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CASE PRESENTATION

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Tumor de vaina del nervio periférico en un paciente pediátrico, reporte de un caso

Peripheral Nerve Sheath Tumor in a Pediatric Patient: A Case Report

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Resumen

Introducción: Los sarcomas representan un conjunto de neoplasias de baja incidencia que se desarrollan en el tejido óseo, conectivo, adiposo y muscular, aquí se incluye el tumor maligno de la vaina del nervio periférico.

Objetivo: Describir el diagnóstico de un tumor de vaina del nervio periférico.

Presentación de caso: Preescolar masculino de cinco años, este presenta en neonatología un íctero fisiológico agravado, con seguimiento en ortopedia por presentar asimetría de ambos hemicuerpos. Tiene el antecedente de parotiditis, tratada con antibióticoterapia, de evolución tórpida, esta requiere ingreso. Al examen físico de cabeza y cuello se palpa un tumor poco movible, doloroso en la región submandibular izquierda. Se realizan estudios que permiten el diagnóstico de un tumor de vaina del nervio periférico. No es posible el tratamiento quirúrgico ni la radioterapia. Se indica quimioterapia paliativa.

Conclusiones: El tumor de vaina del nervio periférico, del tejido blando es raro y agresivo en pacientes pediátricos. La resonancia magnética es el examen de elección. El tratamiento utilizado es la quimioterapia.

Palabras clave: Neoplasias de la vaina del nervio, neurofibrosarcoma, pediatría

Abstract

Introduction: Sarcomas represent a group of low-incidence neoplasms that develop in bone, connective, adipose, and muscular tissues; this includes malignant peripheral nerve sheath tumors.

Objective: To describe the diagnosis of a peripheral nerve sheath tumor.

Case Presentation: A five-year-old male preschooler presented in the neonatal period with aggravated physiological jaundice and was followed up in orthopedics due to asymmetry of both body halves. He has a history of parotitis, treated with antibiotic therapy, which had a torpid evolution requiring hospitalization. Upon physical examination of the head and neck, a poorly movable, painful tumor is palpated in the left submandibular region. Studies are performed leading to the diagnosis of a peripheral nerve sheath tumor. Surgical treatment or radiotherapy is not possible. Palliative chemotherapy is indicated.

Conclusions: Peripheral nerve sheath tumors of soft tissue are rare and aggressive in pediatric patients. Magnetic resonance imaging is the diagnostic test of choice. The treatment used is chemotherapy.

Keywords: Nerve Sheath Neoplasms, Neurofibrosarcoma, Pediatrics

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Introduction

Sarcomas constitute a group of infrequent malignant neoplasms that develop in bone tissue and connective tissues, such as adipose and muscular tissue. In most cases, the etiology of sarcoma is idiopathic, although family history and exposure to radiation or certain chemical products are considered risk factors.[1]

Within this category lies the Malignant Peripheral Nerve Sheath Tumor (MPNST), a type of soft tissue sarcoma. Its pathogenesis begins when nerve sheath cells proliferate abnormally and form neoplasms derived from peripheral nerves, which show variable differentiation towards the cellular components of the sheath.[1]

MPNSTs are rare entities, with locally aggressive behavior, characterized by a clinical pattern of frequent recurrences and metastatic potential. The clinical presentation typically includes pain, the presence of a palpable mass, and functional deficit of the affected nerve. Their classification within sarcomas is based on their histogenic origin and biological behavior.[2]

First termed malignant neurilemoma in 1935, they are also known as malignant schwannoma, neurofibrosarcoma, and neurogenic sarcoma. The World Health Organization (WHO) introduced the term “malignant peripheral nerve sheath tumors” to avoid confusion with previous terminology.[3]

MPNSTs represent 5 % of all soft tissue sarcomas, with a general population incidence of 0.001 %. This frequency increases to up to 10 % in patients with Neurofibromatosis type 1 (NF1). Epidemiologically, half of the cases are sporadic, while the other half occurs in the context of NF1. Incidence is similar between both sexes, with a typical age of presentation ranging from 20 to 50 years.[3,4]

Following an exhaustive review of national and international biomedical databases, no reports or records of identical or similar cases documented in the pediatric age group were found in Cuba. Therefore, the case presented here constitutes the first report in the province of Pinar del Río. This lack of prior documentation underscores the rarity of this condition.

Its diagnosis and treatment are challenging due to the limited information available in the literature, as it is a rare entity in the pediatric population. This case holds significant educational value as it involves an MPNST in a pediatric patient without NF1, which further increases its uniqueness. This study aims to improve early diagnosis, guide treatment options, and contribute to the scientific knowledge about MPNST. It seeks to raise awareness within the medical community and the general public about this condition, facilitating timely detection and appropriate management of affected patients. The objective is to describe the diagnosis of a peripheral nerve sheath tumor.

Case presentation

A five-year-old male preschooler, Caucasian, product of a full-term vaginal delivery at 39.4 weeks, weighing 7.10 pounds. Personal pathological history includes left-sided hemihypertrophy; family pathological history includes maternal dysfunctional thrombopathy. The patient has no known drug allergies, previous surgeries, or history of blood transfusions.

The patient was admitted to the neonatology unit for aggravated physiological jaundice and has been followed up by orthopedics at the Frank País International Orthopedic Scientific Complex due to asymmetry of both body halves.

In May 2023, the patient presented with left-sided parotitis, with symptoms including pain, tenderness in the jaw region, fever, general malaise, and difficulty chewing and swallowing. Due to the severity of these symptoms and the risk of complications, the patient was admitted to the Pepe Portilla Provincial Pediatric Hospital. He received antibiotic therapy to prevent secondary bacterial infections (Amoxicillin at a dose of 80 mg/kg per day orally, administered in three divided doses, for 10 days).

The patient was discharged from the hospital, but the clinical condition worsened over the following days. In June, he was readmitted for further study and treatment.

Physical examination of the head and neck revealed a poorly mobile, painful tumor in the left submandibular region, with no cervical lymphadenopathy. The pediatric assessment triangle was examined, showing no impairment of its components.

Supplementary tests revealed elevated alkaline phosphatase (139 U/L) and lactate dehydro-

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genase (LDH) at the upper limit of normal (446 U/L).

Imaging studies were ordered to establish a clinical diagnosis and rule out other complications.

Magnetic Resonance Imaging

Coronal sequences of the neck and skull T1 and T2, axial T1 and Flair

An extensive occupying lesion, isointense to muscle on T1-weighted images, is observed from the periphery. It occupies the left parotid region, left masseter, and pterygoid muscles. The lesion obliterates the nasopharynx and obstructs and displaces the upper airway. It extends into the carotid, jugular, sternocleidomastoid, and paravertebral compartments. It measures 62 x 58 x 52 mm.

The lesion appears hyperintense on T2-weighted images with central, asymmetric, isointense areas due to solid or proteinaceous content, presenting a multilocular appearance. Partial opacification of the left mastoid air cells is noted, figures 1 and 2.

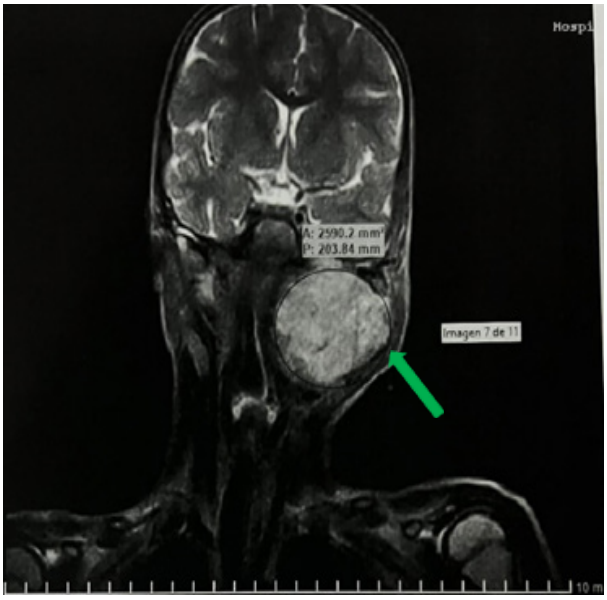

Fig.1. Magnetic Resonance Imaging. Coronal Slice. Extensive lesion occupying the left parotid region, left masseter and pterygoid muscles.

Positron Emission Tomography and Computed Tomography (PET/CT)

A dose of 2.2 mCi of 18F-FDG was intravenously administered. After 60 minutes, blood glucose was confirmed to be 5.2 mmol/l.

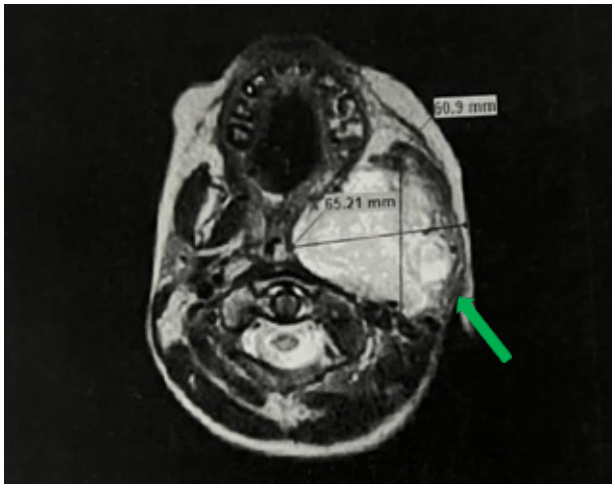

Fig. 2. Magnetic Resonance Imaging. Axial Slice. The lesion obliterates the nasopharynx, obstructs, and displaces the upper airway.

Positron Emission Tomography was performed from the cranial vertex to the mid-third of the femurs, along with a low-dose, non-contrast Computed Tomography for attenuation correction and anatomical localization of hypermetabolic areas identified in the PET study.

Abnormal areas of glycolytic hypermetabolism are observed in the head and neck. Furthermore, an extensive heterogeneous tumoral mass with an average density of 24 HU (Hounsfield Units) is seen in the left parotid region at the level of the nasopharynx. This mass compresses and displaces the upper airway, vascular structures, and the left maxillary sinus to the right without infiltrating it, while infiltrating the soft tissues at that level. Its largest dimensions measure 71.68 x 48.02 x 54.60 mm, with a maximum Standardized Uptake Value (SUV) of 7.11.

In the context of the patient's clinical presentation, the described mass exhibits glycolytic activity consistent with a primary tumor at that site.

Immunohistochemistry

A tissue sample obtained by biopsy was received. Microscopic examination reveals a tumor with histological characteristics of spindle cell proliferation, nuclear atypia, high mitotic activity, areas of tumor necrosis, and an infiltrative growth pattern, consistent with a peripheral nerve sheath tumor.

64-Slice Computed Tomography (CT) Scan

The study of the neck region reveals a hyper-

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dense (40-54 HU) tumoral-appearing, heterogeneous mass occupying the left masticator, parapharyngeal, pre- and post-pterygoid, and parathyroid spaces, with no clear interface with the left medial and lateral pterygoid muscles and part of the temporalis muscle on that side.

It causes left-sided asymmetry of the left lateral wall of the nasopharynx. A lytic lesion is noted in the left mandibular ramus (largest diameter 2.3 cm), the zygomatic process of the temporal bone, and bone lysis with destruction of the infratemporal surface of the greater wing of the sphenoid bone. Bone erosion of the zygomatic bone and pterygoid process is observed, along with anatomical distortion of the mandibular condyle affecting the temporomandibular joint, all on the left side. A tracheostomy tube is in place, figures 3 and 4.

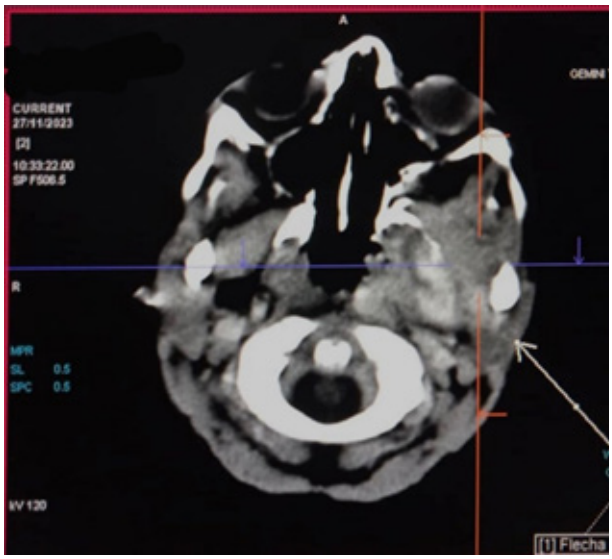

Fig. 3. Computed Tomography Scan. Axial View. Lesion occupying the left masticator, parapharyngeal, pre- and post-pterygoid, and parathyroid spaces.

64-Slice Contrast-Enhanced Computed Tomography Scan

The tumor area is measured to be 30x26 mm in its largest axial diameters. It causes distortion of the adjacent muscles and obscuration of the perilesional fat. A lytic lesion of 12x6 mm is noted in the left mandibular ramus. Bone destruction of the greater wing of the sphenoid and bone erosion of the zygomatic bone and mandibular condyle are observed, with involvement of the temporomandibular joint, all on the left side, showing no change compared to the previous study.

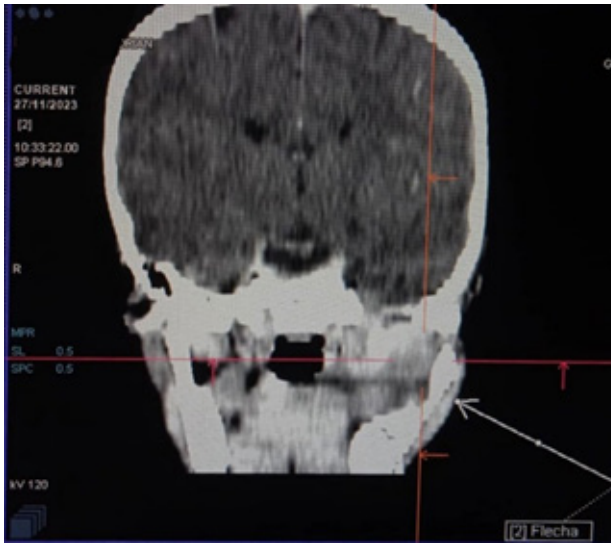

Fig. 4. Computed Tomography Scan. Coronal View. Tumor mass with a lytic lesion of the left mandibular ramus.

Computed Tomography Scan of the Head, Neck, and Thorax

Skull/Neck: A space-occupying lesion is seen in the left temporal lobe, associated with mild effacement of the gyri and sulci in the left cerebral hemisphere. The previous image comes from the infiltration by an extensive tumor mass occupying the entire left facial skeleton, with loss of normal anatomy of all muscle groups. There is infiltration of the left pterygopalatine and masticator spaces, with extension and continuity to the posterior wall of the orbit, the medial and lateral walls of the left maxillary sinus, the body of the sphenoid bone, the lesser wing of the sphenoid, and destruction of the hard palate. The airway is displaced to the right and obstructed at the level of the oropharynx.

Thorax: Presence of multiple bilateral parenchymal pulmonary nodules of secondary appearance, the largest measuring 20 mm in the medial basal segment of the left lower lobe.

Bone: Presence of osteolytic lesions of secondary appearance in the anterior wall of C2 and the left transverse process of T11.

A diagnosis of malignant peripheral nerve sheath tumor is made. The case is discussed in the head and neck consultation at the National Institute of Oncology, where it is deemed inoperable due to the tumor's extent. It is decided to administer oncologic-specific treatment with neoadjuvant chemotherapy according to protocol, for a duration of 6 months. The chemotherapeutic treatment is outlined in Table 1.

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Table 1. Chemotherapeutic Treatment

Week	Medications	Presentation	Dosage
1st	Ifosfamida + Doxorrubicina	Ifosfamida (Ampoule 1 g)	Ifosfamida 3 g/m²/Dose Days 1, 2 y 3 Intravenous way
4th	Ifosfamida + Doxorrubicina		
7th	Ifosfamida	Doxorrubicina (Ampoule 10 g)	Doxorrubicina 37,5 mg/m²/Dose Días 1 y 2 Intravenous way Cumulative dose of doxorrubicina 375 mg/m² Intravenous way
10th	Ifosfamida		
16th	Ifosfamida + Doxorrubicina		
19th	Ifosfamida + Doxorrubicina		
22nd	Doxorrubicina		

Evaluation at week 13: The tumor is still deemed unresectable, and treatment continues.

Upon completion of the treatment, the patient shows no response and is experiencing clear disease progression. The treatment is changed to Cyclophosphamide (50 mg tablet) 25 mg/m² daily, orally, and Vincristina (1 g vial) 1.5 mg/m² on days 1, 8, and 15, intravenously.

The possibility of ionizing radiation therapy is discussed with the radiotherapy team. After clinical and imaging evaluation, it is decided that due to the advanced stage of the local and distant (pulmonary and bone metastases) disease, the patient is not a candidate for radiotherapy with curative or palliative intent.

Given the extent of the disease and after discussion with the multidisciplinary teams, it is reaffirmed that the patient is experiencing clear disease progression. He is admitted to the Intensive Care Unit of the Pepe Portilla Provincial Teaching Pediatric Hospital.

Deemed suitable for palliative care, chemotherapy with Ifosfamide (1 g vial) 3 g/m²/dose intravenously and Etoposide (100 mg vial) 5 g/m²/dose intravenously is proposed, with the aim of improving local symptoms. A tracheostomy is performed to ensure a patent and secure airway for treatment administration, as the airway is currently compromised due to secondary displacement from tumor compression.

For pain relief, treatment is initiated with Paracetamol (Acetaminophen) 300 mg suppositories, at a dose of 150 mg every 6 hours.

Discussion

This case is notable for the unusual occurrence of an MPNST in a pediatric patient, an age group in which these tumors are rare. A wide range of imaging studies, including MRI and CT, were performed. These not only confirmed the diagnosis but also provided detailed information on the tumor's location, morphological characteristics, and its relationship with adjacent structures. The combination of these methods allowed for a comprehensive evaluation of the case, highlighting the necessity of a multidisciplinary approach in the diagnosis of MPNSTs in pediatric patients.

MPNST is an uncommon neoplasm, with an estimated incidence of 0.001 % in the general population. It represents 10 % of all soft tissue sarcomas. This is an aggressive entity whose origin derives from nerve sheath cells: Schwann cells, perineural fibroblasts, or endoneurial fibroblasts.[5]

According to Chávez Sánchez SA, et al.,[6] they are commonly located in the extremities (40 to 45 %), although they can also appear in the trunk (22 %), head and neck (21 %), and retroperitoneum (15 %). Distant metastases have been described in almost 50 % of patients, with the most frequent sites being the lungs, bones, lymph nodes, and liver. In this case, the patient's MPNST is located in the head and neck and, in its final stage, has pulmonary and bone metastases.

NF1 is often associated with multiple tumors, with MPNST being one of the most frequent. It is related to the loss of sequence on chromo-

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somal arm 17q, which leads to the inactivation of this gene. Additionally, tumors of the central nervous system and varieties of gastrointestinal stromal tumors are also associated.[7]

In the study by Parentini F, et al.,[1] the case involves a patient with NF1, which does not coincide with the present case. The patient in this report has no personal history of NF1 nor a family history of MPNST; therefore, the cause of the tumor is unknown, and its appearance is sporadic.

MPNSTs may be detected as palpable masses. For diagnosis, clinical, histological, immunohistochemical, and ultrastructural information suggesting Schwann cell differentiation is important. To complement the study, imaging support is essential, with magnetic resonance imaging being the examination of choice. Irregular margins, perilesional edema, cystic degeneration, heterogeneous enhancement on T1, and irregular contrast uptake on T1 can be observed.[1,6]

Establishing the differential diagnosis between MPNST, fibrosarcoma, and leiomyosarcoma can be even more challenging. This is because these typical lesions consist of a fusocellular population. Although neural tumors are distinguished by the characteristic "wavy" appearance of their cells, and leiomyosarcoma has more eosinophilic cytoplasm with less pointed nuclei compared to fibrosarcoma, these entities have different variants, including the presence of "epithelioid" cells, which can obscure the morphological diagnosis.[8]

Differential diagnosis between a benign peripheral nerve sheath tumor (neurofibroma) and its malignant counterpart (MPNST) can be complex, as some neurofibromas can be quite cellular and even have occasional pleomorphic cells. However, the mitotic count in these cases is crucial for establishing the diagnosis, as MPNSTs exhibit high mitotic activity.[8]

According to Fagúndez Núñez JM, et al.,[9] the choice treatment is complete surgical excision with oncological margins of at least one centimeter, which reduces the risk of recurrence. In their study, a left submaxillectomy was performed. In the clinical case by Ruiz Martín I, et al.,[10] the treatment is adjuvant radiotherapy with a dose of 56 Gy. These data differ from the present investigation, where, due to the tumor's extent, it was deemed inoperable, and chemotherapy was decided upon.

The use of adjuvant systemic chemotherapy is recommended due to the presence of disseminated disease, tumor size, and histological grade. For disseminated disease, first-line treatment with anthracyclines is used, in some cases combined with ifosfamide.

Ethical considerations

This research is conducted in accordance with ethical norms, as outlined in the Declaration of Helsinki [11], adhering to the principles of beneficence, non-maleficence, justice, and autonomy. The information is used for scientific purposes and to expand knowledge. The confidentiality of the data obtained is respected with the corresponding informed consent from the legal guardians.

Conclusions

Peripheral nerve sheath tumors are malignant soft tissue tumors that are rare and aggressive in pediatric patients. Their diagnosis requires clinical interpretation and the support of imaging studies, with magnetic resonance imaging being the fundamental examination of choice. The recommended treatment is complete surgical excision; however, in this case, it was not possible, and chemotherapy was utilized as the treatment.

References

1. Parentini F, Rojas J, Fernández F, Bermeo J. Tumor maligno de la vaina nerviosa periférica del nervio vago: Reporte de un caso. Rev. Otorrinolaringol. Cir. Cabeza Cuello [Internet]. 2021 [citado 11/3/2025]; 81(2): 232-236. Available from: http://www.scielo.cl/scielo.php?script=sci_arttext&pid=S0718-48162021000200232&lng=es.
2. Briceño Morales C, Acosta Ortiz S, Alarcón Durán LA, Hernández-Gómez JA. Resección multivisceral para el tratamiento de un tumor maligno de la vaina del nervio periférico intraabdominal. Rev. colombia. cir. [Internet]. 2024 [cited 11/3/2025]; 39(3): 467-469. Available from: http://www.scielo.org.co/scielo.php?script=sci_arttext&pid=S2011-75822024000300467&lng=en.
3. Lozano P, Almanza L, Juárez S, Gallmann A, Díaz Alfaro R, Gomez Zanni M, et al. Tumor maligno de la vaina nerviosa periférica: reporte de un caso. Rev. argent. dermatol. [Internet]. 2024 [cited 11/3/2025]; 105: 3-3. Available from: https://www.scielo.org.ar/scielo.php?script=sci_arttext&pid=S1851-300X2024000100003&lng=es.
4. Sepúlveda Villegas CA, Jurado Basildo C, No-

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Columna Méd. 2026; Vol. 5: e313

voa Ferro M, Del Campo Estepar S, Aleman Millares R. Tumores del sistema nervioso periférico: el papel del radiólogo, anatomopatólogo y cirujano. seram [Internet]. 2021 [cited 12/3/2025];1(1). Available from: https://www.piper.espacio-seram.com/index.php/seram/article/view/4378

5. Ruiz Martín I, Ramos Zayas A, Torres Calcines N, Sánchez Aniceto G. Tumor maligno de la vaina nerviosa periférica mandibular. Caso clínico y revisión de la literatura. Rev Esp Cirug Oral y Maxilofac [Internet]. 2020 [cited 12/3/2025]; 42(3): 132-135. Available from: http://scielo.isciii.es/scielo.php?script=sci_arttext&pid=S1130-05582020000300006&lng=es.

6. Chávez Sánchez SA, Bellido Caparó Á, García Encinas CA, Gallegos Serruto GS, Bravo Taxa MP, Vásquez Morales VM. Hemoperitoneum secondary to a malignant tumor of the sheath of the peripheral nerve in the liver. Rev. gastroenterol. Perú [Internet]. 2024 [cited 22/3/2025]; 44(2): 140-144. Available from: http://www.scielo.org.pe/scielo.php?script=sci_arttext&pid=S1022-51292024000200140&lng=es.

7. Martínez Barrios E, Bortolatto L, Martínez Bogado E, Noguera OG. Neurofibromatosis 1 asociada a tumor maligno de la vaina del nervio periférico: Un enfoque radiológico. An. Fac. Cienc. Méd. (Asunción) [Internet]. 2020 [cited 22/3/2025]; 53(2): 157-164. Available from: http://scielo.iics.una.py/scielo.php?script=sci_arttext&pid=S1816-89492020000200157&lng=en.

8. Hernández MG, Acosta M, Ramírez AK, Paredes HR, Marín CE, Hernández RJ, et al. Tumor maligno de la vaina del nervio periférico primario de mama informe de caso Unidad de Mastología Clínica Leopoldo Aguerrevere. Rev Venez Oncol. [Internet]. 2017 [cited 23/3/2025]; 29(2):123-129. Available from: https://www.redalyc.org/articulo.oa?id=375650363007

9. Fagúndez Núñez JM, Salazar L, Araujo J, Mota S. Cedeño N. Neoplasias del sistema nervioso periférico en cabeza y revisión de la literatura. Rev Venez Oncol. [Internet]. 2023 [cited 5/9/2025]; 35(4):250-259. Available from: https://www.redalyc.org/articulo.oa?id=375675350006

10. Ruiz Martín I, Ramos Zayas A, Calcines Torres N, Sánchez AG. Tumor maligno de la vaina del nervio periférico mandibular. Caso clínico y revisión de la literatura. Rev Esp Cirug Oral y Maxilofac [Internet]. 2020 [cited 5/9/2025]; 42(3): 132-135. Available from: http://scielo.isciii.es/scielo.php?script=sci_arttext&pid=S1130-05582020000300006&lng=es.

11. Rodríguez Puga R. Nuevas directrices de la Declaración de Helsinki, impacto y ética en la investigación médica actual. Rev Cubana Oftalmol [Internet]. 2025 [cited 7/9/2025]; 38:e2043. Available from: https://revoftalmologia.sld.cu/index.php/oftalmologia/article/view/2043

Authorship declaration

Mitjans Hernández D: Conceptualization, investigation, methodology, resources, supervision, visualization, writing – original draft, writing – review & editing.

Hernández González EA: Conceptualization, investigation, methodology, visualization, writing – original draft, writing – review & editing.

Rivera López SM: Investigation, methodology, writing – original draft, writing – review & editing.

Conflict of interest

The authors declare no conflict of interest.

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