Vision Rehabilitation in a Case of Bilateral Optic Nerve Hypoplasia and Vertical Nystagmus

A 60yo male with history of bilateral optic nerve hypoplasia and longstanding reduced vision presents with vertical nystagmus not previously noted. We describe differential diagnoses and management of optic nerve hypoplasia and nystagmus.

I. Case History

Patient demographics: 60yo WM Veteran born premature and placed in an incubator with 100% oxygen at birth. His mother was a Type I diabetic that had previously given birth to 2 children prematurely, both of whom died at birth.

CC: Presents for advanced low vision services with longstanding reduced vision OU

OHx:
Optic Nerve Hypoplasia OU
Pigment dispersion syndrome OU
Fuch’s Dystrophy OU

MHx:
Ataxic gait and reports balance issues
Type 2 diabetes
PTSD
Bipolar
Panic disorder

Medications:
Carbamazepine
Clonazepam
Fluoxetine
Aripiprazole
Oxybutynin
Previously taking lithium

II. Pertinent findings
Clinical

- Pupils normal OU
- Extraocular motilities full with a low frequency large amplitude vertical jerk nystagmus. Nystagmus present in all positions of gaze with the slow phase down and fast phase up
- Constant alternating exotropia
- Cranial nerve testing WNL
- VA OD is 4/100 wearing +4.25-7.50x090, VA OS is 6/100 wearing +7.25-11.00x090
- Anterior segment: guttatae and 1+ diffuse endothelial pigment OU
- IOP by Goldmann 15/22mmHg pre-dilation and 18/24mmHg post-dilation
- Posterior segment (Photos to be included)
  - Small hypoplastic nerves OU with “double ring sign”
  - Disc-macula distance to DD ratio ~4-5 OU
  - Subtle lightening of macular pigment OU
  - Mild vessel tortuosity OU
  - No signs of diabetic retinopathy or macular edema OU

Radiology: An MRI of the brain without contrast shows mild global cerebral volume loss suggestive of minimal microvascular disease. There is no acute intracranial abnormality.

III. Differential diagnosis

1. Small Optic Nerve Head
   a. Optic nerve hypoplasia
   b. Optic atrophy
   c. Ocular albinism
   d. Bilateral tilted discs

2. Nystagmus
   a. Cerebellar problem (Chiari malformation)
   b. Brain stem problem
   c. Poor VA
   d. Toxic/metabolic
   e. Wernicke encephalopathy
   f. Thiamine deficiency
   g. Thalamic hemorrhage
   h. Tumor
   i. Stroke
   j. Trauma
   k. Multiple sclerosis

IV. Diagnosis and discussion

Legally blind secondary to bilateral optic nerve hypoplasia OU.
Nystagmus workup did not elicit any new causes and is thought to be more congenital in nature due to history of longstanding decrease in VA.

Bilateral optic nerve hypoplasia is present at birth and has been documented to occur in premature infants and infants with diabetic mothers. It is a nonprogressive congenital abnormality with the hallmark sign of small optic nerve sizes usually 1/3-1/2 normal size. The loss of nerve fibers and ganglion cells can lead to a severe reduction in vision. Cerebral malformations are associated more in bilateral cases with the most common abnormalities being septo-optic dysplasia. MRI with attention to the hypothalamic-pituitary area is indicated in new diagnosis of bilateral cases.

V. Treatment, management

Through low vision rehabilitation services, patient meets his visual goals and activities of daily living. Devices prescribed:

- Monocular +12 round segment bifocal for habitual wear and spot reading
- 6x aspheric hand held magnifier and electronic magnification for near tasks
- 2.2x spectacle mounted telescope for watching TV

If current devices are not meeting goals, can also consider non-optical devices such as optical character recognition and inpatient/outpatient blind rehabilitation training.

VI. Conclusion/Clinical pearls

Optic nerve hypoplasia causes severe vision loss in individuals and is common in those born prematurely. With use of low vision rehabilitation, patients can perform activities of daily living. Providers should be aware of its presentation and management as well as common comorbidities. New onset nystagmus requires immediate attention to rule out life threatening disease.

Additional upon request.