

CASE REPORT**A RARE CASE OF NEUROFIBROMATOSIS WITH SQUAMOUS CELL CARCINOMA OF EPIGLOTTIS****Authors:** Harish Vel V (1) Swetha Lakshmi M (1) Jayendiran (2) Srinivasa V (3)**Authors Affiliations:** (1) Post graduates (2) Senior Resident (3) Head of the department; Vinayaka Mission Medical College, Karaikal**ABSTRACT**

INTRODUCTION: The neurofibroma is a nonmalignant new growth of neuroectodermal origin. neurofibromas appear at the end of a nerve, often in the skin, producing small nonencapsulated nodules which may have pigmentation in the overlying skin. Neurofibromatosis with laryngeal involvement presents with dyspnea, followed by hoarseness, stridor, dysphagia, and voice change. most common sites involved in the larynx are the arytenoids and the aryepiglottic folds.

CASE REPORT:

65-year-old female with previously diagnosed of neurofibromatosis since 10 years of age manifested by multiple cutaneous nodules. The patient had symptoms of dysphagia, hoarseness of voice for past 2 months. Direct laryngeal examination using flexible endoscope showed a edematous ulceroproliferative growth in the laryngeal surface of epiglottis and fullness in the left pyriform fossa. Histopathological examination revealed squamous cell carcinoma from epiglottis and fibromatosis changes from pyriform fossa.

CONCLUSION: All neurofibromatosis patient and their family members should undergo regular oral and laryngeal examination to rule out complications at the earliest.

INTRODUCTION

Neurofibromatosis in larynx are extremely rare, especially in the subglottic part. Characteristic symptoms of laryngeal tumors include hoarseness, dysphagia, odynophagia, dysarthria, a globus sensation, dyspnea and shortness of breath¹. Majority of tumors arising in NF1 patients are neurofibromas, particularly plexiform neurofibromas. Malignant peripheral nerve sheath tumors also affect these patients. Furthermore, patients with NF1 have a greatly increased risk of developing gliomas, leukemia, particularly juvenile myelomonocytic leukemia, pheochromocytoma and rhabdomyosarcoma^{2,3}. In addition, certain types of

carcinomas, including breast cancer, may also occur more frequently in patients with NF1^{2,4,5}. Occurrence of cutaneous squamous cell carcinoma (SCC) in patients with NF1 has been rarely documented⁶. The present study describes a case of laryngeal squamous cell carcinoma (SCC).

CASE REPORT

A 65-year-old female with previously diagnosed of neurofibromatosis since 10 years of age manifested by multiple cutaneous nodules. The patient had symptoms of dysphagia, hoarseness of voice for past 2 months. She is a known case of pulmonary tuberculosis 20 years ago for which she took full treatment course. No history of any addiction. Her physical examination was remarkable for multiple café au lait spots and subcutaneous nodules. FIG 1.0

She also has a family history of neurofibromatosis in her mother and her sister and her son. Indirect laryngoscopic examination demonstrated an enlarged epiglottis with lesions on its laryngeal surface with left pyriform fossa fullness. Direct laryngeal examination using flexible endoscope showed an edematous ulceroproliferative growth in the laryngeal surface of epiglottis and fullness in the left pyriform fossa. Fig 1.1, 1.2, 1.3

FIG 1.4 X-ray of neck soft tissue lateral view

To confirm diagnosis patient was posted for laryngeal biopsy under general anaesthesia. Multiple biopsies from epiglottis and left pyriform fossa were taken and sent for histopathological examination. The histopathological examination revealed squamous cell carcinoma from epiglottis and fibromatosis changes from pyriform fossa. FIG 1.5, 1.6

Fig: 1.0 Neurofibromatosis Patient

Fig: 1.1 Videolaryngoscopy



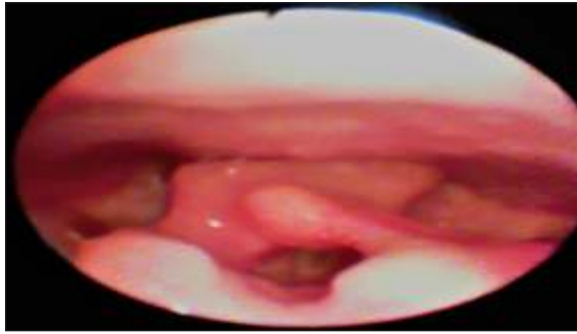


FIG:1.3 Fullness in the Left Pyriform Fossa

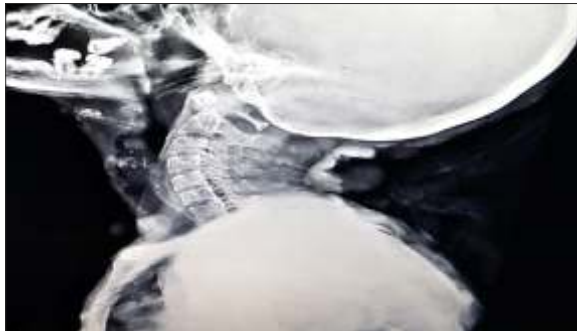


FIG 1.4 Xray of neck soft tissue lateral

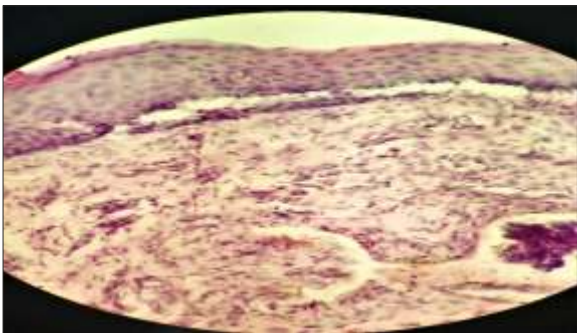


FIG 1.5 Hisopathological examination revealed squamous cell carcinoma from epiglottis and fibromatosis changes from pyriform fossa

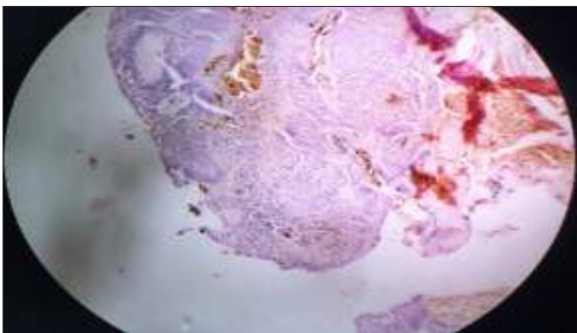


FIG 1.6 Hisopathological examination revealed squamous cell carcinoma from epiglottis and fibromatosis changes from pyriform fossa

DISCUSSION

Neurofibromatosis (NF) is an autosomal dominant disorder mainly characterized by abnormalities in the skin and nervous system. Neurofibromatosis are of 8 subtypes but NF1 and NF2 are the best described variants. NF1 is also known as *peripheral NF*.

National Institutes of Health diagnostic criteria for neurofibromatosis type 1

- Six or more cafe-au-lait macules (>5 mm diameter in children, >15 mm in adults)
- Two or more cutaneous or subcutaneous neurofibromas or one plexiform neurofibroma
- Axillary or inguinal freckling
- Optic-pathway glioma
- Two or more Lisch nodules
- Bone dysplasia
- First-degree relative with neurofibromatosis type 1

NF2 is known as *central NF* and is generally characterized by bilateral vestibular schwannomas presenting with hearing loss during the second or third decades()

Most common sites of laryngeal involvement include the aryepiglottic folds and the arytenoids, areas of the larynx rich in terminal nerve plexuses. It has been postulated that this location implies a tumor origin from the superior laryngeal nerve and from anastomoses between the superior laryngeal nerve and the recurrent laryngeal nerve.⁷⁻⁹

In 1849, Robert W. Smith first published a review of the disease and suggested that the origin of the tumors was the connective tissue surrounding small nerves¹⁰. In 1882, Friedrich von Recklinghausen recognized that the tumors included both neural and fibrous tissue derived from peripheral nerves¹¹.

A review article by Chang Lo in 1977 reported 20 cases of laryngeal involvement with neurofibromatosis. The most common presenting symptom is dyspnea, followed by hoarseness, stridor, dysphagia, and voice change. Half of the cases manifest in childhood and half in young adulthood.

This patient has generalized neurofibromatosis (von Recklinghausen's disease) for many years. Hoarseness and increasing dyspnea brought him to laryngeal examination. Histological examination revealed squamous cell carcinoma. Combined neurofibroma and squamous-cell carcinoma in the

larynx has been reported rarely. This case is considered worthy of documentation

CONCLUSION:

The neurofibromatosis patient should undergo regular oral and laryngeal examination and the family members of such patients also should undergo regular check up to rule out complications at the earliest

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