritance patterns of IRDs (dominant, recessive, X-linked, mitochondrial, sporadic, etc.).
4. Summarize the current status of gene therapy trials for IRDs in the US.

Pre-Work:

1. Watch
 - Watch Don Creel's ERG video: <https://morancore.utah.edu/section-12-retina-and-vitreous/electroretinograms/>
2. Read
 - Chapters 13 and 14 in the Retina BCSC book
 - Eeview article on gene therapy for IRDs: <https://iovs.arvojournals.org/article.aspx?articleid=2791262>
 - [AAO PowerPoint reviews](#): R38-39, R3, R5, R9-11, R42, R70

Lecture outline:

7:00-7:15 am

Practical introduction to OCT and ERG in IRDs.

7:15-8:00 am

Case-based lectures for the following BCSC Retina chapters

13: Hereditary Retinal & Choroidal Dystrophies

14: Retinal Degeneration Associated with Systemic Disease

8:00-8:30 am

Retinal gene therapy in IRDs

8:30-9:00 am

Retina Bowl