Title: Management of Optic Atrophy in the Pediatric Special Needs Population

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Abstract:

A 4 year-old female with severe global developmental delays and history of Lennox-Gastaut Syndrome presents with history of long-term bilateral vision loss. Objective testing and Visual Evoked Potentials are used to assess visual status.

I. Case History

- Patient Demographics: 4 year-old Hispanic female
- Chief Complaint: Grandmother (legal guardian) requests an evaluation of patient’s visual status. Previous eye exams have diagnosed the child as blind, yet multiple spectacles have been prescribed. Grandmother reports that patient will occasionally grab at objects such as light up toys and colorful objects, leading her to believe that the child has some visual ability.
- Developmental History: Patient was born at nine months through natural birth. Guardian reports no complications during pregnancy or birth. Patient currently has not reached age normal motor, verbal, or cognitive milestones. She is unable to speak, walk or crawl at this time. She has been evaluated to have greater than a 75% motor development delay, greater than a 33% receptive and expressive language delay, and falls in the low range daily living skills range for her age.
- Ocular History: Patient was diagnosed with blindness at the age of three months in Coney Island Hospital.
- Medical History: History of Lennox-Gastaut Syndrome, microcephaly, deafness, and infantile spasms. Surgical history of cochlear implant and gastrostomy tube insertion.
- Medications: Pulmicort and Albuterol as needed. History of Zantac, Onfy, Valproic Acid, and Levocarnitine daily.
- Family History: Mother is also mentally delayed. Ocular related family history is unremarkable.
- Other pertinent information: With occupational, physical, and speech therapy the patients muscle strength and vocalization have improved over the past seven months.

II. Pertinent findings

- Visual Acuity: Light Perception OD and OS
- Bruckner: equal reflexes OU
- Lensometry: OD +1.75-2.75x175 OS +1.50-1.50x005
- Dry Retinoscopy: OD +2.00-1.50x180 OS +2.00-1.50x180
- Pupils: OD and OS equal, round and minimally reactive, (-) APD
- EOMs: Gross observation of patient demonstrated full movements horizontally and vertically with both eyes, (-) nystagmus
- Anterior Slit Lamp Examination: unremarkable OD and OS
- Tactile Pressure: Moderate to pressure and equal OU @ 10:29 am
- Dilated fundus exam:
  - Macula: normal structure to view OD and OS
  - Optic Nerve: 0.30 c/d with temporal pallor OD and OS
III. Differential Diagnosis

- Primary: Optic Nerve Atrophy due to Neurodegenerative Disease
  - Diagnosis of microcephaly and history of encephalopathy
- Alternate Causes of Optic Nerve Atrophy in Pediatric Population (ruled out per previous medical records and guardian history)
  - Prematurity
  - Ischemic or hypoxic event at birth
  - Inflammatory or infectious insult
  - Trauma Related
  - Toxic Optic Neuropathy
- Compressive Optic Atrophy
  - MRI’s have been performed after diagnosis of blindness
- Autosomal Dominant Optic Atrophy
  - No family history to support this diagnosis
- Leber’s Hereditary Optic Nerve Atrophy
  - No family history of disease and patient does not fit age demographic

IV. Diagnosis and discussion

- Lennox-Gastaut Syndrome (LGS) - also titled Childhood Epileptic Encephalopathy (Glauser, 2007)
  - A “pediatric epilepsy syndrome that is characterized by multiple seizure types, mental retardation or regression, and abnormal findings on electroencephalography (EEG).”
  - LGS has a prevalence of 0.1-0.28 per 1000 and an annual incidence of 2 per 100,000 children. It accounts for 1-4% of patients with childhood epilepsy and 10% of children with an onset of epilepsy before the age of five. Infantile spasms occur in 9-39% of children before the development of LGS.
  - The pathophysiology of the syndrome in unknown and no animal models currently exist.
  - An EEG is performed for diagnosis of the condition and further neuroimaging is used to determine the etiology. There are no neurological examination findings or laboratory tests that are used for the diagnosis of the condition.
  - Treatment includes a variety of antiepileptic agents and surgery to reduce the frequency of seizures. Long-term prognosis is overall unfavorable for these patients.
- Encephalopathy is a broad term for any condition that alters the structure or function of the brain. Similar to optic nerve pallor, there are a multitude of causes and variable effects depending on severity.
  - Damage is rarely confined to one area of the brain. In these cases, multiple areas of the visual system may be affected: “primary visual cortex, visual associative cortex area, optic radiations, optic nerves, and visual attention pathways.” (Hoyt, 2007)
  - The visual diagnosis, systemic associations, and prognosis of visual ability are highly variable depending on which areas of the brain are injured.
  - One study identified hypoxic ischemic encephalopathy to be the most frequent cause of nerve pallor in their population of 324 children diagnosed with unilateral or bilateral optic atrophy. Of these children, a majority has had a history of delayed developmental milestones along with seizures. The remaining children were associated with prematurity or microcephaly. (Chinta et al., 2014)
• Optic nerve atrophy in the pediatric population has commonly been known to be associated with compressive tumors, but a recent study found that perinatal events and neurodegenerative diseases have become the two most common origins. (Zheng et al., 2016)
  o Perinatal events can include prematurity itself or an array of events: ischemia, hypoxia, inflammation, trauma, and toxicity.
  o Nerve pallor is the result of irreversible damage along any area of the anterior or posterior visual pathway, resulting in permanent visual loss. Anterior pathway damage leads to the loss of retinal ganglion cells while posterior pathway damage can cause transynaptic atrophy.
  o Preliminary testing in these children shows a range of reduced visual acuity, color vision, visual fields and minimal pupillary reaction to light. If the atrophy is congenital and involves both eyes, it is possible to note nystagmus.
• Furthermore, optic nerve pallor and visual disorders are found more regularly among children in the special needs population.
  o “The higher prevalence...is accounted for by the many underlying causes of their disability with prenatal and perinatal factors and acquired injury all of relevance.” (Salt et al., 2014).
  o Regularly associated medical conditions include hydrocephalus, developmental delays, seizures, cerebral palsy, and microcephaly. (Zheng et al., 2016)
• Auxiliary testing can serve in assessing and managing a globally delayed child.
  o Visually Evoked Potential (VEP) testing can objectively symbolize the functionality of the patients’ pathway from the retina to the visual cortex.
    ▪ Visual acuity, contrast sensitivity, and color vision can be tested using a Pattern VEP.
    ▪ The P100 wave is the major component of the VEP that is used to determine visual acuity. This positive wave on the test remains reliable for patients ranging from 5 to 60 years old. (Creel, 2016)
  o B-scan Ocular Ultrasounds can be used as a non-invasive procedure for general evaluation of ocular structure when it is difficult to have a patient fixate or tolerate the light for fundus examination.
• Interdisciplinary management is critical in providing the best functional outcome for these patients. It is important to fully understand their visual capabilities and inabilities so other healthcare professionals are able to successfully integrate the visual system into their rehabilitative methods.

V. Treatment/Management

  1. Exam findings indicate that the patient is light perceptive in both eyes. A Pattern VEP was performed with and without glasses. Testing indicated that there was no difference in cortical stimulus between the trials, signifying that spectacle lenses provide no visual gain for this patient.

  2. Flash VEP is to be performed at a follow up appointment to determine if the patient is indeed light perceptive. This information will assist the patients’ therapists in coordinating activities to continue improving her motor and vocal abilities.
VI. Conclusion

Special needs patients present with high variability in visual and overall function. It is important to assess their visual system to full capacity and determine the degree in which they can be helped. Further auxiliary testing allows determination of appropriate management, both visually and through interdisciplinary coordinated care, to provide these patients with the highest quality of life.

References