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CASE REPORTS

A Technical Problem in Diagnosis Clarity of Solitary Spinal Cord Neurofibroma in an Eleven-month-old Boy

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Spinal neurofibroma is one of the rarest of the neoplasms involving the spinal cord or roots and occurs much less often than neurinoma, meningioma or glioma. The sixth pediatric case of solitary intramedullary tumor was described in 2013, according to B. Eljebbouri et al. We present a rare, difficult to diagnose and maybe the seventh pediatric case of solitary neurofibroma of the cauda equine in an 11-month-old infant. The patient underwent a laminectomy of T12, L1, L2 and L3, extirpation of intradural, intramedullary and extramedullary spinal cord tumor. The patient is fully recovered for 5 years of monitoring. Although rare, spinal neurofibromas in children should be diagnostically considered and radically treated for a favorable outcome.

INTRODUCTION

Spinal neurofibroma is one of the rarest of the neoplasms involving the spinal cord or roots and occurs much less often than neurinoma, meningioma or glioma.[1] They are uncommon, comprising approximately 3% of all spinal tumors and occur both sporadically and in association with neurofibromatosis 1 (NF1; von Recklinghausen's disease).[2] Intramedullary spinal cord tumors in children account for 4-10% of central nervous system tumors.[3] Kim et al.[4] described less than 50 cases of neurofibroma without neurofibromatosis in all age groups. According to Mukerji et al.[3] prior to 2007 the relevant literature describes only 5 pediatric cases of intramedullary schwannomas without neurofibromatosis. The sixth pediatric case of solitary intramedullary tumors was described in 2013 by B. Eljebbouri et al.[5] We present a rare, perhaps the seventh pediatric case of solitary neurofibroma of the cauda equina without neurofibromatosis.

CASE REPORT

An 11-month-old boy, born after a first normal pregnancy, was ill from the second month of birth with complaints involving fluctuating fever of 38°C, anxiety, crying, bilateral unsteadiness and paresis in the lower limbs, more expressed on the left side, urinary dysfunction and constipation. He was treated

for nephritis and viral infections. In the first months of her pregnancy the mother had an unspecified viral infection and received antibiotics. The general examination of the infant was normal, with no signs of neurofibromatosis. The neurological examination revealed lower paraparesis (right lower limb – 1st degree and the left– 2nd and 3rd degree), preserved muscle tone as well as the tendon reflexes and there was inability to stand up. Partial retention of urine and disturbances of defecation were found. The plantar, cremaster and anal reflexes were normal. The local examination of the spine didn't establish visual evidence of any changes in the region.

Laboratory tests including blood and urinalysis were within normal limits except for WBC with dynamics from 22×10^9/L to 31×10^9/L.

The search for a diagnosis of acute meningoencephalitis in the Department of Infectious Diseases lead to five unsuccessful attempts to collect a sample of the cerebrospinal fluid, i.e. so called "dry tap". Therefore, magnetic resonance imaging (MRI) was obtained, which demonstrated an advanced intraspinal tumor with heterointense characteristics of signals on T2-weighted images, localized in the cauda equina region – from the lower border of T11 to the lower border of L4, predominantly dorsal and more to the left side. The components of the thecal sac were dislocated to the right and the posterior part of the arachnoid space at the same segments was strongly and asymmetrically narrowed. Edema of the conus medullaris and inflammatory changes in the region of the right sacroiliac joint were established ***(Fig. 1)***.

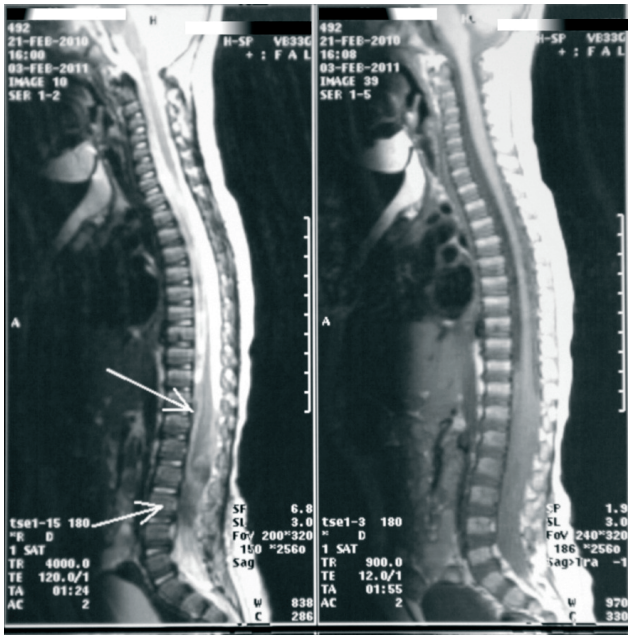

Figure 1. MRI of thoracolumbar spine.

The diagnostic methods proved that it was a case of intramedullary tumor which was most probably congenital, causing spinal cord compression as well as rapid progression of neurological deficit.

Surgical operation was the outcome measure. Laminectomy of T12, L1, L2 and L3 was performed, followed by extirpation of intradural, intramedullary and extramedullary spinal cord tumor - hourglass type in intramedullar part of the tumor. Decompression of spinal cord and cauda equina was achieved and finally the operation ended with plastic repair of dura mater (DuraGen® Dural Graft Matrix (US)) ***(Fig. 2)***.

The diagnosis of neurofibroma is based on histological preparations of the intraoperative specimens. Pathological Findings: Two independent histological analyses were done to double-check and confirm the diagnosis of neurofibroma. Immunohistochemistry revealed that some of the cells with granular cytoplasm demonstrated a strong expression of CD68 and were also highly positive on S-100. The lack of mitosis on the examined preparations as well as the low proliferative index (the Ki-67 labeling index) indicated a benign variant of the tumor.

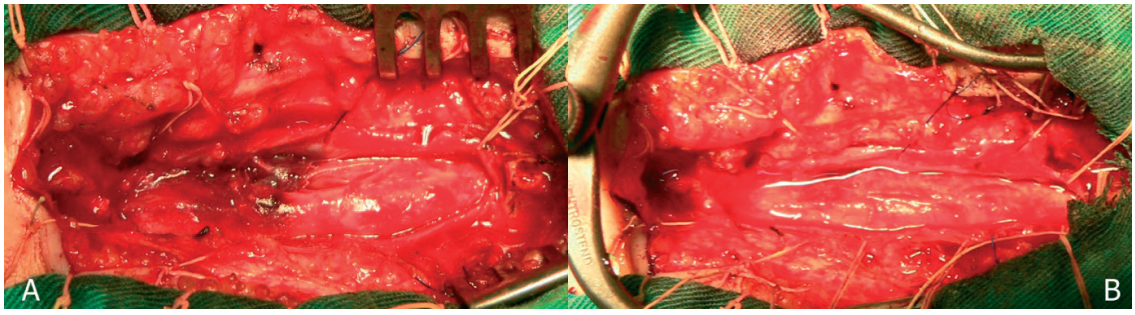

Figure 2. Intraoperative image of the tumor (A) and the same region after excision of the tumor (B).

The histological examination showed hyperplastic interfascicular connective tissue, pleomorphic cells, and tightly packed nerve fibers compressed by the surrounding loose connective tissue. There was hyperplasia of the vascular stroma ***(Fig. 3)***.

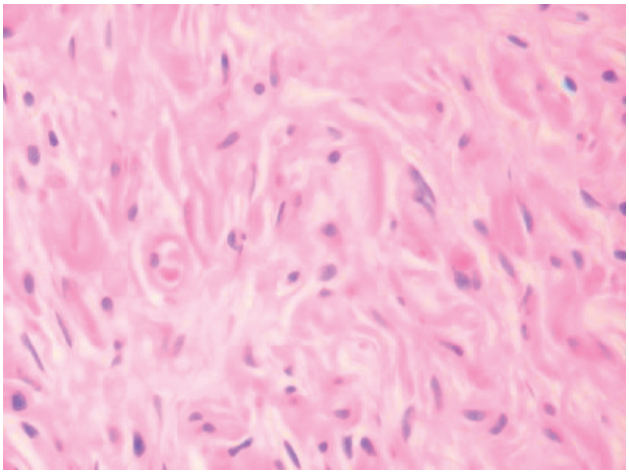

Figure 3. Microscopic studies showed uniformly distributed spindle cells with wavy nuclei, dispersed chromatin, and inconspicuous nucleoli, diagnosed as typical neurofibroma (H-E, ×200). No evidence of atypia or mitosis was present.

The postoperative period was smooth for the baby patient. The surgical wound healed by primary intention healing and all neurological deficits showed rapid regress with a petite paresis of the left lower limb.

Based on the clinical, MRI, histological and immunohistochemical features of the lesion, a diagnosis of solitary spinal neurofibroma was indicated. The child was followed-up for 5 years. At this point the patient is free of disease recurrence or progression.

DISCUSSION

Neurofibroma is the designation for a group of common, closely related benign nerve sheath tumors that, according to present evidence, have a similar molecular pathogenesis. These tumors typically present either as a localized lesion or as part of a generalized syndrome of neurofibromatosis generally known as neurofibromatosis type-1 (NF1) or von Recklinghausen disease. Intramedullary spinal neurofibromas are very rare. In the large Mayo Clinic series reported by Rasmussen et al.[6], no intramedullary lesion was present among 163 cases of spinal neurofibromas. Tonnis et al.[6] reported only one intramedullary case among 82 cases of spinal neurofibromas. Nittner[6] also found one intramedullary case among their 78 cases of spinal neurofibromas. By using PubMed, and to the best of their knowledge, Hidetaka et al.[6] found only 6 case reports of intramedullary neurofibromas. Intramedullary schwannomas are mostly located in the cervical spinal cord (51%), followed by the thoracic region (22%).[4] They may remain asymptomatic for a long time or cause nonspecific complaints, which makes the diagnosis difficult in children. The most common symptom is pain. Sometimes, they present with motor deficits, which can progress to the clinical syndrome, which includes motor deficiency, reduced sensation and loss of genitourinary function.[3]

The symptoms in our patient were similar including urinary dysfunction and constipation. We support the above data about the small number of cases reported in the literature and based on this research and the available evidence could say that a case like ours with lumbar localization is absolutely exceptional in the medical database. It is very likely that we are presenting the seventh case, but this is the first case we know of at this location and at this age.

Intramedullary spinal cord tumors in children account for 4-10% of central nervous system tumors, which is one of the reasons to misdiagnose them.[3] According to the available literature, solitary neurofibroma in the absence of neurofibromatosis cannot be ignored and we accept that although rare, the solitary form of spinal neurofibroma does occur.[1] Spinal neurofibromas are often asymptomatic. If symptoms are present, they usually include pain and/or radicular sensory changes due to the typical location along the dorsal sensory roots.[7]

It is an exception to associate symptoms in a newborn with a rare solitary neurofibroma and its spinal localization without neurofibromatosis 1 and we agree with Bisceglia[8] that the preoperative diagnosis of solitary fibrous tumors of nervous system is challenging, if not impossible. Immunohistochemistry is critical to establish a correct diagnosis.[8]

In our case, it was a real diagnostic challenge for an 11-month period. The discrete atypical symptoms and the fluctuating fever with unknown etiology were the cause of a lot of laboratory examinations and treatment with different antibiotics in the hospital. Several diagnoses seemed reasonable including intestinal infection, pyelonephritis, viral infections and undefined viral meningoencephalitis.

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In our case intraoperative diagnosis of neurofibroma enabled us to do a complete surgical removal that resulted in good recovery with noticeable improvement of his neurological symptoms. The patient is fully recovered for 3 years of monitoring. This case demonstrates the diagnostic difficulty and the good outcome of timely surgery.

CONCLUSION

Sharing cases of rare diseases when the differential diagnosis suggests a potential fatal outcome is important - if a proper approach is followed it will secure a certain recovery of the patient. Detection of unexplained symptoms like paresis in the limbs, urinary dysfunction, constipation and "dry tap", present in our case with spinal neurofibroma, should be thoroughly investigated and prompt the physician to consider the diagnosis and seek specialists' help. Although rare, this diagnosis should be considered when a child presents with a solitary intramedullary tumor, since its total resection can be achieved improving the surgical outcome.

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Проблемы диагностики солитарного нейрофиброма у мальчика одиннадцатимесячного возраста

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Спинальная нейрофиброма считается одной из самых редких неоплазм, поражающих спинной мозг или корешки и встречается значительно реже невриномы, менингиомы и глиомы. Шестой педиатрический случай солитарной интрамедуллярной опухоли был описан в 2013 году Б. Елджебурри и сотр. Нами представлен редкий, трудный для диагностики и вероятнее всего седьмой педиатрический случай солитарной нейрофибромы конского хвоста (cauda equine) младенца одиннадцатимесячного возраста. Пациенту была проведена ламинэктомия T12, L1, L2 и L3, экстирпация интрадуральной, интрамедуллярной и экстрамедуллярной опухоли спинного мозга. Пациент

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полностью восстановился в течение 5-ти летнего наблюдения. Независимо от редкого проявления, спинальные нейрофибромы у детей подлежат тщательному рассмотрению с точки зрения диагностики и радикальному лечению для достижения благоприятного исхода.