Management of Inherited Ocular Disorders Emerging Concepts in the Post-Genome Era

Course Description:

This one hour course will discuss cutting edge treatments for inherited ocular disorders in a case based approach. The lecture will include a discussion of the NIH EyeGene project’s results and future steps. It will also explore stem cell treatment trials. Finally, the course will discuss the next generation of low vision technology including the development of retinal implants and head-mounted display systems. The discussion will be augmented by case presentations of patients who have undergone the above treatments.

Learning Objectives

1. To familiarize optometrists with the current research on treatment of inherited ocular disease
2. To become comfortable determining which patients are appropriate candidates for implantable technology/ “bionic vision”
3. To explore the role of genetic testing
4. To develop the knowledge base to appropriately counsel patients and make appropriate referrals

Outline

1. Overview of genetic ocular disorders
   a. In-depth review of conditions including
      i. What are the clinical characteristics of these patients?
         1. Aniridia-- congenital condition
            a. Glaucoma in later childhood/teenage years
            b. Nystagmus
            c. Foveal Hypoplasia
            d. Early onset cataracts
         2. Stickler’s Syndrome
            a. Ocular, facial, auditory and skeletal/joint problems
            b. High myopia
            c. Possible glaucoma, retinal detachment and earlier cataract
         3. Oculo-cutaneous Albinism and Ocular Albinism
            a. Nystagmus
            b. Reduced acuity
            c. Photophobia
         4. Stargardt Maculopathy
            a. Reduced central acuity
            b. Possible reduction in rod function/difficulty with night vision
         5. Retinitis Pigmentosa
            a. Reduced night vision, evident as early as childhood
b. Progressive loss of peripheral vision (beginning mid-peripheral)
c. Can be syndromal (Bardet-Biedel, Usher, etc)
d. CME/early onset cataracts

6. Dominant Optic Atrophy
   a. Nerve pallor
   b. Reduced acuity
   c. Reduced color vision
   d. Reduced visual field

7. Leber’s Congenital Amaurosis
   a. 13 different types have been identified
   b. Variable vision findings depending on type
   c. High hyperopia, keratoconus (2’ to oculodigital syndrome is noted)

8. Leber’s Optic Atrophy/Hereditary Optic Neuropathy
   a. Presents in teens to 20’s
   b. Blurred vision in one eye with the other following shortly after
   c. Primarily affects central vision and color vision

9. Achromatopsia
   a. Can be complete or incomplete
   b. Loss of color vision in varying degrees (depending on whether condition is complete or incomplete)
   c. May have associated nystagmus, high refractive error (hyperopia > myopia)

b. Discussion of objective findings/clinical exam findings
   c. Discussion of common subjective complaints
   d. Mode/pattern of inheritance
      i. Dominant Inheritance
         1. Aniridia (though ⅓ are a new mutation)
         2. Stickler Types I-III
         3. Stargardt with ELOVL4 mutation
         4. RP
         5. Dominant Optic Atrophy
         6. LCA with CRX or IMPDH1 mutations (rare)
      ii. Recessive
         1. Stickler Types IV-VI
         2. Oculocutaneous Albinism
         3. Stargardt with ABCA4 mutation
         4. RP
         5. LCA (more common)
         6. Achromatopsia
      iii. X-Linked
         1. Ocular Albinism
2. RP
   iv. Mitochondrial
      1. LHON
   e. Low vision rehabilitation goals
2. Care and counseling the patient at time of diagnosis
   a. Counseling on inheritance
   b. Counseling on diagnosis/ prognosis
   c. When to consider formal genetic testing
      i. For your adult patients
      ii. For your pediatric patients
   d. Ethical considerations in genetic testing
      i. Review of literature
3. Review of results and outcomes of the NIH EyeGene project
   a. Current published literature
   b. Future directions for the project
4. Low vision rehabilitation management considerations:
   a. The early years
      i. Role of early intervention programs-- who qualifies and what services are provided
      ii. Role of the low vision optometrists and therapists in the habituation plan
      iii. Preparing for preschool
   b. School years
      i. Providing appropriate accommodations for the classroom and to optimize learning environments
      ii. When to introduce low vision aids
      iii. Review of the IDEA act
   c. Challenges in the teenage years
      i. Importance of peer groups
      ii. Preparing your patient for the college workplace transition
   d. Preparing/entering for the workforce
5. Treatments for the next generation
   a. How to counsel your patient regarding current clinical trials
   b. When should you refer your patient to a clinical trials
   c. Review of publications and ongoing clinical trials in
      i. stem cell treatment
      ii. gene therapy
   d. When is technology an outlet?
      i. Technology in the eye
         1. Retinal implants
      ii. Technology out of the eye
         1. Head mounted display systems
         2. Computer platforms for the workplace
         3. Smart-phone/tablet based systems
6. Case based discussions:
a. Ocular Albinism
b. Oculo-cutaneous albinism
c. Dominant Optic Atrophy
d. Incomplete Achromatopsia
e. Stargardt's macular dystrophy