Thoracoabdominal chordoma in a pediatric patient. A rare entity

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ABSTRACT

Introduction. Chordoma is a rare, slow-growing notochordal neoplasm typical of adults. Less than 5% of the cases occur in children, where they are located at the skull base. Treatment involves surgical resection with or without radiotherapy.

Clinical case. 14-month-old patient with a left dorsal lumbar mass, pain, and limited mobility in the lower limbs. MRI showed a left paravertebral mass infiltrating the spinal canal (T11-L1), in contact with the parietal pleura, along with abdominal extension. Chordoma diagnosis was established based on percutaneous biopsy. Full resection without safety margins was carried out using the posterior approach. One month later, tumor recurrence caused hemothorax, pleural infiltration, and rib infiltration. Two chemotherapy cycles were administered, with a good response. Reintervention was successfully conducted one month later. After 9 months, the patient is free from disease, under chemotherapy treatment and proton therapy.

Discussion. Although unusual, chordoma is included within the differential diagnosis of retroperitoneal masses. Prognosis depends on full resection.

KEY WORDS: Germ cell neoplasm; Chordoma.

CORDOMA TORACOABDOMINAL EN PACIENTE PEDIÁTRICO. UNA ENTIDAD POCO FRECUENTE

RESUMEN

Introducción. El cordoma es una neoplasia de crecimiento lento, infrecuente, típico de adultos, de origen notocordal. Menos del 5% ocurre en niños, localizándose en la base craneal. El tratamiento implica la resección quirúrgica asociando o no radioterapia.

Caso clínico. Paciente de 14 meses con masa dorsolumbar izquierda, dolor y limitación motora en miembros inferiores. La resonancia mostró masa paravertebral izquierda introduciéndose en canal medular (T11-L1), en contacto con la pleura parietal, y extensión abdominal. La biopsia percutánea fue diagnóstica para cordoma. Se realizó resección completa sin márgenes de seguridad, mediante abordaje posterior. Al mes debutó con hemotórax por recidiva tumoral, infiltración pleural e implante costal. Recibió dos ciclos de quimioterapia con buena respuesta, reintervinéndose al mes exitosamente. Tras 9 meses se encuentra libre de enfermedad, en tratamiento quimioterápico y protonterapia.

Comentarios. Aunque inusual, el cordoma forma parte del diagnóstico diferencial de masa retroperitoneal. Su pronóstico dependerá de la resección completa.

PALABRAS CLAVE: Neoplasia de células germinales; Cordoma.

INTRODUCTION

Chordoma is a rare, slow-growing notochordal neoplasm typical of 40- to 70-year-old adults. Malignancy is low or medium, but with a tendency to recurrence. Consequently, in spite of its slow progression, it is locally aggressive, it can infiltrate the surrounding tissues, and it is potentially fatal. With an incidence of 1 in 1,000,000 people, less than 5% of the cases occur in children[1,2]. In adults, it is more frequent in male patients, and in children, it is more common in girls. It can be located along the backbone, mostly at the sacral level, but it is more common at the skull base in pediatric patients[3].

Clinical signs emerge progressively, with pain and gradual, incapacitating neurological impairment according to tumor location. Even though imaging tests such as X-ray, MRI, or CT-scan can help establish diagnosis, the latter will only be confirmed based on biopsy results.

Treatment involves surgical resection with or without radiotherapy. Even though this surgery is typically conducted by neurosurgeons or traumatologists, we present the case of a chordoma found at a rare (thoraco-abdominal) location in a pediatric patient that required the cooperation of pediatric surgeons for resection purposes. Early recurrence, which is unusual, is also described herein.

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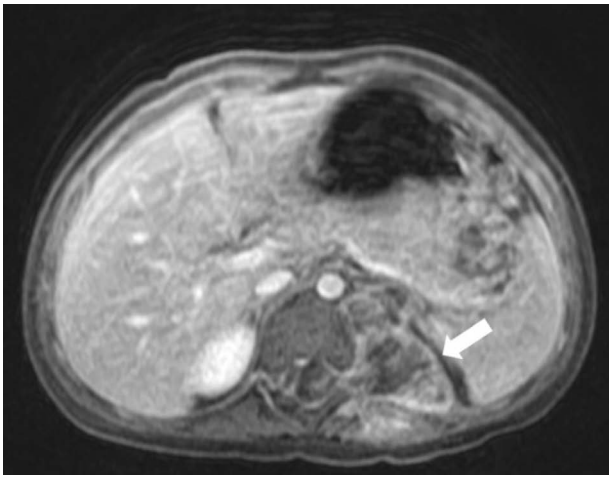

Figure 1. Left paravertebral mass infiltrating the conjunction holes and causing spinal compression at the level of the T11-L1. It also infiltrated the left paraspinal muscles.

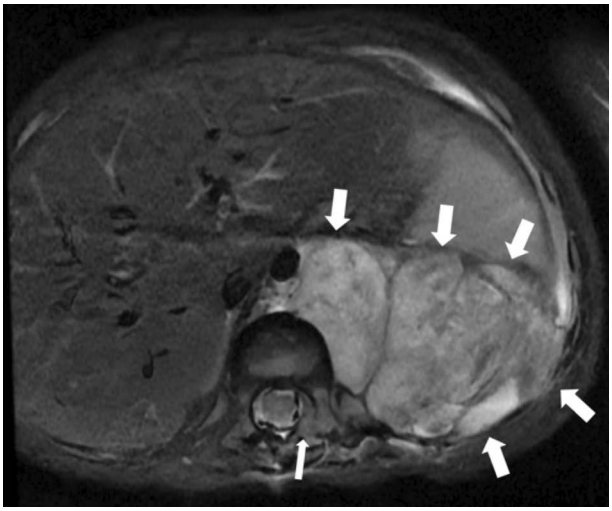

Figure 2. MRI revealed recurrence, with a large tumor mass, infiltration of T11-L1 conjunction holes and of the left posteroinferior rib wall, with thoracoabdominal aorta displacement.

CLINICAL CASE

14-month-old male patient with a left dorsal lumbar mass, dorsal pain, and limited mobility in the lower limbs. MRI revealed a left paravertebral mass infiltrating the conjunction holes and causing spinal compression at T11-L1, in contact with the left basal parietal pleura and with abdominal extension, reaching the posterior renal aspect. It also infiltrated the left paraspinal muscles (Fig. 1).

An ultrasound-guided thick needle biopsy was carried out. Once chordoma diagnosis had been confirmed –in spite of the difficulties in terms of sample processing owing to its rarity–, the patient underwent surgery one month later. Removal was conducted using a posterior spinal approach with T11 costotomy for retroperitoneal access, given that the total surface of the tumor could not be approached differently. Following resection, the rib was put back in place and fixated with a plate. The injury was fully removed, including the biopsy trajectory. At the pathological level, no tumor-free margins could be achieved even though they appeared as necrosis and inflammation in the intraoperative biopsies. The rest of the deferred study was also reported as wide necrosis and inflammation without tumor cells.

One month later, the patient developed respiratory impairment, with chest X-ray demonstrating a left pleural effusion. Thoracocentesis revealed the presence of hemothorax, but the presence of malignant cells was not assessed. Recurrence was confirmed at MRI, which showed that the tumor had grown larger than the initial mass. It also demonstrated pleural infiltration and infiltration of T11-L1 conjunction holes, of the left posteroinferior rib wall, and of the posterior renal wall, with thoracoabdominal aorta displacement (Fig. 2).

Two cyclophosphamide, vincristine, and daunorubicin chemotherapy cycles were administered, with a significant reduction in tumor volume at control MRI two weeks later. Reintervention was carried out one month following relapse. To access the tumor, a dorsal-lumbar incision was performed in the midline, with left lateral discharge at the level of the 12th rib, resection of the 11th and 12th ribs, and lateral costotomy in the 9th and 10th ribs, which were subsequently luxated to cephalad to adequately expose the vertebral and abdominal components. Electrophysiological monitoring was applied at all times, without any disorders noted. The mass was removed along with the parietal pleura, the intercostal and paravertebral muscles, the adhered diaphragm, and the T12 pedicle and left root, this time with free margins. During closure, 9th and 10th rib reconstruction was carried out using titanium plates (Fig. 3). An abdominal Jackson-Pratt drainage and a thoracic drainage were placed and subsequently removed on postoperative days 3 and 4.

Following surgery, the patient stayed at the pediatric ICU for 72 hours, and remained hospitalized for 4 extra days, with a good progression. After discharge, he has received 7 chemotherapy cycles, and most recently, proton therapy at the surgical bed. No further relapses have been observed at the follow-up imaging tests.

DISCUSSION

Chordoma is a rare bone tumor. Originating from notochordal remnants, it emerges at the clivus, the backbone, or the sacrum, which is why it sometimes has an abdominal component, as in our case. It has a high local recurrence rate, and it sporadically presents lung, bone, and liver metastases. It can also be associated with tuberous sclerosis[4].

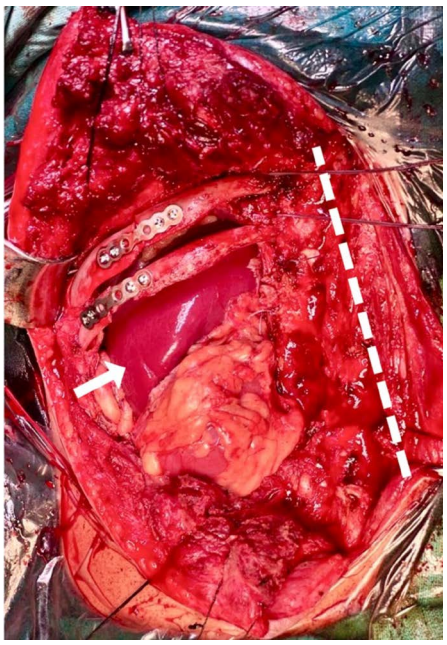

Figure 3. Surgical closure image. 9th and 10th rib reconstruction with titanium plates. Absence of the 11th and 12th ribs, which were resected at surgery. The spleen *(arrow)* and the backbone *(hyphens)* can be identified.

The 5-year overall survival rate in children is 50-80%[1,5]. It is lower in patients under 18 years and above 65, and in the presence of bladder or intestinal disfunction at initiation, recurrences, and metastasis. Prognosis is also worse in younger children[1,5-7].

Given that it is a slow-growing pathology, clinical signs can emerge even years later, with location-dependent symptoms. When located at the skull base, it is associated with headache and cranial nerve disorders –primarily diplopia as a result of cranial nerve VI involvement. It can also present with rhinorrhea. At the cervical level, it is associated with cervical and upper limb pain, radiculopathy, myelopathy, and even odynophagia. At the chest and lumbar levels, it can also cause non-specific pain, bone fractures, and nervous system disorders. These signs can be accompanied by bladder, intestinal, and autonomic nervous system disfunction in the presence of sacral involvement.

For diagnosis to be established, imaging tests combined with biopsy are required. X-ray of the area where the tumor is suspected will show a lytic injury, typically > 5 cm and with a mass effect in the surrounding tissues. CT-scan reveals chordoma in a most precise fashion, with hypodense, lytic areas, irregular calcifications, occasionally peripheral sclerosis, and moderate to high contrast enhancement. It also allows metastasis to be located. MRI defines the spread to soft parts in a more accurate manner and determines spinal and nerve root involvement. In the T1-weighted images, it shows low signal intensity, and in the T2 weighted images, it shows hyperintensity. Following gadolinium administration, it appears as a T1-weighted honeycomb image.

Diagnosis is definitive only after pathological examination. A needle or open biopsy can be performed –in the first case, the needle's trajectory should be included in the tumor resection in light of the high recurrence rates observed. Microscopically, a lobular pattern with physaliphorous cells immersed in the myxoid matrix is observed. At least four histopathological variants –classic, chondroid, undifferentiated, and sarcomatoid– have been described. The classic and the chondroid variants are the most frequent in children, whereas the undifferentiated one is less common and has a worse prognosis, but it is more frequent than in adults[1,8]. In addition, the chondroid variant has a worse prognosis than the classic one[9].

Genetically speaking, changes in the TBXT gene, which encodes the brachyury protein, are involved in sporadic and familial cases. In addition, the BAF47 gene expression loss has a worse prognosis[10], as well as the SMARCB1 gene inactivation, which is characteristic of poorly differentiated cases[11].

Chordoma treatment options include surgery with or without radiotherapy, and tyrosine kinase inhibitor therapy. Full en-bloc resection –or piecemeal resection if the former is not feasible– is key for an adequate progression[12].

When access to the tumor mass is highly complex, a tumor reducing surgery is carried out. This allows for symptom improvement while leaving as few remnants as possible for subsequent radiotherapy treatment. Association with extracorporeal radiotherapy reduces recurrence rates because even though these neoplasms are relatively resistant to radiation, the fact they are frequently located at the skull base in children makes removal more difficult[13]. Proton beam therapy is the most commonly used radiotherapy[5,14,15].

On the other hand, tyrosine kinase inhibitors such as imatinib, sorafenib, or erlotinib have been used in adults, given the overexpression of the PDGFRA, PDGFRB, and *KIT* genes[16,17]. The results are difficult to assess, since partial response is achieved in few cases, whereas disease stability is reached in most of them, and it cannot be ruled out this is due to the slow progression of this neoplasm[18].

The use of chemotherapy in these tumors has not been clearly defined. It has been used as a first-line treatment[19] or as an adjuvant therapy with or without radiotherapy[20] in very few cases. Current essays are focused on synthetizing drugs aimed at specific molecular targets and on combining atezolizumab and tiragolumab in tumors with SMARCB1 deficit.

Frequent follow-up with MRI or CT-scan is required following surgery, with periodicity depending on resection degree, since residual tumors reduce recurrence times.

This clinical case aims to review the current management of chordoma and to highlight the fact it is a very rare pathology that often grows in a slow and progressive manner. However, there are exceptions, such as our case, which should be considered in the differential diagnosis of retroperitoneal neoplasms.

REFERENCES

1. Hoch BL, Nielsen GP, Liebsch NJ, Rosenberg AE. Base of skull chordomas in children and adolescents: A clinicopathologic study of 73 cases. Am J Surg Pathol. 2006; 30(7): 811-8.
2. Lau CSM, Mahendraraj K, Ward A, Chamberlain RS. Pediatric chordomas: A population-based clinical outcome study involving 86 patients from the Surveillance, Epidemiology, and End Result (SEER) database (1973-2011). Pediatr Neurosurg. 2016; 51(3): 127-36.
3. Sebro R, DeLaney T, Hornicek F, Schwab J, Choy E, Nielsen GP, et al. Differences in sex distribution, anatomic location and MR imaging appearance of pediatric compared to adult chordomas. BMC Med Imaging. 2016; 16(1): 1-7.
4. Sebro R, DeLaney T, Hornicek F, Schwab J, Choy E, Nielsen GP, et al. Differences in sex distribution, anatomic location and MR imaging appearance of pediatric compared to adult chordomas. BMC Med Imaging. 2016; 16(1): 1-7.
5. Yasuda M, Bresson D, Chibbaro S, Cornelius JF, Polivka M, Feuvret L, et al. Chordomas of the skull base and cervical spine: Clinical outcomes associated with a multimodal surgical resection combined with proton-beam radiation in 40 patients. Neurosurg Rev. 2012; 35(2): 171-83.
6. Jian BJ, Bloch OG, Yang I, Han SJ, Aranda D, Parsa AT. A comprehensive analysis of intracranial chordoma and survival: A systematic review. Br J Neurosurg. 2011; 25(4): 446-53.
7. Chambers KJ, Lin DT, Meier J, Remenschneider A, Herr M, Gray ST. Incidence and survival patterns of cranial chordoma in the United States. Laryngoscope. 2014; 124(5): 1097-102.
8. McMaster ML, Goldstein AM, Bromley CM, Ishibe N, Parry DM. Chordoma: Incidence and survival patterns in the United States, 1973-1995. Cancer Causes Control. 2001; 12(1): 1-11.
9. Tsitouras V, Wang S, Dirks P, Drake J, Bouffet E, Hawkins C, et al. Management and outcome of chordomas in the pediatric population: The Hospital for Sick Children experience and review of the literature. J Clin Neurosci. 2016; 34: 169-76.
10. Beccaria K, Tauziède-Espariat A, Monnien F, Adle-Biassette H, Masliah-Planchon J, Pierron G, et al. Pediatric chordomas: Results of a multicentric study of 40 children and proposal for a histopathological prognostic grading system and new therapeutic strategies. J Neuropathol Exp Neurol. 2018; 77(3): 207-15.
11. Hasselblatt M, Thomas C, Hovestadt V, Schrimpf D, Johann P, Bens S, et al. Poorly differentiated chordoma with SMARCB1/INI1 loss: a distinct molecular entity with dismal prognosis. Acta Neuropathol. 2016; 132(1): 149-51.
12. Rassi MS, Hulou MM, Almefty K, Bi WL, Pravdenkova S, Dunn IF, et al. Pediatric clival chordoma: A curable disease that conforms to Collins' law. Neurosurgery. 2018; 82(5): 652-9.
13. McDowell MM, Zwagerman NT, Wang EW, Snyderman CH, Tyler-Kabara EC, Gardner PA. Long-term outcomes in the treatment of pediatric skull base chordomas in the endoscopic endonasal era. J Neurosurg Pediatr. 2021; 27(2): 170-9.
14. Delaney TF, Liebsch NJ, Pedlow FX, Adams J, Weyman EA, Yeap BY, et al. Long-term results of Phase II study of high dose photon/proton radiotherapy in the management of spine chordomas, chondrosarcomas, and other sarcomas. J Surg Oncol. 2014; 110(2): 115-22.
15. Rombi B, Ares C, Hug EB, Schneider R, Goitein G, Staab A, et al. Spot-scanning proton radiation therapy for pediatric chordoma and chondrosarcoma: Clinical outcome of 26 patients treated at paul scherrer institute. Int J Radiat Oncol Biol Phys. 2013; 86(3): 578-84.
16. Casali PG, Messina A, Stacchiotti S, Tamborini E, Crippa F, Gronchi A, et al. Imatinib mesylate in chordoma. Cancer. 2004; 101(9): 2086-97.
17. Stacchiotti S, Longhi A, Ferraresi V, Grignani G, Comandone A, Stupp R, et al. Phase II study of imatinib in advanced chordoma. J Clin Oncol. 2012; 30(9): 914-20.
18. Lebellec L, Chauffert B, Blay JY, Le Cesne A, Chevreau C, Bompas E, et al. Advanced chordoma treated by first-line molecular targeted therapies: Outcomes and prognostic factors. A retrospective study of the French Sarcoma Group (GSF/GETO) and the Association des Neuro-Oncologues d'Expression Française (ANOCEF). Eur J Cancer. 2017; 79: 119-28.
19. Melchionda F, Oncology P, Spreafico F, Unit PO, Hemato-oncology P, Ciceri S, et al. A novel WT1 mutation in familial Wilms tumor. Pediatr Blood Cancer. 2013; 60(8): 1388-9.
20. Dhall G, Traverso M, Finlay JL, Shane L, Gonzalez-Gomez I, Jubran R. The role of chemotherapy in pediatric clival chordomas. J Neurooncol. 2011; 103(3): 657-62.