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Case Report

Schwannomatosis of the Posterior and Plantar Tibial Nerve. Case Report

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Abstract

Schwannomatosis is a rare form of neurofibromatosis that has been recently identified. The genetic disorder affects fewer than 1 in 40,000 people and causes the development of benign tumors, called Schwannomas, usually in the spinal and peripheral nerves. These tumors develop when Schwann cells, which form the insulating covering around nerve fibers, grow abnormally. These tumors can cause pain that is difficult to control, usually diagnosed in adulthood. We present a 36-year-old male patient with a health history, who came to the clinic complaining of pain and increased volume in the left leg and foot with cramp on palpation and difficulty walking. A multidisciplinary evaluation was carried out that allowed us to make the clinical, imaging and histological diagnosis of Schwannomatosis of the posterior tibial nerve and plantar nerve with favorable evolution and evident improvement of gait.

Keywords: Schwannomatosis; Posterior; Plantar; Tibial Nerve

Introduction

Schwannomatosis is a rare form of neurofibromatosis that has been recently identified. The genetic disorder affects fewer than 1 in 40,000 people [1-5] and causes the development of benign tumors, called schwannomas, usually in the spinal and peripheral nerves. These tumors develop when Schwann cells, which form the insulating covering around nerve fibers, grow abnormally. The development of the tumor appears to be primarily related to a change or mutation in certain genes that help regulate cell growth in the nervous system. These mutations prevent genes from producing the normal proteins that control cell proliferation, allowing cells to multiply excessively and form tumors. These tumors can cause pain that is difficult to control. Schwannomatosis is usually diagnosed in adulthood [1].

Signs of Schwannomatosis usually occur between the ages of 30 and 60, although they can occur at any age [1,2].

The term Schwannomatosis seems to date back to the 1950s, but other terms, such as Neurilemomomosis, have also been coined. The early literature confuses both Schwannomatosis and Neurilemomomatosis, terms used in Japan to include patients who clearly had NF2 with bilateral vestibular schwannomas. In the mid-1990s, a consensus began to develop that Schwannomatosis was distinct from NF2 [7,8].

Case

A 36-year-old male patient with a history of health presented with pain and multiple increases in volumes of different sizes on the inside of the leg and in the plantar area of the left foot with numbness, positive Tinel and difficulty walking due to pain.

Imaging results

Ultrasound(US) soft tissues in the posterointernal region of the left leg, ankle, inner face and sole of the foot where volume increases were observed, which corresponded to nodular lesions of various sizes, mostly solid, homogeneous with variable echogenicities, hypervascular and some with cystic degeneration of +- 3 x 2 cm in diameter, the largest in the path of the posterior tibial nerve.

A single CT scan of the left leg (Figure 1) showed multiple nodular thickenings in the posterior tibial nerve on its way to the plantar

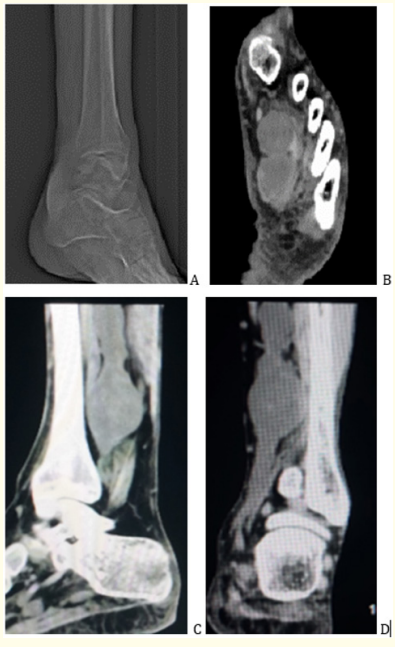

Figure 1: A Topogram showing osteoporosis of the ankle and increased volume and density of soft tissues in the internal retromalleolar region. B Presence of rounded tumors in the plantar region between 1st and 2nd metatarsals. C and D CT reconstructions show Schwannomas in the posterior tibial nerve pathway.

region of sizes and densities ranging from 6 to 38 UH with a compressive effect on adjacent musculotendinous structures.

Anatomical pathological findings

Macro

Several clear-brown, well-defined, soft tumor masses are received. The largest is 6cm in diameter, and the smallest is 1cm. When cut, the surface is light brown with a brown-orange center, the largest, and a soft consistency. The rest of the lesions have the same characteristics, where the central area is reduced in size.

Micro

A well-defined, encapsulated tumour lesion is observed, composed of two types of areas: hypercellular ones where the proliferation of narrow, wavy spindle cells with poor cytoplasmic delimitation and dense chromatin nuclei stands out (Antoni A) and others more lax, hypocellular, myxoid in appearance (Antoni B). In hypercellular areas, nuclear palisades are observed around fibrillar processes, called Verocay bodies.

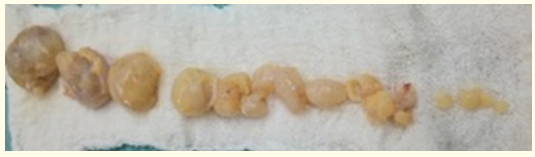

Figure 2

In the presence of these findings, we can say that we are dealing with a biphasic tumor, with benign behavior, called conventional Schwannoma.

In this patient there are several tumors with the same characteristics, all of which is why we can say that he has Schwannomatosis.

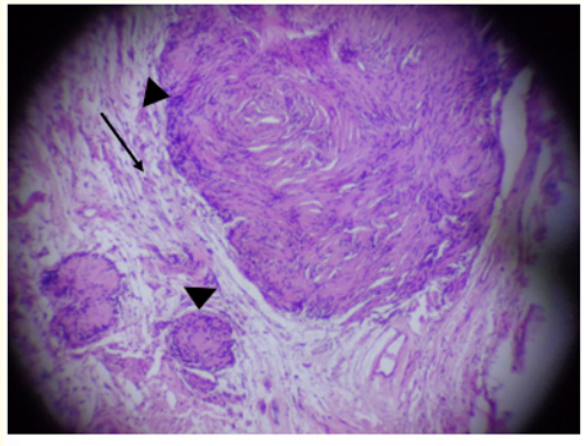

Figure 3

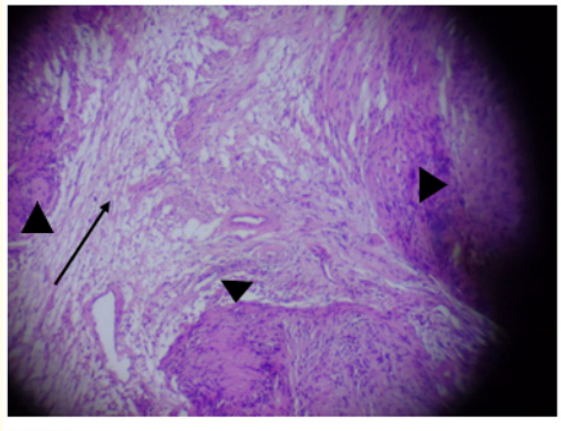

Figure 4

Figure 3 and 4: 4x panoramic views. Hypocellular areas (Antoni B) Arrows. Hypercellular areas (Antoni A) Arrowheads.

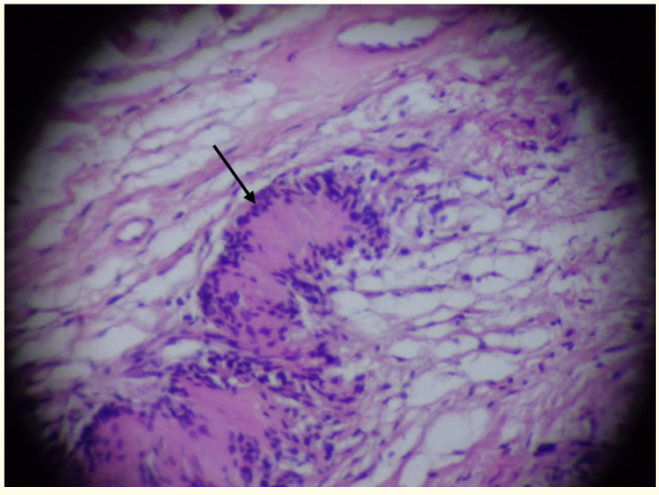

Figure 5: Body of Verocay. 10x.

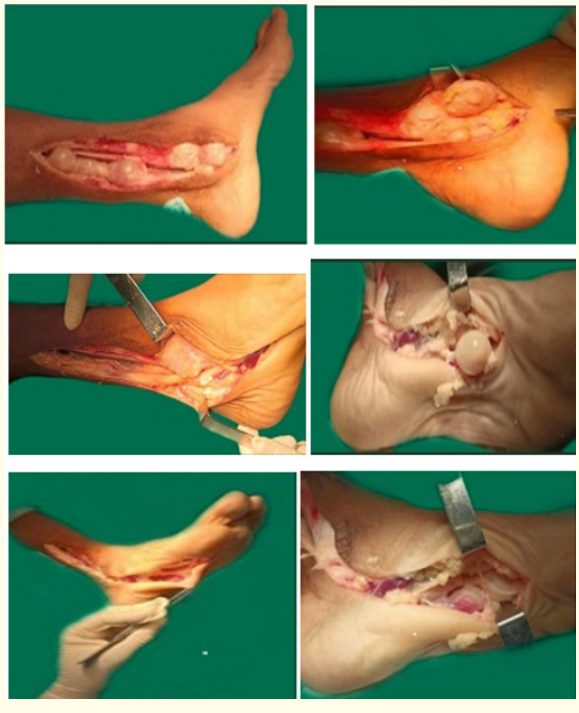

Figure 6

Discussion

Neurofibromatosis is a group of hereditary diseases that affect the bone, central and peripheral nervous system, soft tissues and skin. Up to 8 different types of Neurofibromatosis have been described, with three currently accepted: type 1 (NF-1) or Von Recklinghausen's disease; type 2 (NF-2) or central; and type 5 or segmental neurofibromatosis [2]. Schwannomatosis has been defined in recent years as a pathology different from NF-2 with a partial expression characterized by the appearance of nerve sheath tumors, without the vestibular schwannomas that are diagnostic of NF-2 [2,3,6,9].

In the 90s, Schwannomatosis and segmental neurofibromatosis were used interchangeably to refer to a pathology consisting of the appearance of multiple tumors limited to an anatomical region without crossing the midline and without presenting the rest of the characteristic features of Neurofibromatosis. Today they are considered different pathologies [2]. Approximately one-third of patients with Schwannomatosis have tumors confined to one limb, a specific spinal segment, or a hemibody [2].

Schwannoma, also called Neurilemmoma is the most common benign tumor affecting peripheral nerves [2,4,10], accounting for 5% of benign soft tissue neoplasms. It usually presents itself alone; Multiples are rare, associated with neurofibromatosis and affect one in 40,000 patients [4].

The majority of diagnosed schwannomatoses affect the head or trunk (45%), with less frequent cases documented in the literature locating in the upper limbs (3-19%) [2,11]. Signs of Schwannomatosis usually occur between the ages of 30 and 60 [1-3,12]. although they can occur at any age, [1] any gender and ethnicity [4,12]. The most common is chronic pain, thought to be caused, at least in part, by Schwannoma pressing on nerves, most people with Schwannomatosis experience some degree of pain that requires medical treatment, a small number of people experience only mild pain, although some people may also experience neurological symptoms. Many people with Schwannomatosis experience pain as the only symptom for several years before the source of the pain is identified [1]. They usually have a positive Tinel sign [2,11].

Schwannomatosis can be difficult to diagnose because symptoms vary widely among affected individuals, and many of the symptoms are shared by other disorders [1].

Criteria for a clinical diagnosis include one of the following: two or more non-intradermal Schwannoma, one pathologically confirmed, not including bilateral vestibular Schwannoma by high-quality MRI (a detailed study of the internal ear canal with slices no more than 3 mm thick). We recognize that some segmented patients with NF2 will be included in this diagnosis at an early age

and that some patients with Schwannomatosis have unilateral vestibular Schwannoma or multiple meningiomas [1,2].

It will be considered as a possible diagnosis if there are two or more non-intradermal tumors, but none have been pathologically proven as Schwannoma; the occurrence of chronic pain in association with the tumor(s) increases the likelihood of Schwannomatosis [1].

Differential diagnosis can be difficult, resulting in incorrect diagnosis in up to 75-80% of cases. Ganglion is the pathology with which it is most frequently confused, due to its cystic consistency. MRI can be useful to delimit the lesion, confirm the presence of other associated tumors and plan surgery, but it is ineffective in differentiating it from other pathologies such as neurofibroma or nerve sheath tumor, making it necessary to carry out an anatomopathological study to confirm its condition. Schwannomas are usually hypointense or isointense in T1 sequences, and hyperintense in T2 sequences [2].

In the case of our patient, we made the anatomopathological differential diagnosis Neurofibroma that lacks the Antoni A and B zones and are not encapsulated, Plexiform neurofibroma may be reminiscent of Schwannoma but present nerve grooves through the tumor mass and Dermatofibroma lacks the Antoni A and B zones, they are not encapsulated, nor well delimited and are formed by spindle cells and histiocytes [13].

Treatment options include observation or excision. Surgical treatment is indicated when compressive symptoms appear that make the patient's life difficult, [2] but it should be noted that the possibility of producing a neurological deficit during the intervention is estimated at 4%, with a higher risk in interventions performed after an incisional biopsy or after relapse, and in cases of multiple resections. To minimize neurological sequelae, microsurgical techniques are recommended, and an intracapsular resection of the tumor is recommended after making a longitudinal incision on the epineurium and capsule [2].

The case was submitted to a multidisciplinary evaluation that allowed us to make the clinical, imaging and histological diagnosis of Schwannomatosis of the posterior tibial nerve and plantar nerve with favorable evolution with evident improvement of gait by achieving pain control and negative Tinel sign.

Conclusion

- Schwannoma, also called Eurylemoma, is the most common benign tumor affecting the peripheral nerves.
- Most Schwannomatoses affect the head or trunk, with less frequent localization in the limbs.
- Schwannomatosis is a rare form of neurofibromatosis that affects less than 1 in 40,000 people.
- Signs of Schwannomatosis usually occur between the ages of 30 and 60, although they can occur at any age.

Bibliography

1. Association of People Affected by Fibromatosis. Madrid: Association of People Affected by Fibromatosis; [date unknown]. "Schwannomatosis" (2023): 5.

2. Bartolomé-García S., *et al.* "Bilateral multiple schwannomatosis: a case report". *Revista Española de Cirugía Ortopédica y Traumatología* 55.3 (2011): 17.

3. Children's Tumor Foundation. Have You Just Been Diagnosed with Schwannomatosis?" Basic Information. New York: Children's Tumor Foundation (2016): 20.

4. Montoya Cardero LE., *et al.* "Schwannomatosis of the foot". *MEDISAN* 17.6 (2013): 7.

5. MedlinePlus. Bethesda (USA): National Library of Medicine; Schwannomatosis (2017): 3.

6. Mayo Clinic. Rochester, Minnesota: Mayo Foundation for Medical Education and Research; c1998-2003". "Neurofibromatosis" (2021): 22.

7. Gareth Evans D., *et al.* "ERN GENTURIS clinical practice guidelines for the diagnosis, treatment, management and surveillance of people with schwannomatosis". *Nijmegen: ERN GENTURIS* (2022): 73.

8. Plotkin SR., *et al.* "Updated diagnostic criteria and nomenclature for neurofibromatosis type 2 and schwannomatosis: An international consensus recommendation". *Genetics in Medicine* 24.9 (2022): 52.

9. Columbia Neurosurgery [Internet]. New York: Columbia University Irving Medical Center, The Neurological Institute of New York; c2023. Schwannomatosis (2023): 7.

10. Mariano Cuello L., *et al.* "Schwannomatosis of the internal plantar nerve. Case presentation". *Rev Arget Neuroc* 33.4 (2019): 3.

11. Koontz NA., *et al.* "Schwannomatosis: The Overlooked Neurofibromatosis?" *American Journal of Roentgenology* 200.6 (2013): 39.

12. Alaidarous A., *et al.* "Segmental schwannomatosis: characteristics in 12 patients". *Orphanet Journal of Rare Diseases* 14 (207): 33.

13. Assoc Prof Patrck Emanuel. Dermatopathologist. DermNet. All about the skin. Auckland, New Zeland (2013).