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Neurofibromatosis Type I Otological Manifestations: Case Report

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Abstract **Case Report**

Introduction: Neurofibromatosis type 1 is an autosomal dominant disease, classically caused by a single gene disorder. Its causative genetic defect is determined by a gene on chromosome 17 and is believed to manifest in neural crest cells, which in later stages contribute to the formation of the meninges, adrenal medulla, melanocytes, autonomic motor neurons and Schwann cells. [1] ***Clinical case***: We present a 14-year-old girl with a history of neurifibromatosis type I, with stenosis of the external auditory canal of the left ear secondary to previous neurosurgical interventions. On physical examination, the patient presented a soft, diffuse tumor that affected the left pre- and post-auricular regions, as well as the entire pinna, which showed an anterior-inferior displacement. Stenosis of the external auditory canal. Multiple brown spots were seen on the trunk, neck and extremities, as well as ephelides in the axillary region and multiple neurofibromas, most of them on the head and upper extremities. ***Discussion***: The treatment of neurofibromatosis type 1 must be individualized. It depends on the location of the tumor, whether it causes significant pain or disfigurement, whether it affects function, and its growth rate. Options range from observation, partial or total surgical resection, and the use of chemotherapy.

Keywords: Neurofibromatosis, Ear, Schwannomatosis.

INTRODUCTION

Phacomatosis (from the Greek phakos, meaning freckles) or neurocutaneous syndromes, comprise a wide spectrum of congenital anomalies of tissues derived from the ectoderm, some of hereditary origin; Among them we have tuberous sclerosis and neurofibromatosis, also Sturge -Weber disease and Von Hipple - Lindau disease, although these may have mesodermal origin [1]. Neurofibromatosis was fully described for the first time in 1882 by Friedrich Von Recklinghausen. German pathologist (Although Anthonius R. Smith, a Dutch surgeon, had already seen and described, although unofficially, these cases in 1849) Since that time it is clear that neurofibromatosis is a relatively common genetic disease, the form described on that occasion is the most common (type 1), followed by type 2, about 7 types have been described (others speak of up to 9) variants, but 1 and 2 occur most frequently, the others are so rare that their study is limited. It is also clear that a person with no history of the disease can have the disease, being caused by a genetic mutation [2].

Its variants are designated as type 1 (NF1) or peripheral, type 2 (NF2) or central, and schwannomatosis. Its distinctive feature is the development of benign tumors of the nerve envelope. Each of these disorders has particular clinical manifestations and molecular behavior. Its characteristics and nomenclature were established by the National. Institute of Health (NIH) of the United States. The transmission of these disorders is autosomal dominant, although 50% of cases are new mutations.1 Neurofibromatosis type 1 is the most common, has a worldwide distribution and a reported incidence of one in 2,200 to 4,000 live births, without racial or ethnic predilection [3].

Its particular lesion is neurofibroma, while schwannomas are characteristic of neurofibromatosis type 2 and schwannomatosis. It is difficult to establish the difference with the naked eye, but it can be achieved through a histopathological study. Unlike the other two disorders, neurofibromatosis type 1 also includes non-tumor manifestations, making it a true multisystem disease [4].

CLINICAL CASE

We present a 14-year-old girl with a history of neurifibromatosis type I, with stenosis of the external auditory canal of the left ear secondary to previous neurosurgical interventions. On physical examination, the patient presented a soft, diffuse tumor that affected the left pre- and post-auricular regions, as well as the entire pinna, which showed an anterior-inferior displacement. Stenosis of the external auditory canal. Multiple brown spots were seen on the trunk, neck and extremities, as well as ephelides in the axillary region and multiple neurofibromas, most of them on the head and upper extremities.

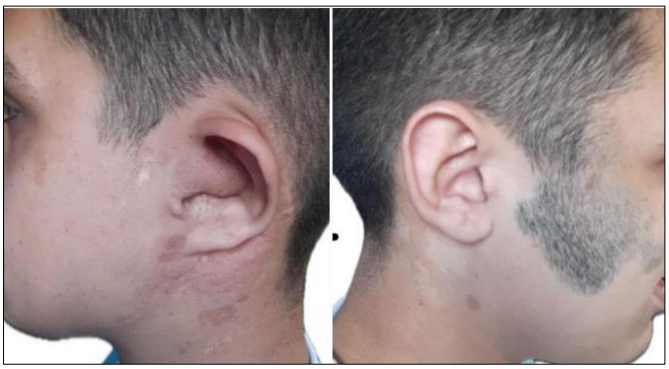

Figure 1: Left ear: complete pinna, which showed an anterior-inferior displacement, preauricular tumor, stenosis of the external auditory canal, brown spots on the trunk, neck and extremities.

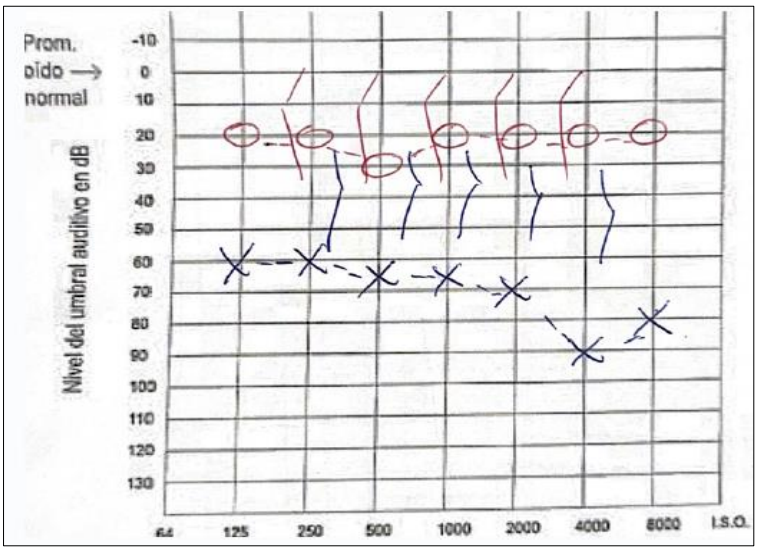

Figure 2: Tonal audiometry

Tonal audiometry shows: Normal hearing loss in the right ear and moderate to severe conductive hearing loss with a GAP of 25DB in the left ear.

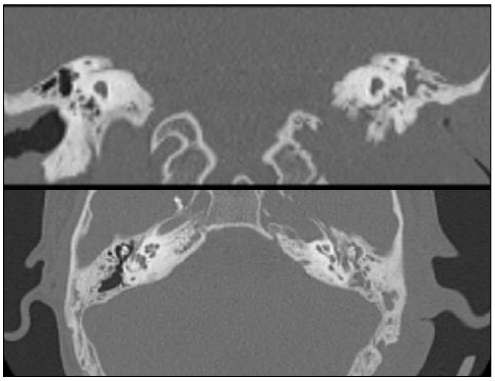

Figure 3: Computed tomography of the brain without contrast axial section

Computed tomography of the brain without contrast in axial section, a patent hypopneumatized mastoid is observed in the right ear, patent external auditory canal, apparently undamaged ossicular and ventilated middle ear, in the left ear, stenosis of the external auditory canal, hypopneumatized mastoid.

RESULTS

Because the patient was asymptomatic and this type of tumor does not have significant morbidity, it was decided to apply conservative treatment with periodic follow-up, with which there were no significant physical changes or progression of hearing loss.

DISCUSSION

Neurofibromatosis type 1 is an autosomal dominant disease, classically caused by a single gene disorder. Its causative genetic defect is determined by a gene on chromosome 17 and is believed to manifest in neural crest cells, which in later stages contribute to the formation of the meninges, adrenal medulla, melanocytes, autonomic motor neurons and Schwann cells [1-2].

The neurofibromatosis type 1 gene is thought to produce a protein similar to Ras GTPase-activating proteins, which in turn are involved in mechanisms that control cell growth and differentiation through interaction with the Ras gene family. This mutant form serves as a catalyst for tumor formation through altered signal transduction [3]

In a series of 434 patients with this condition, An incidence of otological manifestations of 6% was found, of which, the most frequently found was neurofibromas of the auricle (15 patients) (Table 2).

The manifestations of this disease are numerous in the head and neck. A total incidence is reported between 25 and 49%. 2-8 A relatively infrequent site of manifestation is the external ear [4].

Table 1: Otological manifestations of neurofibromatosis

external ear	
Neurofibromas	fifteen
Pavilion	fifteen
Low implementation	3
middle ear	
Neurofibromas	2
cerebellopontine angle	
Unilateral - bilateral acoustic neuroma	5
Neurofibrosarcomas	1

The most common thing is that the diagnosis is made during childhood, due to the existence of a family history of this disorder and obvious skin manifestations, such as light brown spots and neurofibromas. In general, skin lesions, particularly those mentioned above, are asymptomatic and do not represent danger [6].

On the other hand, neurofibromas can be cutaneous (soft, superficial), subcutaneous (causing pain and affecting the nerves) and even plexiform neurofibromas (nodular and diffuse), which cover all layers of the skin and are capable of eroding bone and invade the muscles or even the viscera, therefore, they can produce visible deformities and visceral damage, which being the head and neck region are of great importance [7].

The second division of the trigeminal nerve is the most common site of manifestation of plexiform neurofibromas of the face, however, they can appear in other cranial nerves. Regarding the eighth cranial nerve, neurofibromatosis type 1 is not related to the existence of neurinomas. The hearing loss in these patients is mainly caused by the obstruction of the external auditory canal due to a neurofibroma that, as it grows, compresses the cartilaginous portion and weakens its support [8].

The treatment of neurofibromatosis type 1 must be individualized. It depends on the location of the tumor, whether it causes significant pain or disfigurement, whether it affects function, and its growth rate. Options range from observation, partial or total surgical resection, and the use of chemotherapy [9].

There are several challenges in this regard. For example, the various manifestations of the disease may not result from the same pathological process and therefore will not respond to a single treatment. In addition, it has a greater chance of success if it starts early, preferably before symptoms appear. The last hope for these patients is that progress in understanding the pathogenesis will lead to new treatments. Regarding surgical resection, plexiform neurofibromas can be large and irregular in shape, making them unresectable. This hinders growth monitoring and makes it difficult to assess the effectiveness of the treatment [9-10].

In the present case, after surgical treatment, conservative management and periodic follow-up were decided because the neurofibroma did not cause significant symptoms or functional impairment. Another indication for surgery is malignant sarcomatous degeneration of a neurofibroma. To suspect malignancy, the increase in size of previously asymptomatic nodules; these can grow rapidly and metastasize with a wide distribution. Two other symptoms associated with malignant degeneration are pain or local neurological deficit.

Early diagnosis of malignant transformation is particularly difficult, due, among other factors, to the fact that these malignant tumors originate from preexisting benign lesions and patients with neurofibromatosis type

1 are accustomed to these lesions, which is why they do not seek medical attention.

Fluorodeoxyglucose positon emission tomography (FDG PET) has proven useful in distinguishing between benign and malignant lesions [10].

CONCLUSION

Neurofibromatosis type I is a complicated and misdiagnosed optical condition that requires multidisciplinary work. It can affect the outer ear, although lesions in this area generally have a good prognosis. A complete medical history remains the best diagnostic tool.

Conflicts of Interest: The authors declare that there is no conflict of interest with respect to the publication of this article.

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REFERENCES

1. Theos, A., & Korf, B. R. (2006). Pathophysiology of neurofibromatosis type 1. *Annals of internal medicine*, *144*(11), 842-849.
2. Lustig, L. R., & Jackler, R. K. (1996). Neurofibromatosis type I involving the external auditory canal. *Otolaryngology--Head and Neck Surgery*, *114*(2), 299-307.
3. Smullen, S., Willcox, T., Wetmore, R., & Zackai, E. (1994). Otologic manifestations of neurofibromatosis. *The Laryngoscope*, *104*(6), 663-665.
4. DA, S. (1988). Neurofibromatosis. Conference statement, National Institute of Health development conference. *Arch Neurol*, *45*, 575-578.
5. White, A. K., Smith, R. J., Robert Bigler, C., Brooke, W. F., & Schauer, P. R. (1986). Head and neck manifestations of neurofibromatosis. *The Laryngoscope*, *96*(7), 732-737.
6. Carrillo–Esper, R., Suárez, A., & Paredes–Fernández, M. (2003). Neurofibromatosis tipo 2. *Gaceta Médica Mexicana*, *138*(6), 623-624.
7. Cervera – Paz F., & Manrique, M. (2008). "Auditory implants of the brain stem." Address: www.ctf.org/espanol.neurofibromatosis.
8. Chusid J. (1983). Correlative Neuroanatomy and Functional Neurology. 6th. Ed. Modern Editorial Manual. Mexico City – Mexico. 341 – 396.
9. Cotran, R., Kumar, V., & Collins, T. (2000). Robbins: Structural and functional pathology. 6th. Ed. vol. 1. Mc Publishing. Graw Hill-Interamericana. Mexico City – Mexico. 173 – 175.
10. Falabella, R., Victoria, J., Berone, M., & Domínguez, L. (2002). Fundamentals of Medicine: Dermatology. 6th. Ed. *Editorial Corporation for Biological Research. Medellin Colombia*, *460* - 462-463.