Grand Rounds:
Interesting Presentations of Young(er) Patients
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Why Young(er) Patients?
- Young patients are commonly seen in most practices
- Conditions in the geriatric population may also be present in younger populations
  - BUT, for different reasons
- Becoming a more well-rounded practitioner

Case #1
- 27 yo Caucasian female
- CC: Possible visual field defect detected on confrontation testing
  - Unaware of defects in daily life
  - Denied neurological symptoms
- Ocular History: Unremarkable
- ROS: Unremarkable
- Medication: None
- Family Ocular History: Glaucoma (maternal aunt)

Case #1
- Uncorrected distance visual acuity
  - OD 20/15 OS 20/15
- PERRL (-)APD
- Motilities full and comitant without pain/diplopia
- Confrontation fields
  - Superior nasal constriction OD and OS
- Refraction: +1.00 DS OU
- Anterior Segment: Unremarkable
- GAT (@2:32pm): 24mmHg OD/OS
- Posterior Segment:

Follow up 1 week later
- No changes in vision, no new visual/ocular/neurological complaints
- Uncorrected distance acuity
  - 20/15 OD/OS
- Motilities full and comitant
- PERRL (-)APD
- Ishihara Color Testing
  - 10/10 plates correct OD/OS
- GAT @ 1:29pm: 22/23 mmHg
• Pachymetry: 586/581 microns
• Gonioscopy: flat approach, open to CB 360 OU, tr pigment

http://www.westcoastretina.com/WestCoastRetina/Nov-2010.html

Follow up over ~ 3 years
• No new ocular/visual/neurological complaints
• No treatment has been elected
• Uncorrected visual acuity
  • OD 20/15  OS 20/15
  • Tmax 25/25, range: 22-25

Optic Nerve Head Drusen
• Congenital, acellular, calcific bodies commonly often found bilaterally and associated with small, crowded optic nerves
• It is believed that the formation of ONHD is associated with axonal transport alteration and degeneration.
• The prevalence of ONHD in the general population has been estimated at approximately 0.4%; however, histological studies have reported prevalence as high as 2.4%

Optic Nerve Head Drusen
• In young patients, drusen typically “buried”
• With age, “buried” drusen enlarge due to calcium apposition and extend anteriorly
• As the drusen expand, the nerve fiber layer is disrupted, resulting in visual field loss in 24-87% of adults (Auw-Haedrich et al)
• Central visual acuity is generally spared, and thus this condition is often asymptomatic

Pseudopapilledema
• Critical to differentiate pseudopapilledema from drusen from true optic nerve edema
• Blurring of disc margins
• Obscuration of retinal vessels
• Peripapillary hemorrhages
• Retinal/choroidal folds
• B-scan continues to be the gold standard for detecting drusen
• OCT (and AF) gaining popularity

21 **Optic Nerve Edema**

or

22 **Optic Nerve Drusen?**

23

24

25 **Lee et al**

26

27 **Treatment**

• No universally accepted treatment protocol
• Contrasting views on whether nerve fiber layer loss occurs more rapidly in presence of ocular hypertension (Moussalli et al, Grippo et al)
• Some evidence that treatment with topical alpha-2 adrenergic agonist brimonidine may provide neuroprotection in addition to it’s anti-hypertensive effect

28 **Brimonidine**

• Believed to upregulate intrinsic cell survival signaling pathways, as well as antiapoptotic genes.
• In rat models, has been shown to reduce levels of intravitreal glutamate associated with optic nerve degeneration secondary to ischemia.
• Elevates neurotrophic factors important in preserving retinal ganglion cells after insult.

29 **Brimonidine**

• In laboratory studies, has demonstrated three out of four criteria required to demonstrate neuroprotective effects
  • Receptors on target tissue
  • Adequate penetration to the retina
  • Induction of intracellular changes that enhance neuronal resistance to insult in animals studies
  • To date, has yet to satisfy the fourth criterion: demonstrated efficacy in human clinical trials.

30 **Pregnancy and Glaucoma**
Conventional wisdom is to defer treatment during pregnancy.

There is a statistically significant drop in IOP during pregnancy (from 1st to 3rd trimester) (Dinh; Marris).

Mechanisms?
- Increase in outflow facility
- Decrease in episcleral venous pressure
- Development of a mild metabolic acidosis

Pregnancy Categories - Glaucoma
- Category B: Brimonidine
- Category C: Beta blockers, prostaglandin analogues, CAIs, cholinergics, apraclonidine

Pregnancy Categories
- Category A: Studies have failed to demonstrate a risk to the fetus in the first trimester of pregnancy (and there is no evidence of risk in later trimesters).
- Category B: Animal studies have failed to demonstrate a risk to the fetus and there are no adequate and well-controlled studies in pregnant women.
- Category C: Animal studies have shown an adverse effect on the fetus and there are no studies in humans, but potential benefits may warrant use of the drug in pregnant women despite potential risks.
- Category D: There is positive evidence of human fetal risk based on human studies, but potential benefits may warrant use of the drug in pregnant women despite potential risks.
- Category X: Studies in animals or humans have demonstrated fetal abnormalities and/or there is positive evidence of human fetal risk based human studies, and the risks involved in use of the drug in pregnant women clearly outweigh potential benefits.

Case #2
16yo HF
- CC: blurred distance vision x 1-2 months
- Established patient, LEE 2 years ago: unremarkable
- ROS: unremarkable
- Medication: None
- Ocular history: unremarkable
- Family ocular history: unremarkable

Uncorrected distance visual acuity
- OD 20/150 PH 20/50 OS 20/200 PH 20/50
- PERRL (-) APD
- Motilities full and comitant without pain/diplopia
- Confrontation fields: Generalized constriction OD/OS
- Refraction:
  - OD: -1.25 DS 20/50 OS: -1.00-0.75x005 20/50
- Anterior Segment: Unremarkable
- GAT (@2:55pm): 12 mmHg OD/OS
- Posterior Segment:

Follow up 1 week later
- No new visual/neurological complaints
- Uncorrected distance visual acuity
  - OD 20/150 PH 20/50 OS 20/200 PH 20/50
- PERRL (-) APD
- Motilities full and comitant without pain/diplopia
- Color Vision: OD/OS: test plate correct, 7/11 correct
- Blood Pressure: 104/50

Ordered labs, f/u 1-2 weeks
- CBC
- Serum levels of B1 (thiamine), B9 (folate), B12 (cobalamin)
  - If normal, may consider
    - FTA-ABS/VDRL
    - Heavy metal screen (e.g. Lead)
    - MRI

Follow up
- Patient placed on liquid multivitamin supplement
- Follow up 1 month

1 month follow up
- Vision improving, feeling better and has more energy
- Corrected distance visual acuity
  - OD 20/20- OS 20/20-
- Color vision: 17/17 OD/OS
Common Causes of Anemia

Common Causes of Vision Loss in Teenagers

Anemia

- Condition in which the body does not possess enough healthy red blood cells
- Various classification types
  - Morphological-Based on size (mean corpuscular volume)
    - Microcytic-MCV <80 fl
    - Macrocytic-MCV >100 fl
  - Pathogenic-Based on etiology

Anemia

- Adolescent females are 10 times more likely to develop anemia than adolescent boys (CDC)
- Mexican heritage and poor socioeconomic status have been shown to increase risk for developing the condition
- While anemia is a rare cause of vision loss, it can produce optic nerve ischemia, particularly in the setting of hypotension.
- Ischemic optic neuropathies related to anemia have been reported in cases of repeated gastrointestinal bleeding, trauma involving excessive blood loss, spinal and cardiac bypass surgeries, nasal/sinus surgery, and spontaneous abortion.
- Iron deficiency anemia has been associated with other optic neuropathies, including NAION and papilledema.

Case #3

9 yo HF

Previous Records:

- Subjective:
  - OD: +0.75-1.75×180 OS: -0.50 sph
  → BCVA OD/OS/OU: 20/60
- SLE/DFE: unremarkable
- A/P: New spectacle Rx given, RTO 1 month.

F/U visit 1 month later: No change in vision-BCVA remains 20/60.
- Ishihara: (?)Red-Green and Blue-Yellow Deficiencies
- A/P: Reduced BCVA/color. Refer to University

Case #3

9 yo HF

Case Hx (per parents):

- Difficulty localizing objects
• Excessive sensitivity to sunlight. Avoids outdoor activities.
• Difficulty functioning at night
• All visual difficulties began around age 5
• Mild speech impairment and learning disability
• Mildly overweight
• Normal pregnancy and birth. (+) Polydactyl
• No FHx of any visual/ocular conditions

57 Patient K.H.
• Corrected Distance Visual Acuity
  • OD/OS/OU: 20/60 PH:NI
• Motilities full and comitant
• PERRL (-)APD
• Refraction: OD: +0.75-1.75x180 OS: -0.50sph
  • →20/60 OD/OS/OU
• Ishihara: Test plate correct, all others missed
• Anterior segment: unremarkable
• GAT: 12/12 @ 10:30am
• Posterior Segment:

58

59

60

61 Mom

62 Mom

63 Dad

64 Dad

65 K.H.

66 Dad

67 GDX

68 Patient K.H.
• Summary of relevant findings:
  • Reduced BCVA (20/60)
  • Reduced color vision
  • h/o excessive sunlight sensitivity and difficulty functioning at night, polydactyl, LD,
overweight

- Clinically excessive retinal sheen confirmed by thickened RNFL, possible arteriolar attenuation
- Absence of PIL centrally and peripherally

**Patient K.H.**

- A/P
  - Presumed Early Cone-Rod dystrophy.
  - Educated on condition, potential for future progression of vision loss/visual impairment, sunglasses whenever outside. Parents encouraged to pursue educational services in school.
  - Defer ERG at this time

**Do We Need an ERG?**

- Microstructural retinal changes are commonly observed in patients with inherited retinal dystrophies using high definition OCT. (Gerth et al).
- Absence or interruption of the photoreceptor integrity line, as seen in inherited retinal dystrophies, is associated with reduced visual function.
- High resolution OCT images can be used to predict visual function through the presence, size, and integrity of the photoreceptor integrity line (Aizawa et al).

**Patient K.H.**

- Summary of relevant findings:
  - Reduced BCVA (20/60)
  - Reduced color vision
  - h/o excessive sunlight sensitivity and difficulty functioning at night, polydactyl, LD, overweight
  - Clinically excessive retinal sheen confirmed by thickened RNFL, possible arteriolar attenuation
  - Absence of junction centrally and peripherally

**Patient K.H.**

- Is there more to this case than a cone-rod dystrophy?

**Laurence-Moon-Bardet-Biedl Syndrome**

**Summary of relevant findings:**

- Reduced BCVA (20/60)
- Reduced color vision
- h/o excessive sunlight sensitivity and difficulty functioning at night, polydactyl, LD, overweight
- Clinically excessive retinal sheen confirmed by thickened RNFL, possible arteriolar attenuation
- Absence of junction centrally and peripherally

**Patient K.H.**

- Is there more to this case than a cone-rod dystrophy?
**Primary Features**
- Rod-Cone Dystrophy
- Polydactyly
- Obesity
- Learning Disabilities
- Hypogonadism in Males
- Renal Anomalies

**Secondary Features**
- Speech Disorder
- Brachydactyly
- Developmental Delay
- Polyuria
- Ataxia

78  **Secondary Features**
- Poor Coordination
- DM
- Left Ventricle Hypertrophy
- Hepatic Fibrosis
- Spasticity

**Diagnosis**
- Must exhibit 4 primary characteristics or 3 primary and 2 secondary.
  - Primary:
    - Rod-Cone Dystrophy
    - Polydactyly
    - Obesity
    - Learning Disabilities
  - Secondary:
    - Speech Disorder
    - Developmental Delay
    - Ataxia
    - Poor Coordination

**Treatment**
- The only treatment is the management of symptoms.
  - Vision- Low Vision Services
  - Obesity- Dietician
  - Kidney Problems-Medication/Transplant
  - Polydactyly- surgical removal
Demographics

- Prevalence rate in North America/Europe ranges from 1:14,000 to 1:16,000 live births (Sharifian et al).
- However, in some regions such as Kuwait, the rate is higher, with an estimated incidence of 1:13,500 due to a high frequency of consanguine marriages.
- While this syndrome is considered rare, it is suspected that there is a major problem with its under-diagnosis.

Genetics and Types

- Type 1 11q13 -- Most Common
- Type 2 16q21
- Type 3 3p12 -- Least Common
- Type 4 15q22
- There are patients with the disease that do not have any of the genes listed above.

Conclusions

- The previous cases provides examples where an optometric condition could act as the gateway to a larger system-wide dysfunction.
- Pathology that generally exists in adults can present in younger patients, often for different reasons.
- In giving a thorough optometric evaluation and making the necessary referrals you can improve/extend the lives of these young people.

References

- Gerth C, Zawadzki RJ, Werner JS, Héon E. 2008 Retinal morphology in patients with BBS1 and BBS10 related Bardet-Biedl Syndrome evaluated by Fourier-domain optical


References