Abstract title: Vision loss from myelinated retinal nerve fiber layer with maculopathy.

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General Topic: Ocular Disease
Primary Topic: Posterior Segment

Abstract: The proliferation of myelinated retinal nerve fibers with amblyopia, myopia, and maculopathy as a syndrome has been rarely reported. The review addresses these findings and impact to patients.

- Case History
  - Patient demographics
    - 57 year old black female
  - Chief complaint
    - Presented for routine eye exam: reports longstanding poor vision in the right eye since childhood.
  - Ocular, medical history
    - Pertinent Ocular History:
      - LEE 2 years ago. Patient reports previous doctors have noted poor acuity out of right eye, but patient does not recall diagnosis.
    - Medical History:
      - HTN; diagnosed 2010, takes Amlodipine 5 mg PO QD
        - in office blood pressure: 140/85, 1:42PM
      - (-)DM
  - No pertinent family history

- Pertinent findings
  - Clinical
    - Entering VA’s without specs
      - OD: 20/400 PHNI
      - OS: 20/20
    - Cover Test
      - Distance: 2-3 CRHT, 2 XP (sc)
      - Near: 2 CRHT, 5 XP (with +1.75sph OU)
    - Comitant
  - Refraction
    - Dry ret
      - OD: -5.75-1.50x090 (NI)
      - OS: -0.50sph
    - Wet Auto
4. Pupils: PERRL (+)trace APD OD
5. Extra ocular muscles: full without restrictions OU
6. Confrontation Visual Fields: full to finger counting OD/OS
7. Tonometry (Goldman with 1 gtt proparacaine 0.5% and NaFl strip)
   1. OD: 12 mmHg
   2. OS: 14 mmHg
8. Color Vision
   1. OD: pass (Ishihara; poor response on HRR)
   2. OS: pass 6/6 (HRR)
9. Amsler Grid
   1. OD: (+)general metamorphopsia, reports “blur”
   2. OS: normal
   3. Physical
      1. Slit lamp: unremarkable OU
      2. Dilated Fundus Exam (1 gtt 1% Tropicamide, 1 gtt 2.5% Phenylephrine)
         1. Lens: clear OU
         2. Vitreous: marked syneresis OD, syneresis (+)PVD OS
         3. Optic disc:
            ▪ OD: obscured
            ▪ OS: 0.50; round, pink, distinct
      4. Vessels:
         ▪ OD: normal; peripheral perivasculat hyperpigmentation
         ▪ OS: normal vessels
      5. Macula:
         ▪ OD: pseudohole
         ▪ OS: flat, no hemorrhages, exudates, pigmentary changes, no macular edema
      6. Periphery:
         ▪ OD/OS: flat x 360, no RD, no holes
   4. Extra studies
      1. Posterior segment photos
      2. SD-OCT
         1. Thickening of NFL in area of myelination
         2. Partial thickness hole in ectopically positioned macula
      3. A-scan for axial length measurement
         1. OD: 24 mm
         2. OS: 23 mm
      4. B-scan
         1. OD: myelination, (-)detachments, calcification, tumor, drusen
      5. Goldmann Visual Field (targets III and V)
1. OD: a scotoma corresponding to MRNF is demarcated within a normal functioning peripheral field

6. Humphrey Visual Field
   1. SSA Test Kinetic (III 4 E target)
      - OD: Plots extent of patients peripheral vision without indication of scotoma
   2. SITA-FAST 30-2 Threshold Test
      - OD: Inferior arcuate scotoma corresponding to scotoma from MRNF

➢ **Differential diagnosis**
   - Primary/leading: myelinated retinal nerve fiber layer OD with amblyopia, maculopathy, and myopia.
   - Others: pre-retinal fibrosis, drusen, choroidal osteoma

➢ **Diagnosis and discussion**
   - Elaborate on the condition
   1. Myelinated retinal nerve fibers (MRNF) are a congenital anomaly of the fundus that occurs in about 1% of patients (1). It is more prevalent in patients with congenital anomalies such as neurofibromatosis, Down syndrome, and craniofacial dystostosis (6). Composed of lipoproteins, MRNF is typically a non-visually impeding condition. However, it has been associated with high degrees of myopia and vision loss either due to amblyopia (anisometropic or deprivational) and/or maculopathy. In cases of associated myopia, the degree of anisometropia typically does not correlate with the degree of vision loss and amblyopia.
   2. The myelination of neurons in the central nervous system is coordinated by oligodendrocytes which insulate and accelerate conduction of signals along axons (1). In normal development, optic nerve myelination starts from the optic chiasm to the optic nerve from the eighth month of gestation until birth; the extent of myelination ends at the lamina cribrosa, the site of astrocyte aggregation that is thought to be a barrier to prevent myelination to disperse into the retina (1, 5). Thus while MRNF proliferation can be a developmental anomaly, it can also be due to insult to the lamina cribrosa (i.e. increased intraocular pressure causing deformation of the optic nerve head causing thinning of fibrotic connective tissue structure/scar and peripapillary atrophy. (3)). Given that the lamina cribrosa is believed to be the site of injury to retinal ganglion cell axons in glaucoma, it is a reasonable thought that patients with MRNF proliferation are at a higher risk of the disease. Another theory for the pathogenesis of MRNF is that of the result of abnormal migration of oligodendrocytes into the retina prior to the full development of the retina leading some to describe the condition as an oligodendrocytic choristoma (7).
3. There is a reported association of MRNF with myopia and amblyopia (refractory and/or stimulus deprivational). It has been hypothesized that the MRNF interfering with vision actually stimulates axial elongation, leading to higher levels of myopia (1). Evidently in our patient, the axial length of the eye with the MRNF was roughly 1 mm longer than its normal counterpart.

- Expound on unique features
  1. Our patient’s refractive status only reveals a mildly amblyogenic factor (2). The maculopathy alone was also not consistent with the degree of vision reduction. It is likely that her reduced acuity is contributed to both processes.
  2. MRNF is often congenital but can be acquired or even progress in adolescence (5).
  3. MRNF can regress in the presence of several conditions such as chronic glaucoma, anterior ischemic optic neuropathy, branch retinal artery occlusion, radiation retinopathy following plaque radiotherapy for choroidal melanoma, progressive diabetic retinopathy, and laser photocoagulation of diabetic retinopathy. It has even been reported to disappear in a patient with Behcet syndrome with recurrent episodes of papillitis and vitritis (1).

- **Treatment, management**
  - Treatment and response to treatment
    1. Due to the likely longstanding nature of her condition, we decided to monitor her condition with yearly dilated exams. We advised her to fill her prescription as PALs (balanced OD) to allow for full time wear and protection of her remaining 20/20 left eye.

- **Conclusion**
  - Clinical pearls, take away points
    1. The prognosis of occlusion/vision therapy to improve visual acuity in patients with optic nerve or macular-involving MRNF is variable, but likely guarded to poor, as there is an organic pathology impeding normal visual input.
    2. It is imperative to photo-document MRNF due to the possible progressive nature of this condition. If vitreomacular traction is suspected, a macular OCT should be done. This case demonstrates that MRNF can be associated with maculopathy, along with amblyopia and myopia as previously reported.
    3. Subsequent examinations with dilation is important as retinal tissue in the area of MRNF are more prone to neovascularization and vitreous hemorrhage or branch artery or vein occlusion (1).
Bibliography


