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Pharyngeal Amyloidosis: A Case Report with Review of Literature

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Abstract

Amyloidosis is a disease characterized by predominantly extracellular deposits of non-soluble fibrillar proteins, known as amyloid, in organs and tissues. Amyloid deposition can cause tissue damage, organ failure and death. It can present in localized and systemic forms with nodular or diffuse submucosal presentation. Localized amyloidosis in head and neck is rare with most of the cases presenting with laryngeal amyloidosis. Pharyngeal amyloidosis is very rare. Here we present a case of 59 year old female with pharyngeal amyloidosis who presented with complains of dysphagia and lateral pharyngeal wall swelling, with a review of literature. The patient underwent complete surgical excision of the pharyngeal mass. Although a rare disease, amyloidosis should be considered as differential diagnosis for smooth pharyngeal mass.

Keywords: Amyloidosis; Dysphagia; Pharyngeal Amyloidosis; Localized Amyloidosis; Pharyngeal Mass

Introduction

Amyloidosis is a term used to refer to rare disorders characterized by presence of deposits of amorphous extracellular eosinophilic deposits of unique abnormal protein fibrils called amyloid [1,2]. These protein fibrils demonstrate apple-green birefringence when examined under polarized light, after staining with Congo-red [3]. Amyloidosis may present as a systemic disease or be limited to certain organs and is more prevalent in males at a 3:1 ratio with more presentation in the 5th decade of life [4]. In localized form, amyloidosis can present with head and neck involvement which amounts to about 20 % of cases and most of them present as laryngeal amyloidosis [5]. Localized amyloidosis of the pharynx are very rare with limited reported cases [6,7]. Here we present a case of 59 year old female who presented with dysphagia and right lateral pharyngeal wall mass which was diagnosed as pharyngeal amyloidosis after histopathology examination.

Case Presentation

A 59 year old female with no known comorbidities presented to the ENT OPD with complaints of difficulty in swallowing for about a month and swelling in right side of throat. The swelling had not increased since then. There were no constitutional symptoms or weight loss. On clinical examination a single well-defined, raised, globular mass was seen on right lateral pharyngeal wall that was about 2 x 2 cm in size, yellowish in colour, with smooth surface, non-bleeding and firm in consistency. A lesion of about 1x 0.5 cm in size with similar features was also noticed in vallecular region near epiglottis Figure 1. No clinically palpable

cervical lymphadenopathy was noted. There was no history of similar illness in the past or any other relevant personal history. There was no family history of similar illness or any lymphoproliferative disorders. All routine laboratory blood parameters, ECG, urinalysis and chest X-ray were normal. On radiological evaluation, a fairly well-defined T1 and T2 hypointense with mild peripheral hyperintense rim lesion of approximately 10 x 9 x20 mm (AP x Trans x CC) seen involving right posterolateral wall of nasopharynx and oropharynx, showing subtle peripheral post-contrast enhancement was noted. Patient underwent panendoscopy and complete excision of lesion with assistance of KTP laser under general anesthesia. KTP laser was used for easy accessibility. Histopathology examination of the lesion revealed sections with mucosal tissue bits lined by squamous epithelium without any atypia and stroma with amorphous extracellular eosinophilic material with focal collections of mature plasma cells and few lymphocytes. Few capillary sized blood vessels with unremarkable submucous glands were also noted. Congo red staining was done that showed apple green birefringence, based on which final diagnosis of Amyloidosis was given Figures 2a & 2b. Post-operative period was unremarkable with well healed surgical site. On follow up, no recurrence has been reported Figure3.

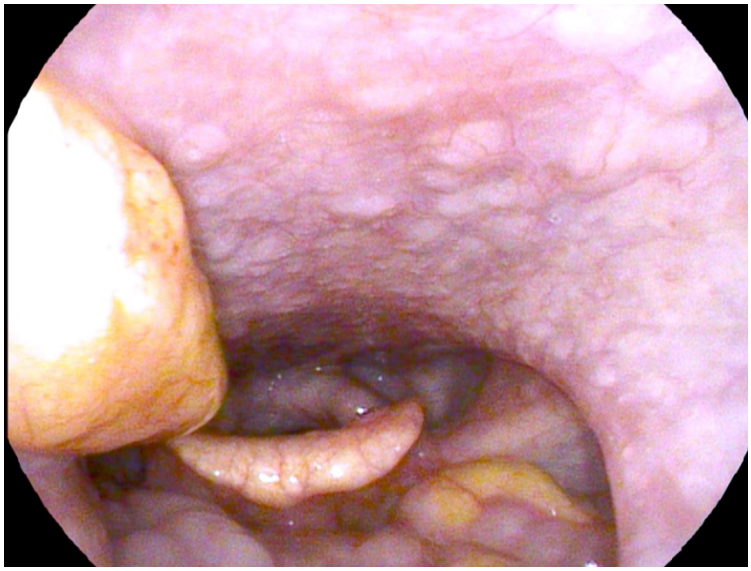

Figure 1: Preoperative endoscopic view showing globular, smooth, yellowish mass of about 2x2 cm in size that was firm in consistency, non-bleeding on touch on the right lateral pharyngeal wall. Similar lesion of about 1x0.5 cm in size was also observed over the left side of vallecula.

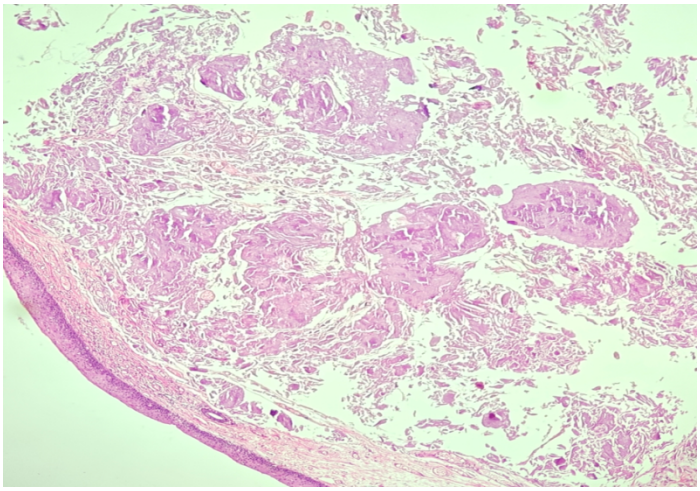

Figure 2a: H & E: Histopathology (X 100 magnification) showing amorphous extracellular eosinophilic material in the stroma.

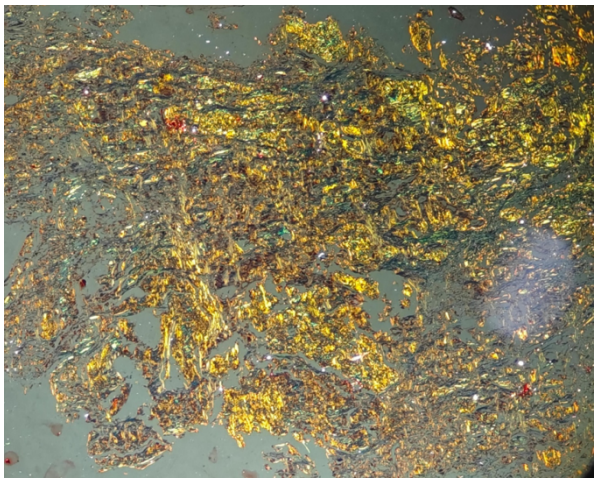

Figure 2b: Congo**:** Red Staining: Apple Green Birefringence Seen under Polarising Lens.

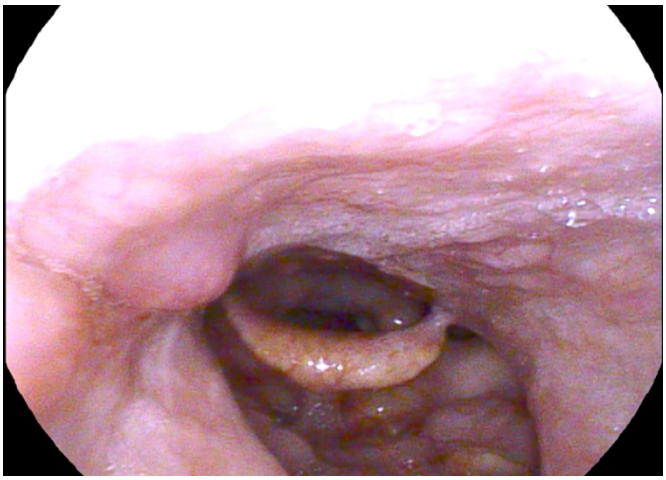

Figure 3: Postoperative endoscopic view showing well healed surgical site on right lateral pharyngeal wall with no evidence of recurrence.

Discussion

Amyloidosis in its different forms is a rare clinical entity which encompasses a spectrum of diseases with extracellular accumulation of protein fibrils in a unique and characteristic beta-pleated sheet polymer [7,8]. If there is limited amyloid deposition to the tissue where its precursor protein is produced, amyloidosis can be defined as localized or systemic if the precursor protein is produced in a tissue different from the site where amyloid protein is found Crosetti [8]. Amyloidosis in the head and neck involving the upper aerodigestive tract is usually localized, with exception of tongue involvement, which is associated with systemic involvement. It may involve nose, paranasal sinuses, orbit, nasopharynx, oropharynx, tonsils, oral cavity, larynx and tracheobronchial tree. Larynx is apparently the most frequent site for upper aerodigestive tract amyloidosis. Amyloidosis limited to pharynx is much rarer with most case reports on amyloidosis in nasopharynx, followed by about 14 case reports of amyloidosis in the oropharynx and the least reports on amyloidosis in hypopharynx [1,8-10].

The clinical manifestations are varied depending on the organ and system involved. The symptoms and presentation in case of localized amyloidosis depends on the size, physical presentation and location of the deposits of amyloid [1, 11]. Upper aerodigestive amyloidosis can present as tumour like nodule formation or diffuse subepithelial infiltrate. Nodule like presentation is more common in larynx and nasopharynx and subepithelial infiltration presentation predominates at other sites [6,7,12].

When suspected, biopsy and histopathology evaluation is the gold standard for diagnosis of amyloidosis. On Congo red staining and viewing under the polarized light, amyloid demonstrates characteristic apple-green birefringence which is pathognomonic of amyloidosis [7-9,13]. Thorough endoscopic evaluation of whole respiratory tract needs to be done to rule out multiple lesions as there is high incidence of multifocal lesion. Imaging studies can help to determine the extent of disease, but there are no characteristic findings on imaging and thus, diagnosis can only be made after biopsy [8].

With the diagnosis of upper aerodigestive tract amyloidosis, it is recommended to workup the patient with routine investigations to rule out systemic involvement. It is the initial step in the treatment of the disease as amyloid fibrils deposition destroys normal tissue structure and functioning of tissue. Systemic amyloidosis carries poor prognosis in comparison to localized amyloidosis [14].

In our case, the patient presented with complains of dysphagia and firm, smooth, yellowish swelling on right lateral pharyngeal wall, underwent direct panendoscopic examination with excision biopsy of the lesion and histopathology confirmation of amyloidosis on observance of apple-green birefringence after Congo-red staining. Patient did not have any other constitutional symptoms of fatigue, weight loss, neuropathy etc., and all routine investigations with chest X-Ray were normal. However, patient was referred to rheumatology to rule out further systemic amyloidosis and other diseases. Patient was followed up three monthly. There was no recurrence on follow up.

Management of upper aerodigestive tract amyloidosis mainly of surgical excision for localized amyloid deposits when possible without significant anatomical and functional sequelae. The basis of surgical removal is to provide symptomatic relief of dysphonia or difficulty in breathing, dysphagia [4,7]. The lesion can be removed endoscopically or by open technique either with cold-steel instruments or with laser. Due to its hemostatic capability, laser excision is considered favourable [7]. Different regimens of medical treatment including systemic corticosteroids and chemotherapy including alkylating agents (melphalan), dimethyl-sulfoxide, interferon-alpha, colchicine have been applied to oropharyngeal amyloidosis with largely unsuccessful results [7,15].

Radiation therapy regimens have been used with differing results and observation. The rationale behind the use of EBRT is the possible mechanism including elimination of the clonal amyloidogenic plasma cells that produce the light chains and amyloid fibrils, modulation of the local blood supply and induction of an immune response against amyloid deposition [16] Plasma cells are radiosensitive. Response to radiotherapy has been demonstrated in local control of multiple myeloma and plasmocytoma [7,17]. Radiation therapy is considered effective for arresting progression of laryngeal amyloidosis [16]. EBRT has also shown successful results in treatment of localised tracheobronchial amyloidosis causing airway obstruction or postoperative therapy [17,18] Radiation dose used in treatment for laryngeal and tracheobronchial amyloidosis ranges from 20 to 45 Gy. Radiation dose is kept low in comparison to what is used for malignant diseases [16,18] Nasopharyngeal amyloidosis has also shown regression with intensity-modulated radiotherapy [19,20]. However, it did not show clinical benefits in case of macroglossia due to amyloidosis [17].

As, there is no standard treatment protocol for localised or else amyloidosis and as there are high chances of recurrence even after surgical excision, it is crucial that the patient be followed up at regular intervals. Clinical evaluation with endoscopic examination of the aerodigestive tract at intervals and whenever any symptoms develop is necessary [8,16].

Conclusion

Amyloidosis is a group of rare disorders; but it should be considered a possibility in smooth lesions of the upper aerodigestive tract. The definite diagnosis of amyloidosis could be done by histopathology evaluation with special stains. Management of each case of amyloidosis is decided on individual basis as there are variable clinical presentations of the disease and may vary from surgical excision to adjuvant chemotherapy and radiotherapy. All cases of amyloidosis need to be followed up to observe for any possible recurrence.

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