Clinical presentation of Stargardt Disease and Low Vision Management

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Abstract: We discuss the clinical findings of Stargardt disease. Relevant ophthalmic literature is reviewed including interim findings of the Natural History of the Progression of Atrophy Secondary to Stargardt Disease (ProgStar) Reports.

I. Case History
- Patient demographics
  - O.D., 42 year old Caucasian female
- Chief complaint
  - Reported vision has slightly worsened since last low vision examination in September 2014
  - Felt her vision prevented her from having the visual speed and efficiency needed to keep up in fast-pace work environment (previously worked in medical coding and billing)
  - Interested in returning to school to explore other employment options (specifically a career in teaching)
- Ocular, medical history
  - Stargardt disease
    - Diagnosed at 14 years of age
  - History of gestational diabetes
- Medications
  - None reported
- Other salient information
  - Currently uses Amigo Portable CCTV and non-illuminated hand-held magnifier for reading and changes resolution on computer/TV to improve contrast

II. Pertinent findings
- Clinical
  - Entering (unaided) Visual Acuity, measured with back-lit Bailey-Lovie Chart
    - 20/320 OD/OS
  - Retinoscopy
    - -3.50 DS OD, -2.50 -1.50 x045 OS
  - Trial Frame Refraction
    - -2.50 -1.00 x 045 OD, -2.50-1.00 x 045 OS
  - Best Corrected Visual Acuity, measured with back-lit Bailey-Lovie Chart
    - 20/200 OD, 20/160 OS
  - Contact Lens Prescription Released
    - -3.00 DS OD/OS
    - Alcon Fresh Look Clear Blend 8.6/14.5
  - Contrast Sensitivity (Mars Chart)
    - 1.6 log, Mildly reduced, 2.5% Weber
  - Low Luminance Contrast Sensitivity (using 4% gray transmission NoIR U23 fitovers)
    - 1.16 log, Moderately reduced, 6.9% Weber
  - Confrontational Vision Fields (with transilluminator)
- Full in all meridians OD/OS
  - Berkeley Central Visual Field Test (tangent screen test)
  - Bilateral superior field scotoma

### III. Differential diagnosis
- Primary/Leading
  - Stargardt disease
- Others
  - Fundus albipunctatus
  - Retinitis punctate albscens
  - Cone or cone-rod dystrophy
  - Batten disease and Spielmeyer-Vogt syndrome

### IV. Diagnosis and discussion
- Stargardt disease
  - Even though Stargardt disease is the most common hereditary macular dystrophy, there is limited information about the natural course of the disease and associated risk factors for progression. Recently, the results of the ProgStar studies were published which aimed to establish the natural disease progression using both structural and functional measures and whether yearly change in visual acuity was a sensitive enough outcome measure for change.
- Unique Features
  - Atrophic-appearing lesions within macula
  - Presence of yellow-white round or pisciform lesions at RPE level (referred to as “flecks”) in an annulus pattern encircling macula

### V. Treatment, management
- Treatment and response to treatment
  - No effective treatment. Cigarette smoking, second-hand smoke and vitamin A supplementation should be avoided.
  - Released updated spectacle prescription and contact lens prescription for general wear.
  - Prescribed low vision aids:
    - Eschenbach 4x12 Microlux monocular telescope for distance spotting
    - Beechers 4x20 wide angle binocular mounted telescope glasses for distance viewing
    - Mattingly 4x/12D LED-illuminated hand-held magnifier for near spotting
    - Mattingly Stella desk Lamp for task lighting

- Bibliography

### VI. Conclusion
- Clinical pearls, take away points
  - The ProgStar study interim findings indicated that visual acuity may be an inadequate outcome measure to determine progression in Stargardt patients. More sensitive outcome measures that are readily available to eye-care providers would include SD OCT, FAF and microperimetry. An exception to this would be younger patients with Stargardt disease or those with minimal visual impairment at baseline, where visual acuity is an adequate outcome to detect any progression.
  - The importance of educating patients on the possible genetic component of Stargardt
disease.

- Recall the functional implications Stargardt disease can have on a patient’s daily activities and the importance of low vision rehabilitation as they transition into different stages of their lives.