

CASE REPORT

DYSPHONIA DUE TO RARE METABOLIC CAUSE: PRIMARY LARYNGEAL AMYLOIDOSIS

Abhay Havle D¹, Ashok Purohit², Gautam Purohit³, Vishal Jain⁴, Swapna Shedge⁵

HOW TO CITE THIS ARTICLE:

Abhay Havle D, Ashok Purohit, Gautam Purohit, Vishal Jain, Swapna Shedge. "Dysphonia due to rare Metabolic Cause: Primary Laryngeal Amyloidosis". Journal of Evolution of Medical and Dental Sciences 2015; Vol. 4, Issue 01, January 01; Page: 130-134, DOI: 10.14260/jemds/2015/21

ABSTRACT: Primary localized laryngeal amyloidosis is an extremely rare condition of unknown etiology. It usually presents with hoarseness of voice. Dysphagia is seen in some cases. We present the case of a 28-year-old woman with primary localized laryngeal amyloidosis who presented with hoarseness of voice. There were no signs of any systemic disease in our patient and diagnosis was established histopathologically. She was treated surgically by micro laryngoscopy under general anesthesia and the mass was excised using a CO2 laser technology method.

KEYWORDS: Congo red stain, CO2 Gas laser, Hoarseness, Laryngeal Diseases, Primary amyloidosis.

INTRODUCTION: Amyloidosis is a benign, slowly progressive condition characterized by the presence of extracellular fibrillar proteins in a variety of organs and tissues. Localized deposition of amyloid may be observed in individual organs without any systemic involvement. The progressive accumulation of amyloid deposits interferes with the normal structure of affected tissues resulting eventually in impairment of their function.

Primary laryngeal amyloidosis is a rare lesion representing 0.2 to 1.2 % of all benign laryngeal tumors.¹ Amyloidosis was initially described by Rokitansky in 1842. The first laryngeal amyloidosis case was documented by Borow in 1873.²

Immunohistochemical stains, and Congo red staining viewed under polarized light microscopy, or electron microscopic findings of a laryngeal biopsy specimen can confirm the presence of amyloid in the larynx.

A high degree of suspicion is necessary since clinical presentation of the disease may mimic that of a laryngeal neoplasm.

CASE REPORT: 28 years old Asian lady from India presented with history of hoarseness of voice since 6 months. She reported no weight loss and she denied the use of tobacco/mishri/alcohol consumption. She was taking T. carbamazepine for 5 years for epilepsy.

Physical examination of the neck was normal. Indirect and direct laryngoscopy revealed a smooth, red-yellow, cystic mass arising from left false vocal cord. (Fig. 1)

Chest X-ray was normal. MRI neck demonstrated the extent of the lesion. (Fig. 2)

The thyroid cartilage appeared intact and no nodal involvement was detected.

We proceeded to perform microlaryngoscopy under general anaesthesia with punch biopsy of the lesion. On macroscopic inspection, there were few irregular gray white soft to firm bits of tissue largest measuring 0.4*0.3*0.1 cm. On microscopy, section showed fragments of tissue lined by respiratory epithelium. Sub epithelial tissue showed extensive deposits of eosinophilic acellular hyaline material. Also noted was focal dense infiltration by lymphocytes and plasma cells. (Fig. 3) 20% ZN staining revealed no acid fast bacilli.

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Staining with Congo red stain, under polarized light, revealed apple green birefringence throughout the mass due to the presence of amyloid. (Fig. 4) No signs of malignancy were seen.

Further examinations were done to rule out systemic amyloidosis. Further examinations were done to rule out systemic amyloidosis. The patient's complete blood count, erythrocyte sedimentation rate, basic metabolic and biochemical panel and liver function tests were within normal limits. Serum calcium was also normal. Serum and urine electrophoresis were normal.

Histopathological examination of subcutaneous fat was negative for amyloidosis. Based on these findings systemic amyloidosis and multiple myeloma were excluded from the differential diagnosis.

The mass was removed by CO2 LASER excision by microlaryngoscopy. (Fig. 5)

Patient was on regular follow up. The voice improved after the surgical excision of the mass.

DISCUSSION: Amyloidosis is a metabolic protein disturbance in which extracellular protein fibrils are deposited in various organs and tissues. Amyloid deposits are extracellular and not metabolized by the body, thus progressive deposits will eventually impair the function of the organ where they accumulate.³

The amyloidosis is classified as systemic or local as shown in table-1.

A. Systemic amyloidosis;	
i. Hereditary amyloidosis (e.g., amyloidosis in familial Mediterranean fever)	AA, AF
ii. Idiopathic systemic amyloidosis.	AL
iii. Secondary systemic amyloidosis (reactive amyloidosis) and chronic infections, neoplastic diseases.	AA
B. Localized amyloidosis	AL, AA, AK

Table 1: Classification of amyloidosis

AA - Serum amyloid associated protein, AF- Familial amyloidosis, AL- Amyloid light chain, AK- Amyloid keratoepithelin.

Amyloid deposits rarely show remission but usually tend to increase in size at a slow but progressive rate. In the past, the extracellular deposition of amyloid in the larynx was commonly misdiagnosed as vocal cord nodules. Deposits of amyloid in the larynx are rare, accounting for between 0.2 and 1.2% of benign tumors of the larynx.¹ Thus the high degree of suspicion is needed in the diagnosis.

The ventricles and the false and true vocal cords are the most common sites for localized amyloidosis in the respiratory tree.⁴ Other sites are the eye, the orbits and the major and minor salivary glands, while sub mucosal deposits have been observed in the nose, paranasal sinuses, nasopharynx, oral cavity, oropharynx, tracheobroncheal tree and lungs.⁵ Oral and paranasal amyloidosis is usually a manifestation of systematic amyloidosis, mainly plasma cell dyscrasia.⁶ Laryngeal amyloidosis usually appears during the fifth and/or sixth decade of life, without specific symptoms, though it most frequently involves hoarseness of the voice.¹ Dyspnoea is a common manifestation of the disease. Dysphagia is rare in laryngeal amyloidosis.

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There are indications that immunological mechanisms are involved in the pathogenesis of human amyloidosis and that the latter could be a complication in immunodeficient conditions. Factors that affect the human immune response, such as steroids, immunosuppressive and ionizing radiation, may accelerate the appearance of the disease. Some types of amyloid deposits may be the result of an immunocyte dyscrasia or mucosa associated lymphoid tissue neoplasms.⁷ Although systemic amyloidosis presents a poor prognosis due to accumulation of amyloid protein in a variety of vital organs impairing their structure and function, localized primary amyloidosis carries a much better prognosis.

Although immunophenotyping has become universally available, the "gold standard" for the diagnosis of Amyloid remains a tissue biopsy demonstrating characteristic apple green birefringence with Congo red stain under polarized light (The so called dichroism). This should be distinguished from pseudoamyloid, which is often found in vocal cord nodules. This is of fibrous consistency and represents an amorphous granular degeneration of collagen fibers with sparse disseminations between the fibroblasts.⁸ Potassium permanganate may be used for the discrimination of protein composition between type a protein (AA) which dissolves, and amyloid (AL) of light chain, which is resistant and appears in the sections. Laryngeal amyloidosis is a type of localized amyloidosis that is characterized by monoclonal deposits of the light chain type (AL).⁹

Magnetic resonance imaging (MRI) is the technique of choice to detect the most specific features, since Amyloid deposits present an intermediate T1-weighted signal intensity and low T2-weighted signal intensity, and MRI is thus considered to be a more specific technique than CT scanning.¹⁰ Unfortunately, in our case, it was not possible to conduct an MRI scan due to technical reasons.

The treatment of choice of primary localized laryngeal amyloidosis is endoscopic CO2 LASER excision of the lesion. The course of the condition under discussion is slow but sudden relapse is possible¹¹. However, relapse may occur after a long time period; long-term follow up is essential for at least 5 to 7 years.¹ In our case, evolution into the systemic form of the disease was not observed in a 6-month follow-up; there is no disease recurrence and the patient is free of symptoms in this time period and is in a good state of health.

In the study by Biewend et al¹ in a mean 7.6-year follow up of two patients no recurrence was observed, while in another study by Piazza et al, 17 out of 32 patients were asymptomatic in a 20-year follow up. In secondary amyloidosis, a reduction in amyloid deposition may occur following successful treatment of the underlying disease.

CONCLUSION: We present here a rare case of dysphonia secondary to primary laryngeal amyloidosis. The diagnosis of the disease was established histopathologically using Congo red stain and polarizing microscopy of the laryngeal lesion obtained at complete surgical excision by microlaryngoscopy and CO2 laser. During one year post-operative follow up there was no recurrence of the lesion at the local site as well as systemically. Relapse may occur after a long time period; long-term follow up is essential for at least 5 to 7 years.

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Fig. 1: Smooth reddish yellow cystic mass arising from left false vocal cord



Fig. 2: CT scan at the level of supraglottis

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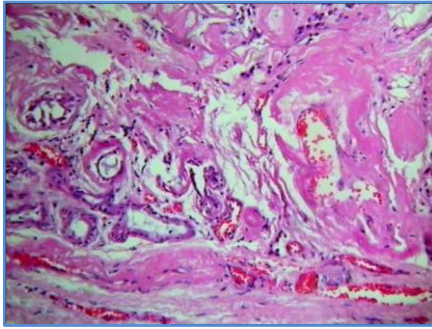


Fig. 3: H & E staining showing acellular, amorphous, eosinophilic material

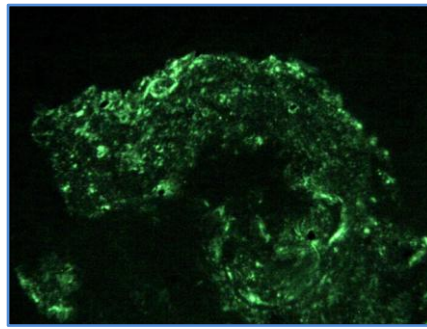


Fig. 4: Amyloid deposits seen on Congo red staining-Apple green birefringence

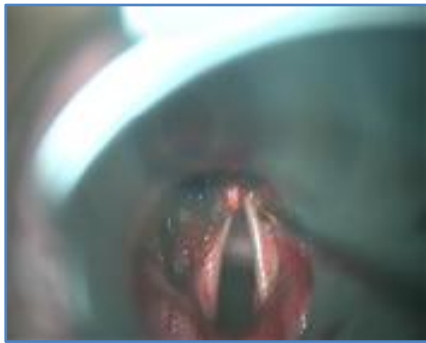


Fig. 5: Mass being excised using CO2 LASER



Fig. 6: Post-operative after 3 months LASER

AUTHORS:

1. Abhay Havle D.
2. Ashok Purohit
3. Gautam Purohit
4. Vishal Jain
5. Swapna Shedge

PARTICULARS OF CONTRIBUTORS:

1. Professor & HOD, Department of ENT, KIMSU, Karad.
2. Associate Professor, Department of ENT, BVDU, Sangli.
3. Resident, Department of ENT, KIMSU, Karad.
4. Resident, Department of ENT, KIMSU, Karad.

5. Assistant Professor, Department of Anatomy, KIMSU, Karad.

NAME ADDRESS EMAIL ID OF THE CORRESPONDING AUTHOR:

Dr. Abhay Havle D,
Professor,
Department of ENT,
KIMSU, Karad-415110.
E-mail: entpubkimsu@gmail.com

Date of Submission: 03/12/2014.

Date of Peer Review: 04/12/2014.

Date of Acceptance: 23/12/2014.

Date of Publishing: 01/01/2015.