



Xerophthalmia in a 7 y.o. patient with autism and severe dietary restriction

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Introduction

Xerophthalmia from vitamin A deficiency is a rare cause of vision loss in developed countries. However, worldwide it is estimated to blind half a million children each year (1). Etiologies include a severely restricted diet, malabsorptive process, inflammatory bowel disease, or post-gastric bypass surgery. The typical ocular presentation includes dry eye, corneal scarring and ulceration, and nyctalopia as a result of the metaplasia and keratinization of mucus-secreting epithelium and impaired rod function. Ocular findings include: conjunctival and corneal xerosis, bitôt spots, corneal scarring, and decreased scotopic amplitudes on electroretinogram (2).

Case Presentation

- Seven year old male with history of autism spectrum disorder presents with photophobia, angioedema, and headache, worsening over multiple months.
- Severe lifelong dietary restriction consisting of chips, muffins, pasta, and dry apples.
- Ophthalmic exam reveals angioedema, meibomian gland dysfunction, blepharitis, conjunctival xerosis, severe superficial punctate keratopathy, right sided corneal stromal haze, and mild bilateral optic nerve head edema (Figures 1 and 2).
- MRI brain and orbits, MRV, CT head, Bscan and lumbar puncture with opening pressure are within normal limits and without evidence of optic canal stenosis or disc drusen. Infectious and inflammatory workup unremarkable.
- Labs revealed microcytic anemia w/ hemoglobin 7.4 g/dl and hypoalbuminemia 2.2g/dl (normal 3.8-5.4 g/dl) which were attributed to malnutrition. Vitamin A levels were undetectable. Vitamin B12 levels were deficient 182 pg/ml (normal 211-946pg/ml).
- MRI abdomen and MRCP revealed pancreatic pseudocyst with pancreatic duct laceration and ascites. One week after starting vitamin A replacement and frequent ocular lubrication, the patient's ocular symptoms improved greatly. Vitamin A levels one month after replacement were within normal limits.

Testing

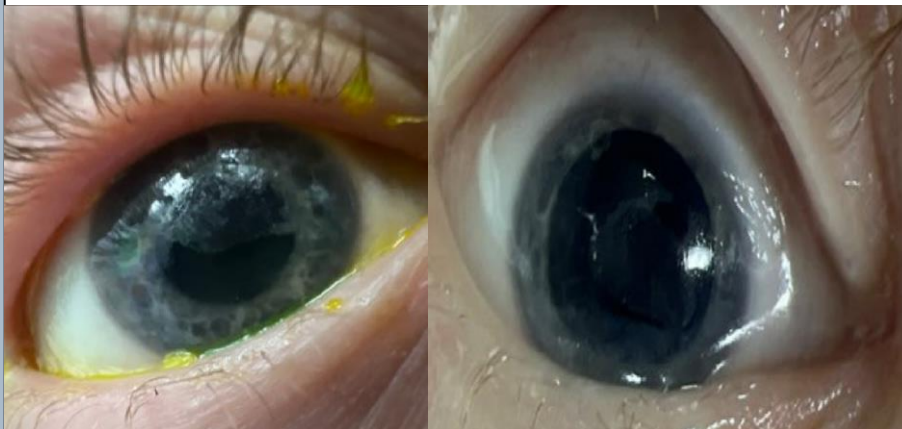


Figure 1. Superficial keratitis right eye (image on left), conjunctival xerosis right eye (image on right)

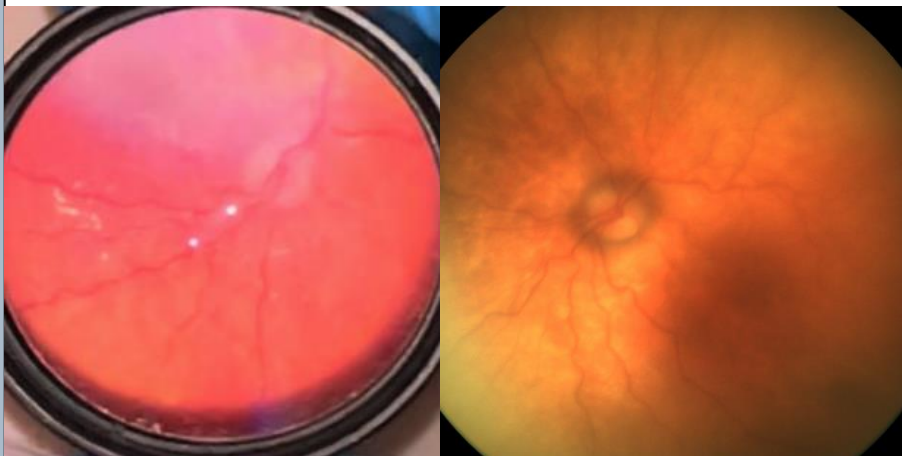


Figure 2. bilateral mild optic nerve head edema

Discussion

Xerophthalmia remains a challenging diagnosis in the developed country due to its rarity. Early identification and prompt treatment may result in complete resolution of symptoms in as little as two months (3). Workup should include evaluation for malabsorptive process and accurate history taking. In our case, the deficiency was likely multifactorial as a deficiency in pancreatic enzymes that help metabolize fat soluble vitamins and dietary restriction due to autism. Zinc levels should also be evaluated as it is important in the synthesis of retinol-binding protein which is required to metabolize retinol from the liver (4). Xerophthalmia results from goblet cell dysfunction and impaired rod function as vitamin A is a precursor to the photopigment rhodopsin (1).

Conclusion

- Xerophthalmia remains a rare cause of vision loss in developed countries but is the leading cause of childhood blindness worldwide.
- Vitamin A deficiency should be suspected in those with dry eye and a severely restricted diet, history of gastric bypass surgery, or other GI malabsorptive process.
- Zinc deficiency should also be ruled out
- Patients may have resolution of their visual function after initiating prompt treatment.

References

1. World Health Organization. Prevention of Childhood Blindness. Geneva: The Organization, 1994.
2. Chiu M, Dillon A, Watson S. Vitamin A deficiency and xerophthalmia in children of a developed country. J Paediatr Child Health. 2016 Jul;52(7):699-703. doi: 10.1111/jpc.13243. PMID: 27439630.
3. Steinemann TL, Christiansen SP. Vitamin A deficiency and xerophthalmia in an autistic child. Arch Ophthalmol. 1998 Mar;116(3):392-3. doi: 10.1001/archoph.116.3.392. PMID: 9514500.
4. Hodge C, Taylor C. Vitamin A Deficiency. [Updated 2021 May 9]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2021 Jan-. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK567744/>



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