Abstract:
A discussion of the serious ocular manifestations, decreased visual functions and visual prognosis for patients with Bardet-Biedl syndrome by reviewing the case of a 14-year-old Hispanic female diagnosed with this rare condition.

I. Case history
   a. Patient demographics
      i. 14-year-old Hispanic female
   b. Chief complaint:
      i. Decline in night vision
      ii. Increasing frequency of outward eye turn
      iii. Parents feel patient’s vision is getting worse overall
   c. Ocular history:
      i. Exotropia first noted around age 10
      ii. First pair of glasses also prescribed around age 10
      iii. Parents report difficulty with night vision since early childhood
   d. Medical history:
      i. Severe fever and subsequent diagnosis of kidney disease at 1-month-old, which led to extensive work-up including genetic testing
      ii. Genetic testing revealed BBS9 homozygous mutation
      iii. Recent history of recurrent urinary tract infections
      iv. Monitored regularly by nephrologists
      v. History of post-axial polydactyly; extra fingers surgically removed in infancy
      vi. Presents with mild truncal obesity
      vii. Family history of consanguinity; parents distantly related
      viii. No current medications and no known drug allergies
   e. Developmental history:
      i. Freshman in a public high school; enrolled in a special day class
      ii. Developmentally delayed

II. Pertinent findings
   a. Refractive status:
      i. Cycloplegic refraction shows low myopia and astigmatism similar to entering glasses
         OD: -1.25 -1.75 x 180  OS: -1.25 -1.50 x 180
   b. Binocular status:
      i. Full versions and ductions
      ii. Anomalous head posture: right head turn
      iii. Constant alternating exotropia, convergence insufficiency type
         1. Variable: 40-60 pd at near, 10-40 pd at distance
         2. Alternating but 80-90% right exotropia
      iv. Steady central fixation OD/OS although fixates primarily OS
      v. Patient frequently squints or closes right eye
      vi. Unable to appreciate 480 sec of stereopsis with the PASS test
   c. Visual function:
      i. Decreased best corrected visual acuity with crowding effect
         1. 20/32 OU using isolated Lea symbols
         2. 20/40 OU using Lea symbols with crowding bars at 100% spacing
         3. 20/60 OU using Lea symbols with crowding bars at 50% spacing
         4. 20/32 OU (detection) and 20/50 OU (identification) using Cardiff cards
ii. Decreased contrast sensitivity, worse in dark
   1. Mild-moderately decreased CS in light conditions: 1.6% using Cambridge Contrast Sensitivity Test, 3.2% using Mr. Happy Contrast test
   2. Severely decreased CS in dark conditions (simulated using NoIR u23 4% gray tint): 8% using Cambridge Contrast Sensitivity Test after adaptation period

iii. Normal color vision
   1. Passes Mr. Color, F2 Squares, and Portnoy saturated & desaturated plates

iv. Confrontation visual fields constricted in light and more constricted in dark
   1. Moderate constriction in light; greater inferiorly and to the left
   2. Severe constriction in dark; diode wade noted only in central 30-40 degrees

d. Anterior segment:
   i. Unremarkable findings

e. Posterior segment:
   i. Bilateral disruption of macular pigment and yellow macular deposits OD>OS
   ii. Mild retinal blood vessel attenuation OU
   iii. Mild temporal pallor of the optic discs OD>OS
   iv. “Patchy” mid-peripheral RPE disruption OU; absence of bony spicules OU
   v. Macular cube OCT showed:
      1. Severe retinal thinning particularly in the outer retina OU
      2. Absence of EPIS and COST lines throughout macular region OU
      3. Mild epiretinal membrane OU

III. Differential diagnoses
   a. Leading diagnosis: retinopathy associated with Bardet-Biedl syndrome
   b. Other differential diagnoses:
      i. Juvenile macular degenerations, e.g. Best disease, Stargardt disease
      ii. Retinitis pigmentosa
      iii. Retinopathy associated with Laurence-Moon syndrome or Alström syndrome

IV. Diagnosis and discussion
   a. Bardet-Biedl syndrome (BBS)
      i. A rare, autosomal recessive disorder
      ii. Prevalence of 1:100,000 in North America
      iii. Affects males and females equally
      iv. Primarily characterized by retinal dystrophy, polydactyly, truncal obesity, hypogenitalism and genitourinary abnormalities, kidney problems, and developmental delay
      v. A pleiotropic condition with highly variable expressivity and severity of clinical manifestations
      vi. Diagnosis typically made based on clinical findings alone
      vii. Mutation of BBS genes affects immotile cilia, which are located throughout the body and in retinal photoreceptors
   b. Ocular manifestations of Bardet-Biedl syndrome
      i. Retinal dystrophy is the clinical feature with greatest penetrance; >90% of patients show signs of retinal dystrophy
      ii. Decreased vision and subtle maculopathy is often primary reason for additional investigation that leads to diagnosis
      iii. Less common ocular findings include: strabismus, nystagmus, posterior subcapsular cataracts
      iv. Visual symptoms include:
         1. Nightblindness and elevated dark-adapted thresholds
         2. Decreased visual acuity
         3. Visual field loss
      v. Typical fundus appearance includes:
1. Some degree of macular involvement (i.e. pigmentary changes, vitelliform-like lesions, bulls-eye maculopathy)
2. Less likely to see bony spicules
3. Attenuation of retinal vessels, particularly arteries

vi. OCT scans typically show:
   1. Severe thinning of overall retinal thickness
   2. Disruption of photoreceptor integrity; loss of EPIS/COST lines
   3. Thinning of RPE
   4. Loss of retinal lamination at severe stages
   5. Deposits anterior/adjacent to Bruch’s membrane
   6. Epiretinal membrane formation

vii. All patients tested have shown abnormalities on ERG
   1. Typically rod-cone dystrophy, however, cone-rod also documented
   2. Not uncommon for patients to have non-detectable ERG signal

viii. Generally poor although variable visual prognosis
   1. Most patients are legally blind by the second or third decade
   2. Visual field, visual acuity, fundus appearance and OCT findings minimally correlated to age

V. Treatment and management
   a. Extensive discussion with parents regarding patient’s functional vision and visual strengths and weaknesses
      i. Continue with full-time spectacle correction
      ii. Variable, alternating exotropia with absent stereopsis
      iii. Relatively good visual acuity with isolated symbols (20/30), reduced to 20/60 with 50% spacing. Consider increased spacing in educational materials.
      iv. Visual field extent and contrast sensitivity dependent on light levels
      v. Unknown rate of progression for visual field reduction and central vision loss

b. Wrote report summarizing exam findings for parents to use to advocate for vision services in public school including:
   i. Services from a teacher for children with visual impairment (TVI)
      1. Modification of educational materials to increase visibility
      2. Desk placement in classroom to avoid fluctuation in light levels and glare sources
   ii. Referral for orientation & mobility training
      1. Consider introducing white cane due to progressive nature of vision loss
      2. Absent stereopsis but availability of contrast and color as depth clues

   c. Emphasized importance of regular eye exams

VI. Conclusion
   a. Patients with Bardet-Biedl syndrome have a poor visual prognosis with an uncertain time course for progression; efforts should be made to maximize remaining vision and to connect patients with low vision services
   b. Syndromic diagnoses should be considered when suspected retinal dystrophies present with other significant systemic findings/history
   c. Optometrists can play an important role in helping patients, parents/guardians, and educators to understand functional vision in visually impaired and developmentally delayed patients
References


