Abstract: A 7 year old boy presented with vision loss due to optic nerve compression from the rare autosomal recessive disorder of osteopetrosis, previously diagnosed as amblyopia.

I. Case History
- 7 year old boy recently moved from Oklahoma City
- Chief complaint: Referred to neuro-opthalmologist Dr. Rosa Tang for disc swelling OS by optometrist who was following child for possible amblyopia of OS x 1 year.
- Medical History: The patient is diagnosed with Osteopetrosis. C3 spinal fracture, and rib fracture in 2010. No past surgical history, no current medications, no allergies.
- Ocular History: Patient was noted to have possible optic nerve edema of left eye 1 year ago, and was sent for imaging. MRI of the brain came back normal.
- Social History: Patient is currently in the second grade. The child is able to read. There are no developmental issues noted, and no frequent infections.

II. Pertinent findings -
- BCVA: 20/20 OD, 20/400 OS
- Manifest refraction: OD: +0.75-0.75x175 20/20
  OS: +0.50-1.00x085 20/400
- (+) left APD. (-) Horner’s (-) efferent defect (-) ptosis (-) proptosis.
- Sensory motor exam: 20 pd Esotropia OS. (-) restrictions on ductions and versions. (-) nystagmus. (-) cranial nerve paresis.
- Worth 4 dot: Suppression of OS @ distance + near
- IOP: 14mmHg OD, 16mmHg OS. No intraocular inflammation, no cranial nerve abnormalities.
- Fundus evaluation: OD: c/d 0.15x0.15 with disc edema. Vessel Tortuosity. OS: c/d 0.4x0.4 with disc edema and temporal pallor. Vessel Tortuosity.
- HVF: non-specific changes on both VF with scotomas.
- OCT: reduced thickness of NFL OS> OD with nerve fiber bundle defect.
- VEP: normal for OD. Normal amplitude but moderately delayed latency OS.
- Fundus Photography: Disc edema OU with temporal pallor OS
- Ultrasound: Bilateral perioptic fluid highly suggestive of disc edema.
- MRI of brain: normal

III. Differential diagnosis
- Strabismic Amblyopia
- Optic neuritis
- Compressive lesion
- Orbital pseudotumor
- Other inflammatory conditions (lyme disease, non-demyelinating disease, infectious disease)
IV. Diagnosis and discussion

- **Diagnosis**: Optic nerve atrophy due to optic nerve compression from osteopetrosis
- Osteopetrosis is a rare autosomal recessive disorder caused by mutations in the TCIRG1, CLCN7, and OSTM1 genes. The genetic mutations result in the inability of the osteoclasts to properly breakdown old bone as new bone is being formed. Therefore, the bones in the body overgrow, become more dense and are more prone to fractures than the normal bone. Visual loss from osteopetrosis is progressive, and almost always occurs within the first year of life.
- Osteopetrosis is not associated with any mental or physical disabilities. Caution should be taken to not isolate the child in their everyday activities, however it is often recommended for the child to refrain from contact sports and for safe play to be encouraged.

V. Treatment, management

- Currently, first line treatment for optic atrophy due to osteopetrosis is decompression of the bones of the optic canal to alleviate the compression of the optic nerve and salvage the vision that the patient has remaining. An improvement in visual status of the patient is unlikely, despite treatment efforts. The goal of treatment is to maintain the vision that is still remaining.
- Visual Evoked Potential is the most useful way of monitoring optic nerve involvement. Severely affected children may show absent or severely attenuated VEPs within the first 3 months of life.

VI. Conclusion

- Loss of vision with optic nerve atrophy is the most common neurological finding in osteopetrosis. Retinal degeneration is a new hypothesis for the cause of the vision loss, however this theory is unproven. Affected children usually present within the first year of life with visual loss, due to the bony encroachment of the optic nerve. Orbital decompression is first line therapy to try to prevent further loss of vision, and to maintain the remaining vision. This patient’s vision loss in the left eye was first diagnosed as possible amblyopia. Amblyopia is a diagnosis of exclusion, and all efforts should be made to not diagnose and treat someone as amblyopic when an underlying condition could possibly exist. Osteopetrosis is a rare progressive disease, and had the diagnosis of optic nerve compression due to osteopetrosis been made sooner, then some of the patient’s vision may have been preserved. As optometrists, we are capable of providing proper care and treatment for these patients by conducting ancillary testing such as OCT to display NFL defect, VEP, and by referring for imaging scans or for further evaluation. Careful examination of all data is needed to provide the proper diagnosis and to rule out more serious causes for the vision loss.

Bibliography:
