



Neurofibromas in otorhinolaryngology. Literature review presentation of a rare clinical case of intramastoid solitary neurofibroma

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Abstract: Neurofibromas are precancerous neoplasms originating from the sheaths of peripheral nerves with a slow growth rate. A large proportion of cases (90%) are solitary tumors, while the rest (10%) are associated with the manifestation of neurofibromatosis type 1 (Morbus Recklinghausen) - an autosomal dominant genetic inherited disease. This type of tumor is relatively rare in the head and neck region. It involves mainly the skin and subcutaneous tissue of the trunk, and represents about 5% of all benign soft tissue neoplasms. In the available English articles 22 cases of intratemporal solitary neurofibroma have been reported of which only 2 were intramastoid. We present a rare clinical case of a solitary intramastoid neurofibroma in a 39-year-old woman mimicking the clinical representation of latent mastoiditis with a brief literature review.

Keywords: neurofibroma, solitary tumors, latent mastoiditis

Introduction

Neurofibromas are benign, slow-growing tumor formations, well-circumscribed and non-encapsulated, originating from non-myelinating type Schwann cells of the peripheral nervous system [1], which exhibit biallelic inactivation of the NF1 gene, encoding Neurofibromin – a GTPase-activating protein, which exhibits down-regulation of RAS/MARK signaling pathway activity, thereby losing tumor-suppressor function. Schwann cells hyperplasia, secrete chemotaxis factors such as KIT-ligands, chemoattractant stem cell factor (SCF), neurite growth-promoting factor 2 (NEGF2), which support the taxis of fibroblasts, perineural, endothelial cells and mast cells and the last secrete mitogens and NGF, influencing the microenvironment and tumorigenesis [2] [3] [4]. Most often, the development of this type of neoplasm is in 3–4 decades of life. There are 3 types of neurofibromas: localized/solitary, diffuse and plexiform [5]. A large proportion of these tumors are solitary (up to 90%), with a very low malignant potential [5]. Plexiform neurofibromas are associated with the development of Neurofibromatosis type 1 (NF1). Diagnostic criteria for NF1 are: ≥ 2 body neurofibromas or ≥ 1 plexiform neurofibroma, family burden, ≥ 6 café-au-lait spots, Lisch nodules (pigmented hamartomas of the iris), ≥ 2 armpit or inguinal freckles, optic nerve glioma

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and sphenoid dysplasia or skeletal abnormalities. While diagnostic criteria for NF2 are: bilateral acoustic neurinomas or family history of a first-degree relative and unilateral acoustic neurinoma or development of 2 of the following – meningiomas, gliomas, schwannomas, juvenile posterior subcapsular cataract [6]. The rate of local recurrence in solitary neurofibromas after surgical treatment is very low, provided the tumor is completely excised from the surrounding tissue [5]. In the clinical case described below, we find a highly atypical site for the development of a neurofibroma in the processus mastoideus, with no other diagnostic criteria for association with von Recklinghausen's disease.

The aim of the present study was to present rare clinical case of intramastoid solitary neurofibroma.

Materials and methods

We present a clinical case of a 39-year-old woman with proven isolated neurofibroma of the mastoid process. Clinical examinations such as otolaryngological status, anterior rhinoscopy, audiometry, computer tomography, tympanometry, as well as paraclinical and pathohistological examination were conducted to establish the diagnosis of neurofibroma of the mastoid process. Method used in the operative intervention: mastoidectomy with typical retroauricular access.

Results

The following examinations have been conducted subsequently.

Medical History: After a viral infection of GDP 3 weeks ago, there are complaints of fever, throbbing pain, fullness and pressure in the right ear, noise and hearing loss. She was treated on ambulatory care basis with amoxicillin/clavulanic acid, which proved to be ineffective. After reexamination by an ENT specialist, therapy with a macrolide antibiotic, nasal decongestant and ear drops were prescribed. Pain and noise were resolved, but inducing deafness and autophony persisted.

Patient reports of frequent childhood otitis media.

Otorhinolaryngological status: right ear - dull tympanic membrane, deformed light reflex, the area around the processus brevis – smoothed, manubrium mallei – not well defined. Slight palpatory tenderness when tapped on the processus mastoideus. Left ear normal.

Anterior rhinoscopy – nasal passages passable, pale pink nasal mucosa, no pathological secretion.

Other ENT organs – without pathological changes. Café-au-lait spots and tumor formations of the skin and mucous membranes on the trunkus and face are not detected.

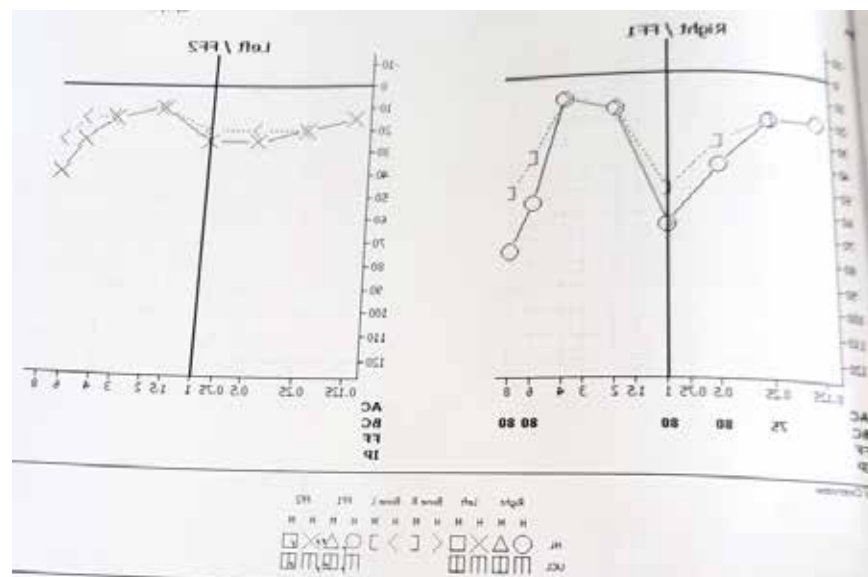


Figure №1.

Air and bone conduction audiogram of the patient



Investigations: Audiometry – sound-receiving hearing loss for the right ear with an average hearing loss of 33 dB, tympanometry – normal. Computer tomography – Mastoid cells on the left with normal pneumatization. Right mastoid – fluid-distended mastoid cells with destruction of bone partitions - density about 29–30 NE. Preserved lateral wall of the mastoid as well as the sigmoid plateau to the sigmoid sinus. In the middle ear, at the level of the tympanum, a linear thickening with a soft-tissue equivalent density is visualized – inflammatory changes /chronic otitis/.

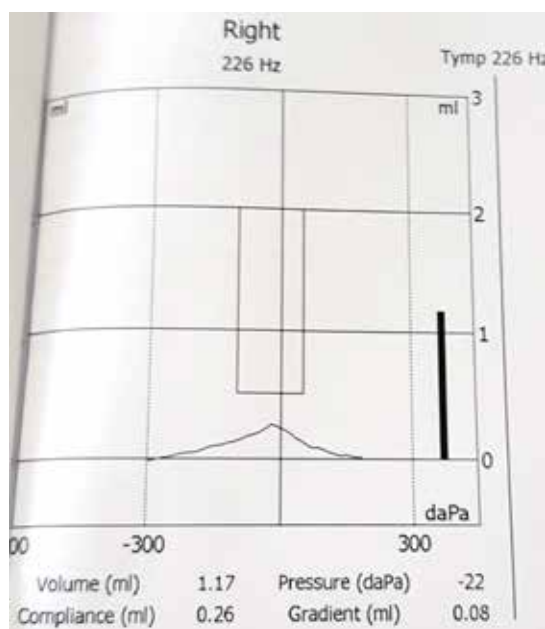


Figure №2.

The patient's tympanometry showed an As type tympanogram for the right ear

Paraclinic - in reference values.

Operative intervention: A mastoidectomy was performed under general anesthesia with a typical retroauricular approach. The mastoid process was trepanned, the antrum was opened, granulation tissue and a tumor formation of a white-like color with moderately dense consistency, filling the entire antrum mastoideum and contacting the dura matter at 2–3 mm, were removed. Due to nudation of the dura during excision of the tumor, a fascial flap was placed on the dura matter covered with gelspon. A ventilation tube was placed on the membrana tympani and a drain was placed in the trepanned processus mastoideus. There was a smooth and short postoperative period, without complications.

Pathohistological examination: histological picture of neurofibroma.

Discussion

Solitary neurofibromas are relatively rare tumors. They are most often found in the skin and subcutaneous tissues or along the course of peripheral nerves and represent about 5% of all benign soft tissue tumors. Although the head and neck region is a common location for these tumors, the development of neurofibroma in the mastoid process is extremely rare [7], [9]. The development of this



Figure №3.

Computed axial tomography of the head with evidence of otomastoiditis on the right; chronic pansinusitis

type of tumor is more likely to occur in the parotid gland, submandibular gland, tongue, gingiva and palate [7], [8]. In our available English literature, 22 cases of neurofibromas in the temporal bone have been described, of which only 2 were intramastoid [7], [9].

In terms of differential diagnosis, in patients with similar complaints and radiological findings, otomastoiditis, schwannoma of the nervus facialis, invasion of squamous cell carcinoma in the mastoid process or adenoid cystic carcinoma of the head and neck should be considered [7]. Any of these tumors could have a primary focus in the mastoid or invade it, causing destruction of the bony septa. Despite the similar localization of the processes listed above, it is possible to make a differential diagnosis based on anamnestic, clinical, and radiological data.

Otomastoiditis is a more likely diagnosis in young children. The presence of febrility, otalgia, otorrhea, redness and fall of the upper-posterior wall of the external auditory canal, as well as the changes in the retroauricular soft tissues – edema, pastiness and redness – lead us to the diagnosis. CT of the middle ear and mastoid process shows fluid in the tympanic cavity and ossification of the mastoid with or without signs of bone destruction.

Schwannoma of n. facialis KT and MRI images are identical as in neurofibroma in most cases, but some specific features of the images would help to differentiate it – more heterogeneous density with degenerations and cavitations, the visualization of a capsule around the tumor and multiple small ring-shaped hyperintense structures of T2-weighted MRI images are more expressive of a schwannoma diagnosis.

Squamous cell carcinoma could also present similar radiological images with destruction of bony septa, but physical examination may reveal skin lesions and lymphadenopathy [7].

According to a study by Chung-Ping Liao et al. 2018 chronic irritation and inflammation of nerve fibers combined with heterozygosity for the NF1 gene and the secretion of chemoattractant stem cell factor (SCF) underlies and subsequently sustains the tumor genesis of neurofibromas [10].

Conclusion

In the clinical case described by us the preceding latent mastoiditis left without adequate therapy and persisting for a prolonged time, may have triggered the development of the tumor. Unfortunately, the pathogenesis of neurofibromas still remains unclear.

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