Conjunctival Amyloidosis: An Unlikely Cause of Unilateral Ptosis

Outline

I. Case History
   a. 41 year old Caucasian male presents for ptosis evaluation
   b. Chief complaint: Ptosis OD, gradual progression over 10 years
   c. Ocular History: Low risk glaucoma suspect secondary to elevated IOP OU
   d. Medical History: History of hernia repair
   e. Medications: Advil PRN
   f. Additional case information: Patient initially presented at age 39 with a ptosis OD. He reported progression over 5+ years; however, at that time he elected for observation of his condition.

II. Pertinent findings
   a. Clinical: Significant ptosis OD, MRD -1mm OD vs 4mm OS, Salmon-colored lesion on superior palpebral conjunctiva
   b. Physical: None
   c. Lab studies: Normal CBC, Metabolic Panel, SPEP, SIEP, spot urine, UPEP
      1. Systemic work-up completed by PCP after tissue biopsy confirmed amyloidosis OD.
   d. Radiology studies: MRI shows enhancing lesion isolated to the superior preseptal soft tissue along with chronic left maxillary sinusitis with antral contraction. The patient has a history of trauma as a former hockey player, to which his left sinus findings can be attributed.
   e. Other: Tissue biopsy consistent with localized conjunctival amyloidosis.
      1. Remarkable for aggregates of eosinophilic material.
      2. Congo Red Stain showing green birefringence on polarized light exam. Pathognomonic for Amyloidosis.

III. Differential diagnosis
   a. Conjunctival lymphoma
   b. Sebaceous cell carcinoma
   c. Ligneous conjunctivitis

IV. Diagnosis and Discussion
   a. Periocular/orbital amyloidosis is a rare condition
      1. Most commonly involves the conjunctiva; however, can involve the lacrimal gland, lacrimal sac, extraocular muscles, or ocular adnexa.
      2. Typically, presents as a visible or palpable mass. Specifically, conjunctival lesions present with edema, conjunctival irritation, papules, and yellow plaques.
      3. May cause ptosis, proptosis, discomfort, subconjunctival hemorrhage, or ocular motility restriction.
   b. Amyloidosis may be localized or systemic, and is caused by extracellular deposition of misfolded, insoluble fibril proteins.
c. Two main types: AA Amyloidosis, AL Amyloidosis – Differentiated by immunohistochemistry; Named for the involved protein.
   1. AA Amyloidosis – Involves Amyloid A protein, which is formed as a result of longstanding inflammation.
      1. Associated systemic conditions: RA, Psoriasis, Inflammatory Bowel Disease, Sjogren’s syndrome
   2. AL Amyloidosis – Involves light chain proteins
      1. Associated systemic conditions: Multiple myeloma, Waldenstrom’s macroglobulinemia, Non-Hodgkin’s lymphoma
      2. More commonly associated with mucous membranes
d. Other potential organ involvement: Heart, Kidneys
e. Diagnosis of localized amyloidosis requires a systemic work up to rule out other organ involvement.
   1. Systemic involvement can be confirmed with a biopsy of subcutaneous fat. In order to determine specific organ involvement, blood testing, urinalysis, and cardiovascular work up are required.

V. Treatment and Management
   a. Treatment options for localized lesions include:
      1. Observation
         - If patient is comfortable and lesion is not progressing, observation is a viable option for treatment.
      2. Surgical excision
         - Most common treatment option.
      3. Cryotherapy
         - Typically adjunctive therapy, interrupts blood supply to amyloid lesions and may prevent recurrence.
      4. Radiotherapy
         - Typically adjunctive therapy, especially in the case of deep orbital disease which cannot be fully excised.
   b. Systemic treatment:
      1. AA Amyloidosis is typically treated by treating the underlying inflammatory condition.
      2. AL Amyloidosis is commonly treated with immunosuppression, such as chemotherapy.
   c. Since our patient was found to have localized amyloidosis, he elected surgical excision, which is being scheduled at this time.
   d. Literature Review:

VI. Conclusion
   a. Amyloidosis, though rare, must be considered as a differential diagnosis in orbital and periocular lesions.
   b. A systemic work up following a diagnosis of amyloidosis is crucial to the proper treatment of the disease.