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Solitary Plexiform Neurofibroma of the Parotid Facial Nerves in a Child with NF1: *Case Report and Review of Literature*

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Abstract **Case Report**

Plexiform neurofibromas (PNF) are benign tumors originating from peripheral nerve sheaths; generally associated with Neurofibromatosis Type 1 (NF1) usually involving a long segment of a major nerve trunk and extending into the nerve branches and they result in the so-called bag of worm's appearance. The MRI features of neurofibromas are characteristic and can be helpful in the evaluation of a mass in a patient with known NF1.Theyhave mostly a predilection towards the limbs, in contract less than a dozen cases in all literature report a juxta-parotidien PNF.

Keywords: Neurofibroma, NF1, MRI, bag of worms.

INTRODUCTION

Neurofibromatosis type 1, previously known as von RECKLINGHAUSEN disease, is a brain and skin condition in which tumours or swellings, called neurofibromas [2]. It is pertinent to add that NF1 belongs to the group of phacomatoses: including a multitude of syndromes involving developmental anomalies that occurred during the embryonic life. Their initial cause is the microdeletion of the NF1 gene located on chromosome 17 seems [6, 8]. Plexiform neurofibroma (PNF) is a rare benign tumor practically pathognomonic of type 1 neurofibromatosis. This prominent enlargement of a nerve with tumor nodules results in the gross pathologic appearance [10]. Plexiform neurofibromas of the salivary glands are rare (0.4% of all salivary neoplasms), often presenting in the parotid gland. They are very rare in submandibular salivary gland. Less than ten has been reported so far. We are highlighting this case because of the extreme rarity of its anatomical situation [11].

CASE REPORT

A 5 years old child presenting painless right cervical mass with tendency to grown. Neither facial paralysis nor sign of infection were described.

On clinical examination, a renitent mobile mass was noted in regard of the parotid gland. Further examination revealed café au lait spots spread predominantly in the back and the upper body.

The Ultrasound revealed the presence of an oval right parotidien mass, hypoechogenic, discreetly heterogeneous, within it, tubular formations which seem to be vascular-likestructurs.

A follow up MRI showed a lesion centered on the right parotid, well limited, with polylobed contours in hyposignal T1, heterogeneous in hypersignal T2 containing some serpiginous structures enhancing in a moderate way after injection of the gadolinium (figure 1), associated with few signal abnormalities at the level of the supra- and sub-tentorial area called unidentified bright object or 'UBOs' (figure 2).

The diagnosis of NF1 was initially retained due to the pathognomic imaging features found on the MRI and further confirmed via molecular genetic testing.

DISCUSSION

Plexiform Neurofibromas is one of the most common inherited genetic conditions,witha birth incidence of one in 1,900 to 2,800 and a diagnostic prevalence of one in 4,150 to 4,950 [3]. Topographycaly, of 300 neurofibromas in the minor salivary gland tumors, two were found to be plexiform neurofibromas. Furthermore, their predilection is astonishingly low in submandibular and sublingual salivary glands which are the case in our patient. In fact a wide review of literature showed that only five case of PNF in those glands have been describe until 2010[11].

The possible differential diagnosis for neurofibromas varie depending on the site of occurrence (intra vs. extraneural), including a number of neoplastic and non-neoplastic nerve lesions, such as schwannoma, nerve sheath myxoma, neurothekeoma, ganglioneuroma and traumatic neuroma, as well as a variety of non-nerve sheath tumors, in particular dermatofibrosarcoma protuberans and desmoplastic malignant melanoma. Among benign lesions, traumatic neuroma, a non-neoplastic proliferation at a site of nerve transection and ganglioneuroma, since neurofibroma may infiltrate dorsal root or sympathetic ganglia, represent the main entities in the differential diagnosis [12].

7 criteria were demarcated, of which 2 or more are indispensable to form the diagnosis of NF1[8], those 2 clinical criterias were in our case a plexiform neurofibroma hanged to the right jaw causing disfigurement , and a multitudes of Café-Au-Lait Macules (CALMs).

It has been also found that one of the child's medical antecedent that he has impaired learning skills and has suffered one episode of seizure at 3 years old. On that matter, epilepsy is known to be one of the neurological manifestations of neurofibromatosis type 1 (NF1), However, is not a constant feature [7]. The prevalence estimates of epilepsy in NF1 are extremely variable that is because of the variability in sampling and the multiple imprecise connotation that the word epilepsy.

Plexiform neurofibromas can extend superficially beneath the skin, or involve deeper tissues, and can affect the face, lower extremities, or spinal column [9]. It usually involving a long segment of a major nerve trunk and extending into the nerve branches and they result in the so-called bag of worm's appearance. Radiological evaluation with MRI can provide diagnostic information in 75% of cases making it the gold standard [2].

The gross pathologic appearance termed "bag of worms" consititutes the pathognomonic characteristic of plexiform neurofibromas (figure1).The second major feature is the occurrence of hyperintense lesions on T2-weighted MRI of the brain. These are located predominantly in the basal ganglia, brainstem, and cerebellum. They have been called "unidentified bright objects," "focal areas of signal intensity [FASI]," "NF spots," or "spongiform changes" because the true nature and significance of these lesions is still undetermined [1, 2] (figure 2).

Both of the previous mentioned radiological features are present in our case, in which the PN appear to be low signal intensity on T1 weighted images and heterogenous high signal intensity on T2 weighted images. The high T2 signal corresponds pathologically to areas of cystic degeneration or myxoid matrix and the low T2 signal represents collagen and fibrous tissue. The enhancement is variable: central, diffuse, peripheral or target [10].

Moreover, the MRI is best suited to look for, cerebellum hypoplasia, spheno-orbital dysplasia, spinal lesions such as duralectasia with meningocele and erosions of surrounding bony structures, peripheral nervous system neurofibromas and neurofibrosarcomas which further confirm the diagnosis of NF1 [6]. Yet, the main imaging features to safely retain the diagnosis of NF1 are the the bag of worms aspect and the unidentified bright objects both observes in this case.

Conventional X ray alongside withcomputed tomography have also a slightimpute in characterizing bone anomalies: vertebral scalloping and various forms of scoliosis.

Surgical excision is the most undertook course of action intraneural dissection is recommended for all schwannomas and many neurofibromas, although some degree of fascicular sacrifice may be necessary and the degree of excision should be tailored to the nerve involved. In the present case, the tumor was far from any major nerve and was well limited. Therefore, the mass was completely removed with no relevant complications [2].

Presurgical and postsurgical adjuvant therapy may include radiation and chemotherapy. The value of these discussions rested with the nuances of each case and the process by which surgeons elect to proceed with interventions.

It highlighted the lack of consensus in the overall approach ,very well understood since this tumors have no fixed location and therefore the surrounding anatomy varies along with it potential tendency for malignant behavior [5], surveillance is key to nuance and optimize the therapeutic approach and that includes Annual history and physical exam : including skin and neurologic exam, ophthalmic assessment, assess with history and clinical examination annually for typical signs of malignant peripheral nerve sheath tumors: any non-dermalneurofibromas with rapid growth, loss of neurologic function, or increasing pain or change in consistency should be enough to order further imaging and /or biopsies [3].

CONCLUSION

Neurofibromatosis type 1 (NF1) is a genetic condition that causes tumours to grow along the nerves. It's usually easy to diagnose NF1 in adults and older children by checking for the typical symptoms. The imaging modality in this topic is the MRI which confirms the diagnosis by looking for a plexiform neurofibroma renowned for its bag of worm like aspect

at the MRI and the presence of unidentified bright objects in multipleanatomical locations in the brain.

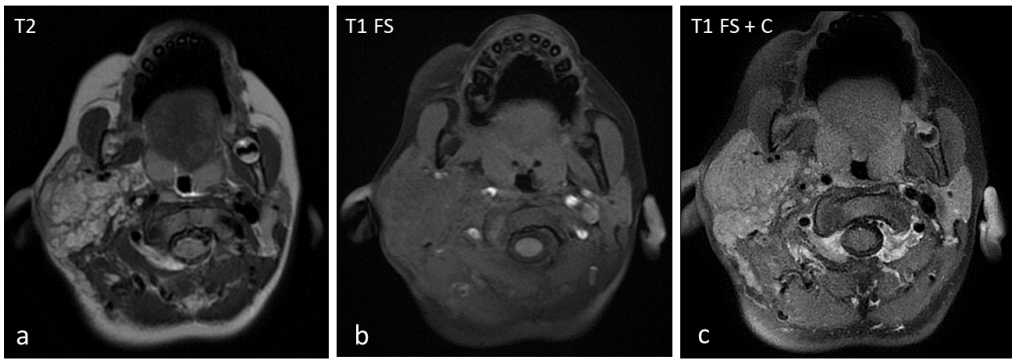

Fig-1: Cervical MRI images demonstrating right parotidplexiformneurofibroma: Axial T2 (a), T1 Fat Sat (b) and post contrast T1 Fat Sat (c) images showing a large right parotid gland mass, lobulated, well circumscribed, exhibiting an intermediate signal on T1 weighted images, heterogeneous high signal on T2 weighted images dottedwith multiple serpiginous structures which has been described as a " bag of worms ", moderatelyenhanced by contrast

Note the extension of the PN to the adjacent cervical soft tissues as well as to the parapharyngeal and carotidspaceswithoutvascular invasion.

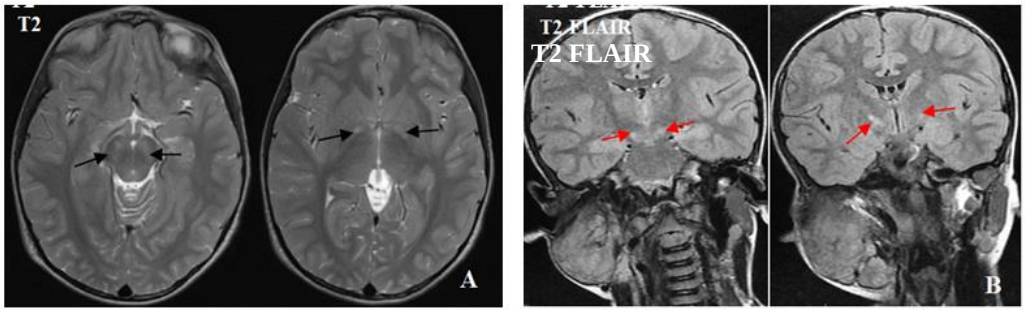

Fig-2: Cerebral MRI images showing signal abnormalities at the supra tentorial and sub tentorial level distributed asymmetrically and bilaterally involving the pallidum nuclei and midbrain in hyper signal T2 (A) (black arrows) and T2 flair (B) (red arrows), without restriction in diffusionnor enhancement after injection of Gadolinium

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