CLINICAL VIGNETTE

Ocular Amyloidosis

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The patient was a 67-year-old female with a recent diagnosis of amyloidosis. The patient noted a left eye conjunctival lesion since 2005. She was monitored by Ophthalmology as the area looked benign and the patient had no symptoms from the abnormality. The lesion slowly grew over time and eventually began to impact her vision and caused an eye fullness as it moved towards the cornea, so resection was recommended. The pathology incidentally noted amyloidosis, confirmed by Congo stain. Further amyloid typing distinguished it as AL-kappa subtype. She was consequently referred for hematology evaluation.

At presentation the patient noted chronic shortness of breath related to asthma but no other symptoms. Her history included morbid obesity, gastric reflux, and sleep apnea as well but no history of cardiac, hepatic, gastrointestinal, or renal issues. Laboratories were obtained to look for a monoclonal gammopathy and indicated a normal serum protein electrophoresis, serum immunofixation, complete blood count, complete metabolic panel, quantified immunoglobulins, and beta2microglobulin. While her kappa light chains were mildly elevated at 2.02 mg/dL (upper limit of normal 1.94 mg/dL) her kappa/lambda ratio was very normal. Positron emission tomography noted no hypermetabolic disease. Echocardiogram noted normal ejection fraction of 55% and normal ventricular wall thickness. Computed tomography of the orbits showed likely postoperative changes in the left medial globe and no other masses or abnormal findings. Magnetic resonance imaging of the orbits similarly indicated no residual or recurrent left medial conjunctival mass and her anatomy otherwise appeared normal. Bone marrow biopsy pathology showed a normocellular marrow, 40-50% cellularity with multilineage maturation and no evidence of amyloid deposition or plasma cell neoplasm. FLOW cytometry and cytogenetics were normal. The patient was referred to Radiation Oncology to consider further local therapy, but given potential secondary side effects, she opted to not proceed.

Given the patient was relatively asymptomatic and evaluation did not indicate any systemic disease, she was monitored expectantly. She had serial ophthalmology exams with no changes over the next two years, and serial monitoring for a monoclonal gammopathy on bloodwork remained unremarkable including no changes in her mildly elevated kappa protein.

Amyloidosis can be a difficult disease impacting multiple organs in the body.[1,2] Symptoms vary depending on pattern of deposition.[1,2] It occurs due to misfolded proteins depositing into sheets on normal tissue.[1,2] The heart, kidneys, gastrointestinal tract, and liver are common areas of deposition and the various eye structures are rarely involved.[1,2] Amyloidosis refers to a diverse group of illnesses but the most common is light chain amyloidosis (AL), which is associated with a clonal abnormality such as seen with multiple myeloma or other B-cell disorders.[2] Usually the disease is systemic and can be fatal if organs are involved.[2] Rarely, localized disease is noted.[2]

This case illustrates, another report of localized amyloidosis. One case reported localized disease occurs in 7-12% of all diagnoses.[2] Of those, about 4% are estimated to occur in the orbit.[2] Some reports indicate that amyloidosis of the eyelid tends to be associated with systemic disease while conjunctival findings are generally more localized.[1] Regardless of the location in the eye, symptoms tend to be blurriness, eye bulging, diplopia.[1,2] Appropriate evaluation includes biopsy of the lesion to confirm the diagnosis and to exclude other etiologies.[1,2] Further evaluation for systemic disease is imperative as it may guide further systemic treatment plans.[1,2]

Given the rarity of ocular amyloid deposits, there is no consensus on treatment.[1] Some advocate lubricants and steroids for symptom control.[1] Surgical intervention can be considered but there is ongoing concern for recurrence and impairment of function with aggressive resection.[1,3] Other modalities have been used including electrocauterization, cryotherapy, radiotherapy, and curettage.[3] While the patient presented here did not proceed with radiation due to limited data and no clear residual disease, there have been reports that radiation after debulking surgery can help reduce progression of amyloid deposition.[2] Glaucoma is a risk with the disease due to interruption of the normal anatomy with amyloid infiltrates.[1] In these cases, surgery may be favored as medications are less likely to be effective.[1]

This patient had already completed excision of the ocular lesion given impact on her eyesight. She was not felt to need further surgery and deferred radiation. Systemic workup was unremarkable at baseline as noted above and subsequent lab follow up for her minimal monoclonal gammopathy has continued to be stable three years after diagnosis. She has had no further growth in the area and remains asymptomatic. As such, it highlights that while in general AL amyloidosis can be a very dangerous illness, there are rare cases that may not require immediate systemic treatment. Given the rarity of

localized disease, it is imperative to continue to monitor for the development of more serious systemic manifestations.

REFERENCES

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