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Case report

Thoracic synovial cyst as cause of cord compression in a patient with Charcot-Marie-Tooth disease

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Abstract: An 80-year-old male patient affected by Charcot-Marie-Tooth (CMT) disease came to our attention in July 2020, for the occurrence of low back pain and lower limb weakness, and also saddle anesthesia, urinary and faecal retention were referred. His diagnosis of CMT is dated back to 1955 and through the years, the clinical picture slowly worsened but never got particularly severe. The quick symptoms outbreak and the presence of urinary disturbances were red flags, which lead us to direct the diagnostic orientation elsewhere. A Magnetic Resonance Imaging of the thoraco-lumbar spinal cord was then performed and it was suggestive for synovial cyst at T10-T11. The patient underwent a decompression with laminectomy and then stabilized through arthrodesis. In the very next days after the surgery, the patient showed a sudden and significant improvement of his condition. At the last visit, he showed a remarkable relief of the symptoms, walking by himself. (www.actabiomedica.it)

Key words: Synovial Cyst, Charcot-Marie-Tooth Disease, Cord compression, Thoracic

Background

Spinal cyst is a rare occurrence in the daily practice of a spinal surgeon, though it could give significant clinical pictures and represent an important challenge for the patient. They are cystic lesions linked to the facet joint with a synovial lining, as opposed to the so-called "pseudo-cyst", which is lined instead by fibrous-connective tissue (1, 2). The pathogenesis has not been defined yet, but destabilization and degeneration of the joint are the most reliable causes at the roots of the cystic shaping (3). Therefore, the vast majority of them occurs in the lumbar tract (4), the segment with greater mobility. Thoracic localization is uncommon (1), instead, due to the relatively limited mobility. Most of the time synovial cysts cause no symptoms at all, and they could become symptomatic when they gain size and induce a compression on the adjacent structures due to the mass effect (5,6). Clinical picture of cord compression is commonly seen then, however it depends on the localization and on the size of the cyst (7, 8).

Case Report

A 80-year-old male patient affected by Charcot-Marie-Tooth (CMT) disease came to our attention on July 2020, for the occurrence of low back pain and lower limb weakness, suddenly started in October 2019 and worsened since January 2020, but due to the SARS-CoV-2 Pandemic he delayed his decision to go

to the doctor (9, 10). The patient also referred to saddle anesthesia and urinary and faecal retention during the visit. His diagnosis of CMT dated back to 1955, with the disease presenting with progressive weakness in his hands and lower limbs. Through the years, the clinical picture slowly worsened but never got particularly severe: the patient reached the age of 80 able to walk autonomously with crutches and to carry out all the daily activities without help. He was even a non-agonistic cyclist, too, and used to ride a bike regularly during the weekends.

The last neurological examination before the occurrence of new symptoms showed a mild motor deficit of the lower limbs and left foot drop, therefore he used to wear an Ankle-Foot-Orthosis to walk autonomously. He had a slight symmetric hyporeflexia and a mild hypoesthesia of the lower limbs.

The sudden precipitation of the clinical picture led him to consult different physicians, but due to his medical history, those changes were repeatedly attributed to the progression of his primary disease. However, the quick symptoms outbreak and the presence of urinary disturbances were red flags, which led us to direct the diagnostic orientation elsewhere.

At the neurological examination conducted in October 2020, the patient was bright and alert, with a good overall state. Upright position was hardly kept. Both the lower limbs were paretic and the gait was limited. Extensor hallucis longus deficit was complete at the left foot, with a full drop foot, and partial at the right foot, both already present. He referred to tinglings and a worsening of the hypoesthesia in the lower limbs, prevailing on the right side; Thermal- tactile hypoesthesia with a D8 level and saddle anesthesia were also found. Deep tendon reflexes showed a brisk response. Babinski's sign was bilateral negative. Those findings suggested a central origin of the symptoms rather than a worsening in the peripheral nerve disturbance. A Magnetic Resonance Imaging (MRI) of the thoraco-lumbar spinal cord was then performed. What emerged was unexpected: an image of an extradural well-delineated lesion of cystic aspect in T10-T11, with a central component hyperintense on T2-weighted sequence and hypointense on T1-weighted

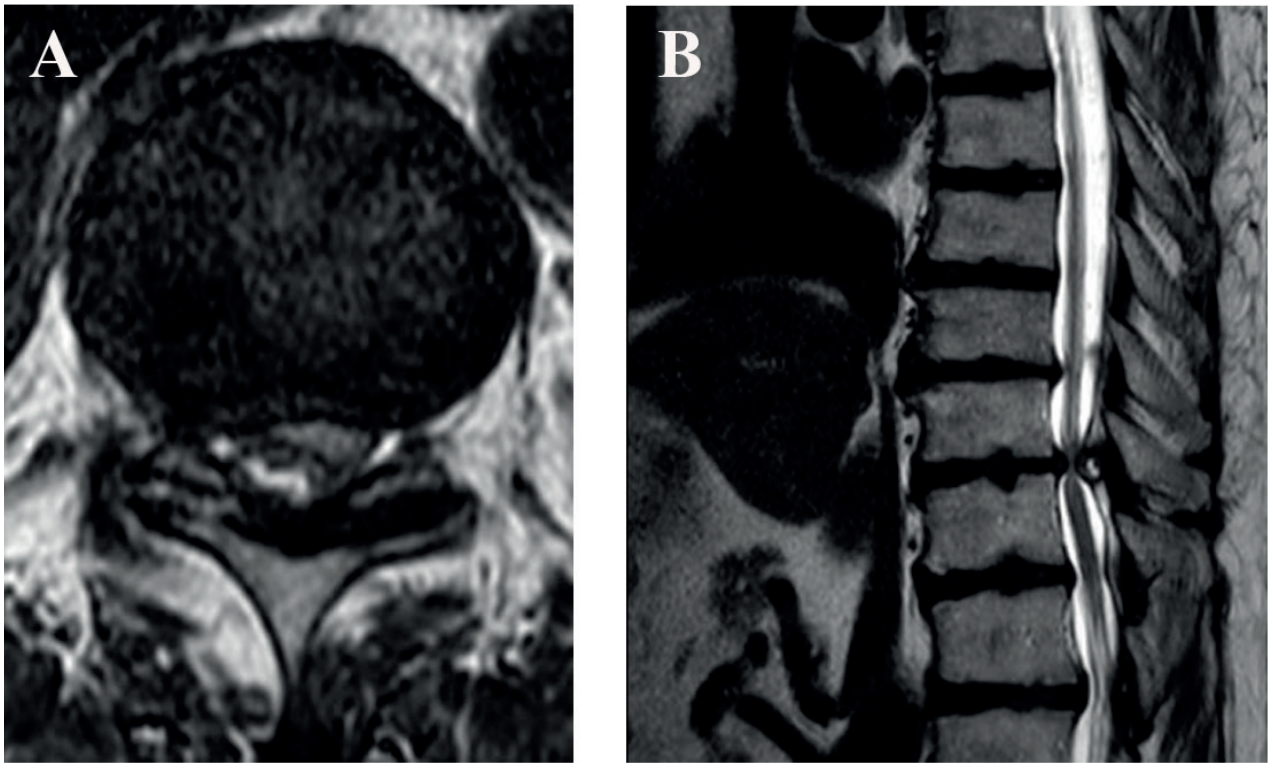

Figure 1. Preoperative T2-weighted axial (A) and sagittal (B) MRI of the thoraco-lumbar spine, showing the cystic lesion. In these sequences could be seen the hyperintense central component of the cyst.

sequence, suggestive for synovial cyst (Fig. 1). This lesion determined thoracic spinal cord compression from the right side.

Due to the result of the MRI and the clinical signs of myelopathy caused by the marked spinal cord compression, we opted for surgery. The patient underwent a decompression with laminectomy, through the use of the microscope. The cystic lesion intraoperatively seemed to originate from the ligamentum flavum, near to the facet joint and adherent to the dura. A complete excision of the lesion was gained. The spinal segment was then stabilized through arthrodesis with local morselized auto-graft, two bars and four pedicle screws in T10 and T11. The cystic lesion was preserved and sent to the anatomical pathology laboratory to be examined. Neuromonitoring remained stable throughout the procedure.

In the very next days after the surgery, the patient showed a sudden and significant improvement of his condition. He managed to walk alone again, with the help of two crutches. The sensitivity and motor deficit, previously quickly worsened, was then improved, and no more bladder and bowel incontinence was reported.

Five days after the surgery, we got the results of the histological examination, which confirmed the diagnosis (Fig. 2).

The findings were referable to a proper synovial cyst, due to the synovial lining. Deposits of calcium pyrophosphate and small fragments of ligamentum flavum were present at the periphery of the cystic structure. The patient went home from the hospital on his feet, ready to re-start his daily activities on the sixth day after the surgery.

The patient came back for the next follow-up at one, three and six months since the surgery. At the last visit, he showed a remarkable relief of the symptoms, walking by himself. He referred minimal low-back pain at his last follow-up, Visual Analogue scale (VAS) was 2 instead of 8 as it was before the surgery. His quality of life was improved, too, and it was assessed through the SF-12. Before the surgery, the patient had a Physical Score (PCS-12) of 16.82 and a Mental Score (MCS-12) of 44.22. At last follow-up, his PCS-12 and MCS-12 increased at 48.64 and 63.07, respectively. A new x-ray was performed to assess rods and screws position, without any evident alteration (Fig. 3).

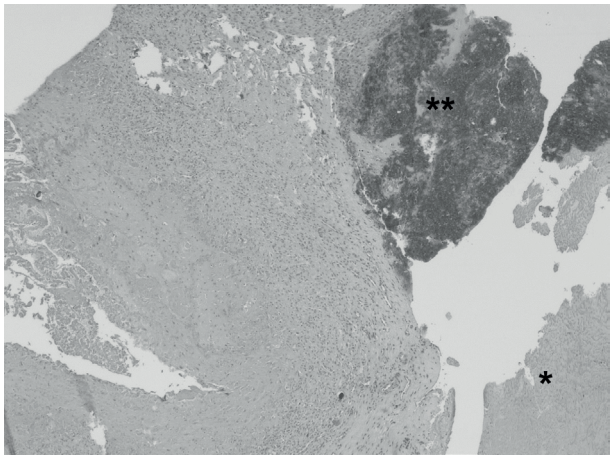

Figure 2. The histological examination revealed small fragments of a cystic structure composed of granulation tissue and synovial tissue. Deposits of calcium pyrophosphate (**) and small fragments of ligamentum flavum (*) were present at the peryphery of the structure.

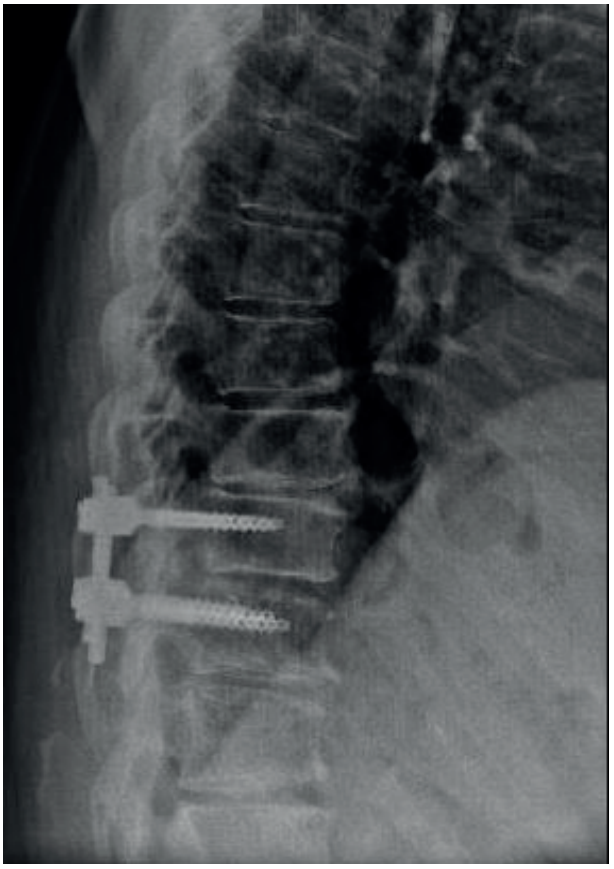

Figure 3. Post-operative X-Ray, where could be assessed the results of arthrodesis T10-T11.

Results and Conclusion

Thoracic cord compression due to synovial cyst is hardly found in the daily routine of a spine surgeon (1). Thoracic spine, in fact, has a smaller mobility than the lumbar tract, thus degeneration and instability of this segment are less accustomed to generating synovial cyst (5, 6).

In the Literature, it was possible to find 14 patients with similar clinical picture and the confirmed diagnosis of thoracic spine synovial cyst (11-21), eight female and six male patients, with an average age of 57.5 years. All the patients were admitted with chronic back pain and a progressive myelopathy started with weakness and numbness of the lower limbs. Two of them showed a clinical picture of Cauda syndrome. The group of patients showed similar diagnostic paths, and in different cases were recorded a delay of the diagnosis or a misdiagnosis, as it occurred in our case. In fact, two of the patients underwent other surgical procedures before to get the right diagnosis, due to the attribution of the symptoms to the wrong origin. For all the patients MRI was necessary and sufficient to hypothesize the diagnosis, and in ten cases the diagnosis was confirmed, as in this case, with an histological exam of the surgical sample. The most common site of the cystic lesion was the T11-T12 level, strictly followed by the T9-T10. Among the 14 patients, two of them were diagnosed with a different type of synovial cyst, that is the hemorrhagic cyst. The differential diagnosis between them was made possible through an accurate MRI exam. In common synovial cyst the lesion appears as hypointense in T1-weighted images, and hyperintense in T2-weighted images. In the haemorrhagic ones, instead, the exact opposite occurs, due to the presence of haemoglobin products (22-24).

Surgical treatment was chosen in all the cases. Decompression and a complete cyst excision was performed when possible, save for the lesions which were too adhered to the Dura, to avoid dural laceration. Two of the patients were treated also with an arthrodesis through pedicle screws and rods. All the patients recovered from the neurological deficits in the months after the surgery, and were able to progressively get back to their daily activities.

Back to our patient, an overlap of peripheral motor and sensory neuropathy and a spinal cord compression is a really rare occurrence, as they both represent uncommon conditions in themselves.

However, an accurate physical examination was sufficient not to linger over the underlying disease. In fact, as we started to sample the patient's medical history and to visit him, signs and symptoms turned out to be partially incongruous. Saddle anesthesia, urinary disturbances and the presence of brisk reflexes are uncommon findings for peripheral neuropathies like CMT; moreover, their subacute onset, after a long period of a stationary clinical picture, made the hypothesis of disease progression unconvincing.

Deepening the investigation through the MRI, allowed us to find out the synovial cyst, whose nature was later confirmed by the histological exam. It is possible that the lesion was already there before the onset of the new symptoms, and it progressively gained size during the years, until clinical manifestation of the cord compression appeared.

The excision of the cyst and the subsequent decompression of the spinal medullary canal, allowed the patient to get a significant and quick relief. We had the chance to meet him again one month after the surgery for the first follow-up, and he showed an improvement of his general conditions. He started physiotherapy treatment, looking forward to riding his bike again.

In prompt worsening of the clinical picture in patients with hereditary motor and sensory neuropathy, such as Charcot-Marie-Tooth disease, investigation on new onset conditions is needed, such as avoiding to focus exclusively on the underlying disease. Misdiagnosis or delayed diagnosis are a common bias in rare diseases, due to the usual routine-based analysis of clinical findings. Furthermore, an early diagnosis could give to the patient an immediate improvement of the clinical picture, with a deep and quick impact on the quality of life.

The presence of a chronic disease, especially when it is rare and offers limited treatment options, could represent a dangerous bias that may blind the physician in coping with the patient. Physical examination remains the cornerstone of the diagnostic process and the preliminary, essential tool to orientate the diagnostic algorithm.

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Informed Consent Statement: Informed consent was obtained from all individual participants included in the study.

Data Availability Statement: The data presented in this study are available on request from the corresponding author. The data are not publicly available due to privacy and ethical reasons.

Conflicts of interest: Each author declares that he or she has no commercial associations (e.g. consultancies, stock ownership, equity interest, patent/licensing arrangement etc.) that might pose a conflict of interest in connection with the submitted article.

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