

# A case of rhinolaryngoscleroma in a patient with neurofibromatosis type 1

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## Introduction

Scleroma is a chronic-specific granuloma of the nose and it may involve other parts of the respiratory tract. The condition is endemic in Egypt. Neurofibromatosis type 1 (NF-1) is one of the most common genetic conditions of the nervous system. Before this report, no case had been published documenting scleroma in a patient with NF-1.

## Aim

To describe a case of rhinolaryngoscleroma in a patient with NF-1.

## Methods

Case report and literature review.

## Results

Presentation of clinical, imaging and pathological findings, as well as management considerations.

## Conclusion

This is the first case to be reported in the literature documenting the coexistence of rhinolaryngoscleroma and NF-1. The biological events in the immune system await further studies.

## Keywords:

genetic disease, granuloma, immunity, neurofibromatosis, scleroma

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## Introduction

Rhinoscleroma is a chronic-specific granuloma of the nose and it may involve other parts of the respiratory tract. The causative organism is *Klebsiella rhinoscleromatis*, a Gram-negative capsulated bacillus [1]. The disease is considered endemic in Egypt. It is a progressive disease involving initially a catarrhal/atrophic rhinitis, progressing to a proliferative granulomatous stage and finally to a fibrotic cicatricial stage with adverse effects [2].

Neurofibromatosis type 1 (NF-1), also known as von Recklinghausen's disease, is one of the most common genetic conditions of the nervous system, affecting ~1/3000 individuals worldwide. It is characterized by neurofibromas, café au lait macules, freckling, Lisch hamartomas in the iris, optic gliomas and bone deformities [3]. NF-1 is a progressive condition, in which the clinical manifestations appear at different times of life. Patients with NF-1 generally have limited life expectancy, mainly because of the development of malignant peripheral nerve sheath tumours in up to 10% of all cases [4].

In this report, a case of rhinolaryngoscleroma in a patient with NF-1 is presented. A PubMed search did not indicate any previous report of both conditions coexisting in the same patient.

## Case report

A 41-year-old woman presented to the outpatient department with nasal obstruction for 2 years, hoarseness

and increasing dyspnoea of 2 months' duration. The dyspnoea had progressed to the extent that she had respiratory distress at rest.

On examination, the patient had inspiratory stridor and hoarseness. There was widening of the nasal pyramid and multiple cutaneous nodules were evident over her face (Fig. 1). There were multiple café au lait macules over her trunk as well as axillary freckling. Examination of the nose indicated complete bilateral nasal obstruction by reddish masses. Endoscopic examination of the larynx indicated a 2 × 2 cm solitary swelling over the laryngeal surface of the epiglottis and purulent exudates (Fig. 2). A computed tomography (CT) scan of the nose indicated marked expansion of the nasal cavities by soft tissue swellings (Fig. 3). The CT scan of the neck and chest showed no involvement of the subglottis or trachea, and the lung fields were clear. A nasal smear was sent for bacteriological studies. The culture indicated the growth of *K. rhinoscleromatis*, sensitive to ciprofloxacin.

The patient underwent surgery under general anaesthesia with orotracheal intubation. The epiglottic mass was excised from its base by microlaryngoscopy and diathermy was applied to the raw area. A throat pack was applied and the nasal masses were removed completely with endoscopic guidance. Histopathological examination of all the laryngeal and nasal specimens indicated a submucosal inflammatory infiltrate composed of lymphocytes, plasma cells, Mikulicz cells and scattered Russell bodies (Fig. 4).

**Figure 1**



Photograph of the patient showing widening of the nasal dorsum and multiple nodules on the face (taken after obtaining the permission of the patient).

**Figure 2**



Endoscopic view of the larynx showing lesion on the laryngeal surface of the epiglottis and exudates.

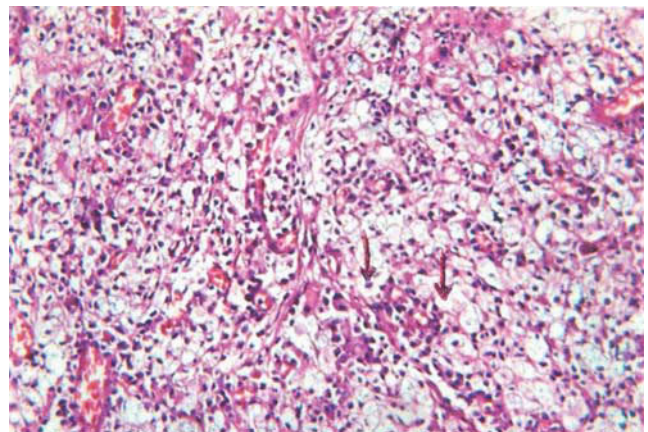
A Ziehl–Neelsen 5% stain showed multiple bacilli within the Mikulicz cells, confirming the diagnosis as rhinolaryngoscleroma in the cellular phase.

**Figure 3**



Coronal computed tomography scan showing expansion of the nasal cavities by the nasal masses.

**Figure 4**



Histopathology of nasal lesion showing lymphocytes, plasma cells, Mikulicz and Russell bodies (H&E, × 150). Arrows point to two Mikulicz cells.

On further questioning of the patient on the cutaneous lesions, she mentioned that her father and aunt also had multiple skin nodules and pigmented patches. The patient was therefore affected by NF-1. The patient was prescribed ciprofloxacin 500 mg twice a day for 4 months. She was compliant with the follow-up appointments, during which endoscopies of the nose and larynx were performed. At the most recent follow-up, 10 months after surgery, the patient was symptom free and there was no evidence of recurrence of the scleroma.

### Discussion

Rhinoscleroma generally affects the nose in 100% of cases [1,2]. Expansion of the nasal cavities by the granuloma causes pressure atrophy of the bones forming the nasal skeleton, leading to widening of the nasal

pyramid (Figs 1 and 3) [1]. In NF-1, alterations in bone mineral metabolism are frequent, with reduced bone mass and abnormal bone remodelling [5]. In the context of rhinoscleroma, abnormal widening of the nasal cavities may be a conspicuous feature. Facial dysmorphisms, including a saddle and broad nose, are also recognized features in some NF-1 patients [6]. Although rhinoplasty has been described in the literature in the context of neurofibroma [7], there are no published reports of rhinoplasty in scleroma patients.

The reported incidence of laryngotracheal involvement in scleroma patients ranges from 15 to 80%. The typical site of laryngeal involvement is the glottic-subglottic region [8,9]. The epiglottic involvement of the granuloma seen in our patient is considered rare. The CT scan did not show subglottic or tracheal involvement. The stage of laryngeal scleroma appears to parallel the nasal disease [8].

NF-1 is a tumour predisposition syndrome, and NF-1 patients are prone to developing a variety of tumours during their life [10]. NF-1 is associated with mucosal neurofibromas. In the upper aerodigestive tract, these have been most commonly reported in the oral cavity [11]. However, there are reports of neurofibromas developing in the nasal cavity [12,13] and larynx [14]. In the obvious setting of NF-1 in our patient, the presence of a neoplastic process in the nose and/or the larynx had to be excluded. All the specimens removed from the patient were subjected to pathological analysis, and no evidence of a neoplastic process was found.

Studies on the cells and molecules of the immune system have been reported in both rhinoscleroma and NF-1 patients. In rhinoscleroma patients, the CD4/CD8 ratio is inverted, leading to inefficient function of the macrophages in killing the bacteria by phagocytosis [15], thereby contributing towards the chronicity of the infection [16]. A genetic predisposition to the development of the progressive clinical features of rhinoscleroma has been suggested [17]. In NF-1 patients, multiple molecular pathways and at least two cellular mechanisms in the immune system have been detected that contribute towards the development of tumours [18]. Whether there is an interaction between the molecules and cells of the immune system in the setting of scleroma and NF-1 in the same patient awaits further studies.

Granulomatous conditions reported in patients with NF-1 include leprosy [19] and central giant cell granuloma [20]. However, to the best of the author's knowledge, scleroma has not been reported previously in the literature in a patient with NF-1.

## Conclusion

To the best of the author's knowledge, the present report is the first to document rhinolaryngoscleroma in a patient with NF-1. An interesting field of study would be the study of immune cells and molecules in such cases.

## Summary

Rhinoscleroma is a chronic-specific granuloma, endemic in Egypt. NF-1 is one of the most common genetic conditions of the nervous system. A case is presented of rhinolaryngoscleroma in a patient affected by NF-1.

Such a case has not been reported previously in the literature.

## Acknowledgements

### Conflicts of interest

There are no conflicts of interest.

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