Septo-optic Dysplasia: A Case Report
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Abstract:
A 16-year-old boy with history of seizures presents with reduced visual acuity in the left eye and left exotropia. Fundus findings include left optic nerve hypoplasia. MRI reveals absence of septum pellucidum.

I. Case History:
-CC: Blurry distance vision with Rx and eye turn
-Ocular History:
  LEE: 3 years
  Current Rx (3 years old): OD: -0.50D, OS: -0.50D
  (+) Watery eyes occasional
  (+) Left eye turned out since birth.
-Medical History:
  (+) Seizures since age 3. Patient was started on Depakote (500 mg daily) at age 16 and discontinued 6 months ago.
  (+) TB exposure, no infection, patient treated prophylactically with Isoniazid.
-Medications: Isoniazid
-Other information:
  Normal pregnancy.

II. Pertinent Findings:
-Clinical:
  -VA (c Rx): OD: 20/25, OS: 20/200
  -Confrontation visual field: OD: full, OS: generalized constriction.
  -Humphrey visual field: unreliable due to high false positives, with no descriptive visual field defect.
  -CT: 45ΔXT OS at distance and near.
  -EOMs: full, ductions full OD/OS
  -Pupils: OD: normal, OS: sluggish, trace APD (+)
  -Refraction: OD: pl/-0.75 X 90 VA: 20/20
OS: +0.50/ -1.00 X 20 VA: 20/20
- SLE: normal findings OD/OS
- IOP Goldmann (3:40pm): OD: 14mm Hg, OS: 16mm Hg
- Fundus exam: OD: C/D: 0.65, VH: 2.3, temporal pallor
  OS: C/D: 0.8, VH: 2.3, no temporal rim, general pallor
  Macula: flat and clear OD/OS
  Periphery: flat, intact OD/OS
- Red Cap test: inconsistent responses
- Stereo: 0’
- Photos of fundus
- Physical: unremarkable
- Laboratory tests: non-contributory
- Radiology tests:
  Brain MRI shows closed-lip schizencephaly, absence of septum pellucidum, and small-appearing optic nerves.

III. Differential Diagnosis

- Primary diagnosis: Septo-optic Dysplasia
- Differentials:
  Hereditary optic atrophies (Kjer, Behr, and Wolfram syndromes)
  Stabismus OS with secondary amblyopia
  Glaucoma

IV. Diagnosis and Discussion

Patient was diagnosed with Septo-optic dysplasia (SOD), also known as de Morsier’s syndrome. Septo-optic dysplasia is a rare congenital anomaly and is defined by having at least 2 out of three features of the classical triad: (i) unilateral or bilateral optic nerve hypoplasia, (ii) midline brain defects, including agenesis of the septum pellucidum and/or corpus callosum, (iii) pituitary hormone abnormalities.

With an incidence of 1/10 000 births, and affecting both sexes equally, Septo-optic dysplasia is associated with younger maternal age. Certain genetic mutations (HESX1 and SOX2) have been identified; however, these mutations are found in less than 1 % of the cases. Other associated features are developmental delay, seizures, short stature, obesity, visual impairment, strabismus, nystagmus, mental retardation, sleep disturbance, precocious puberty, cardiac anomalies and sensorineural hearing loss.
Our patient presented with two out of the three features: unilateral optic nerve hypoplasia and absence of the septum pellucidum. Seizures and strabismus are other associated features that were observed in this patient.

V. Treatment, management

Early diagnosis is important, as untreated hormonal abnormalities places a child at risk of neurodevelopmental abnormalities, hypoglycemia, adrenal crisis, and death. If patients are suspected of having septo-optic dysplasia, MRI and base-line endocrine tests should be performed, and hormone insufficiencies should be treated. Since GH replacement is available, the short-stature of some of these patients can be treated or prevented.

Optometrists and ophthalmologists play a crucial role in early diagnosis, as optic nerve hypoplasia is often the first manifestation of the syndrome. A thorough examination of the optic nerve appearance (hypoplastic margin and outer halo) is key, as well as other elements to look out for such as reduced visual acuity, nystagmus and strabismus.

The management of these patients consists of regular follow-ups, at least every 6 months with a multi-disciplinary team. Since pituitary insufficiencies may evolve over time, a life-long medical follow-up is necessary.

-Bibliography:

VI. Conclusion

Septo-optic dysplasia is a rare developmental disorder that is characterized by unilateral or bilateral hypoplasia of the optic nerve, pituitary hormone abnormalities, and midbrain defects. Optometrists play a crucial role in diagnosing this disorder, as the ophthalmoscopic findings are often the first manifestations. These patients should be forwarded to the pediatric-endocrinologist since pituitary deficiencies may be present.