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CASE REPORT

A case of Type 1 Neurofibromatosis involving the external auditory canal

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Introduction: Type 1 Neurofibromatosis (NF1) is a rare pathology with heterogeneous clinical presentation and only a few cases involving the external auditory canal (EAC) have been reported so far (1-3).

Objective: To report a case of NF1 with EAC involvement and literature review

Discussion and Conclusions: Size and location of tumors in NF1 most influence presentation of clinical symptoms. Complete tumor resection is possible only in patients with small tumors. Patients with single neurofibromas of the head and neck can benefit from surgery in order to 1) exclude malignancy in a rapidly enlarging mass; 2) enhance cosmesis in those with disfiguring disease; 3) provide relief from neurogenic pain; 4) improve symptoms caused by compression.

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Introduction

Type 1 neurofibromatosis (NF1), or Von Recklinghausen's disease, is an autosomal dominant neurogenetic disorder. It is associated with significant morbidity and results in substantial loss of function as well as significant cosmetic problems. NF1 is characterized by cutaneous manifestations (such as café-au-lait spots and freckles) neurofibromas, skeletal deformities, somatic and endocrine abnormalities; also central nervous system tumours and disorders have been reported [1-6]. Neurofibromas may develop anywhere in the body, including cranial and peripheral nerves. The type of impairment associated with neurofibromas depends largely on the site of the lesion. Cutaneous lesions, especially in the head and neck, generally cause deformity, whereas lesions that affect deeper systems (e.g., the auditory and ocular systems) are more likely to cause functional impairment.

The incidence of NF1 is reported to be about 1:2500 new births; it affects equally all races and both sexes [2]. Estimates show that currently in Italy there are about 20.000 cases; worldwide, there are about 1.5 million cases of NF1 [3].

While neurofibromas of the head and neck are not uncommon, they rarely affect the EAC and the pinna, also causing functional impairment (hearing loss).

Surgical management can be performed for functional and cosmetic reasons. The goal of the surgical procedure is to excise the tumour, as completely as possible, while restoring or preserving the ear anatomy and functions.

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Case Report

A 64-year-old Caucasian male was admitted to our clinic with a growing, mobile, soft temporo-parietal mass involving the right ear canal. The patient received a NF1 diagnosis at age 12; it was the first case in his family.

His primary complaint was a recurrent right otitis externa; hearing loss and cosmetic deformity were also present. No other relevant disease were present in his past medical history.

On examination, the temporo-parietal mass was causing head deformity on the right side; the lesion was displacing the right ear causing a significant deformity of the pinna and of the EAC that resulted collapsed (Fig. 1). Diffuse cafe au lait spots of various sizes (average diameter of 0.5 cm) were present in different parts of the patient's body, especially on his left arm. He also had freckles in his axillary areas.

The tonal audiogram showed a moderate conductive hearing loss in the right ear and normal hearing on the left side.

Ophthalmologic and neurologic examination resulted normal.

CT scans revealed a mass on the right temporal and parietal region causing obstruction of the right EAC (Fig. 2).

Surgery (debulking of the mass via a retroauricolar approach) was performed for functional and cosmetic reasons and resulted in a good functional and cosmetic outcome. The goal of the surgical procedure was to excise partially the tumour while restoring/preserving the external ear anatomy and function.

The histological evaluation of the specimen identified it as a neurofibroma (Fig. 3).

Discussion and Conclusions

Neurofibromas are circumscribed but non encapsulated neoplasm of the nervous system. They can arise in all peripheral nerve elements, including Schwann's cells, neurons, fibroblasts, and perineural cells. They may occur in isolation or as part of von Recklinghausen's syndrome in combination with cafe au lait skin patches, Lisch's nodules, or freckles [1-8], as in the case presented.

Neurofibromas can be deforming, especially those developing in the head and neck including eyebrow regions, eyelids, nose, lips and ears. In these location, neurofibromas may affect the functions of the surrounding anatomic structures and also may cause concerns for aesthetic reasons [9].

Neurofibromas are tumours derived from Schwann cells, fibroblasts and the supporting cells known as perineural cells. They can grow anywhere in the body where there are nerve cells [10]. Neurofibromas are usually benign; malignant transformation has been reported to occur in 2 to 16% of cases [7].

Most tumours caused by Neurofibromatosis need no treatment. But tumours that are painful, disfiguring,

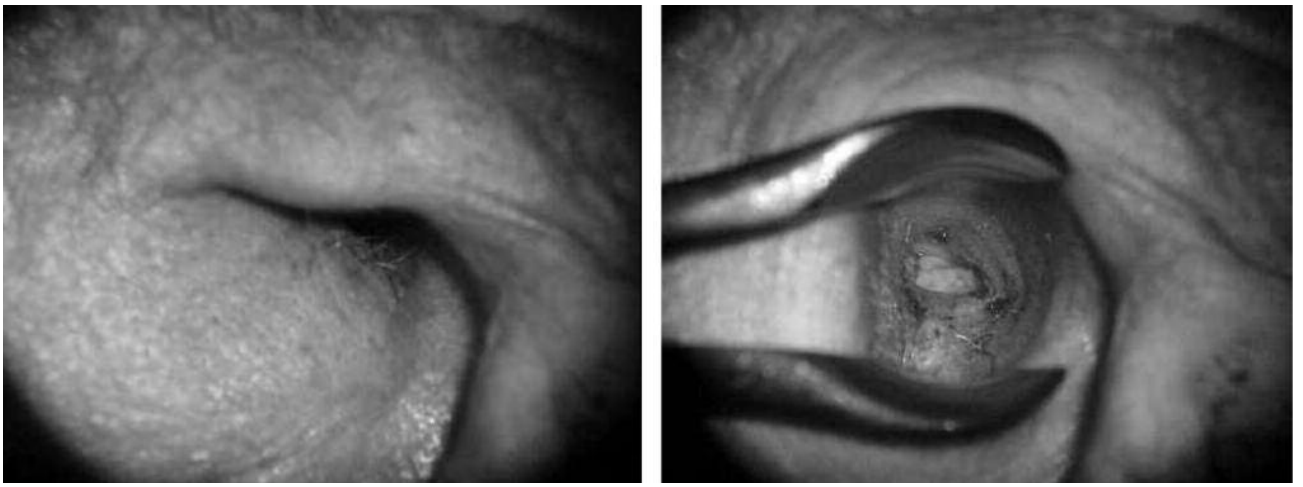

Figure 1. The mass was displacing the right ear causing a significant deformity of the pinna and of the EAC that resulted collapsed.

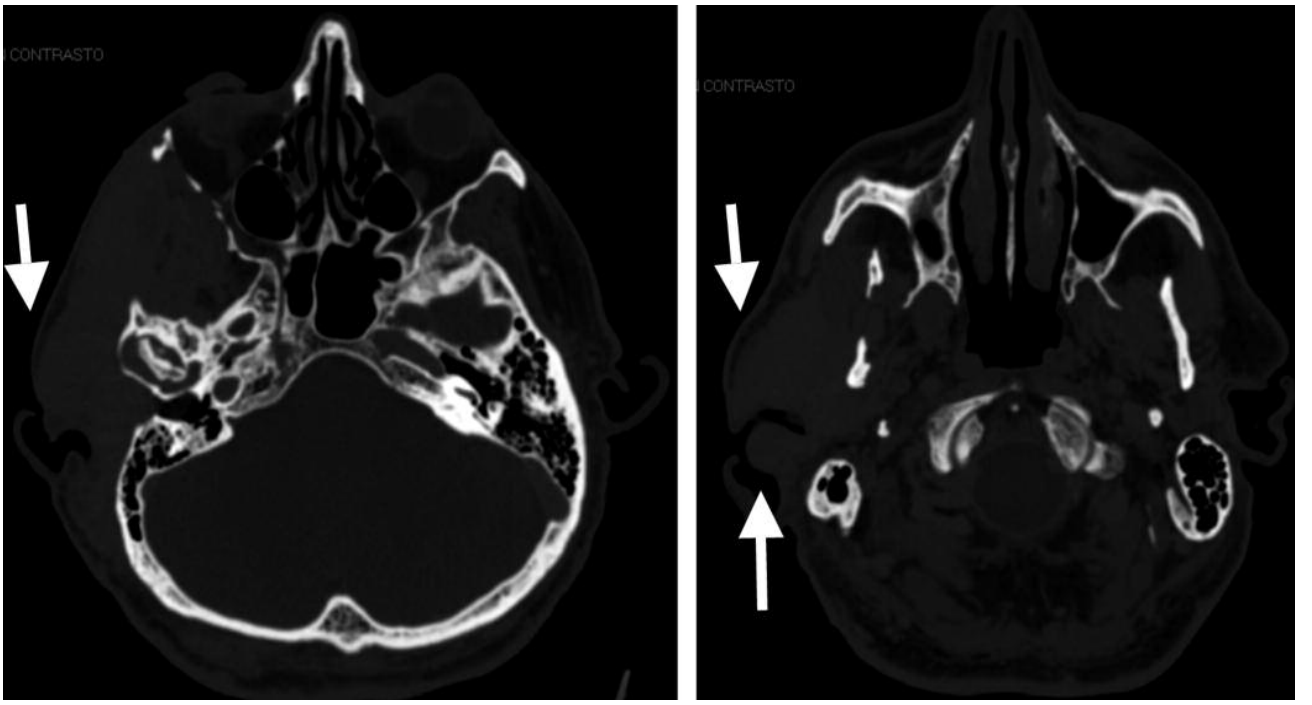

Figure 2. CT scans, axial sections: right temporal and parietal region mass causing obstruction of the right EAC (arrows).

growing rapidly, impairing function or compressing other body parts may need treatment [10]. The presented case demonstrates that neurofibroma involving the EAC can produce functional as well as cosmetic impairment, and that surgery can produce a satisfactory result.

Size and location of tumours in NF1 most influence presentation of clinical symptoms. It has been reported [9-10] that complete tumour resection is possible only in patients with small tumours. Patients with a single neurofibromas of the head and neck are more likely to benefit from surgery if the indications are to 1) exclude malignancy in a rapidly enlarging mass; 2) enhance cosmesis in those with disfiguring disease; 3) provide relief from neurogenic pain; 4) improve symptoms caused by compression [9-10].

Moreover, some Authors advise that [4,8] that subtotal resection of neurofibromas that cause deformity or a function loss is preferable to a complete excision; the latter procedure may result in a large loss of tissue and still not prevent a recurrence. Also, Trevisani et al [4] stated that "complete surgical excision of these lesions is virtually impossible, if not contraindicated," and Crikelair and Cosman [8] emphasized that amelioration rather than complete eradication should be therapeutic goal in these cases.

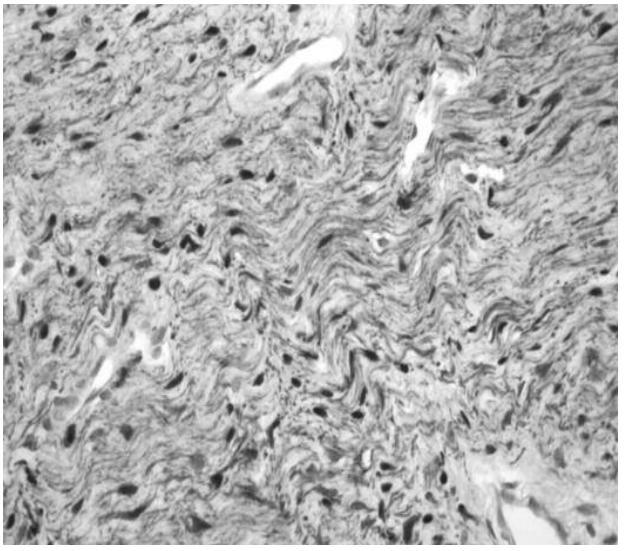

Figure 3. Microphotograph showing spindle shaped cells with fusiform twisted nuclei. Immunohistochemi-cal staining with S-100 protein antibody; 200X.

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