

Medullary Lesions in Erdheim Chester Disease: A Case Report

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Abstract

Case Report

ERDHEIM CHESTER disease (ECD) called also non-Langerhans Histiocytosis, is a rare illness, which can present various unusual clinical aspects. It is a Histiocytosis characterized by an infiltration of various tissues and organs, bones, retroperitoneum, pleuro-pulmonary sites, skin, and central nervous system. The most frequent central nervous system manifestations of ECD are diabetes insipidus, cerebellar syndromes, orbital lesions, and extra-axial masses involving the dura and here we report a rare case with spinal cord compression on the context of ERDHEIM CHESTER disease. Our work highlights another different clinical, radiological and pathological manifestation associated with ECD that should be considered.

Keywords: Erdheim Chester disease, medullary lesion, non-Langerhans Histiocytosis.

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INTRODUCTION

Erdheim-Chester Disease (ECD) is a rare form of non-Langerhans' cell histiocytosis. The multi systemic form of ECD is associated with significant morbidity, which may arise due to histiocytic infiltration of critical organ systems. Among the more common sites of involvement are the skeleton, central nervous system, cardiovascular system, lungs, kidneys (retroperitoneum) and skin [1]. The spinal cord lesion is rare, very few cases have been reported in the literature [7], here we present a case of ECD associated with spinal cord involvement including epidural lesions.

CASE REPORT

A 35 years old women patient with known diagnosis of ERDHEIM CHESTER disease, follow-up in department of clinical hematology from 2010, with pituitary lesion and Bone involvement, taking an interferon- α with a satisfying clinical amelioration, presented a complete spinal cord compression syndrome three months before her admission. Neurological examination showed paraparesis (2/5) with sensory level T6, and hyperreflexia both legs with bilateral Babinski responses.

The spinal MRI showed an epidural and dura mater infiltration by pathologic processes at T4 to T7 and from T12 to L2, with reel compression of the spinal cord, without vertebral involvement (Figure 1).



Figure 1: Medullary MRI demonstrating an epidural processes extending from T4-T7 and T12-L2 hypo intense on T2

The patient received laminectomy from T4 to T6 and from T12 to L2 with stabilization at the two sites. The epidural lesion was well visualized, pale, firm, and rubbery, readily dissected from the dura and removed partially.

The histological findings were compatible with spumous histiocytic infiltrating the spinal cord, with an aspect of round nucleus and foamy cytoplasm with positive expression for CD68 and negative for S100- and CD1a- compatible with ERDHEIM CHESTER histology (Figure 2).

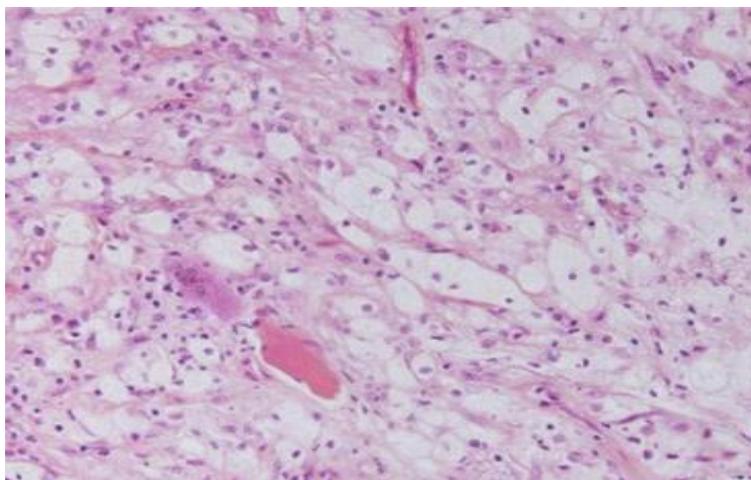


Figure 2: Histological appearance of proliferation of xanthomatous cells composed of foamy cytoplasm (foamy histiocytes). Foamy histiocytes are immunoreactive for (CD68) but not for S-100 and CD1a

The patient had an uneventful postoperative course. She was discharged to home in a stable condition on postoperative day eight, with discrete improved lower extremity sensation and strength. After a year of hard functional rehabilitation, she gained a few points in motor skills but could not walk.

DISCUSSION

Erdheim Chester disease (ECD) is an extremely rare and aggressive form of non-Langerhans cell histiocytosis. The more common sites of involvement are the skeleton, central nervous system (CNS), cardiovascular system, lungs, kidneys (retroperitoneum) and skin [1]. CNS involvement appears in approximately 30% of Erdheim Chester disease patients and accounts for 29% of all deaths, as reported by Arnaud *et al.*, [2].

Although it primarily affects adults between their 5th and 7th decades of life [3], some pediatric cases have been documented in the literature [4]. Its etiology is unknown but it is thought to be either a reactive or a neoplastic disorder [5]. Despite recent advancements, the pathogenesis of this disease is still poorly understood. The broad and complex manifestations of ECD, in conjunction with its rarity and with only a handful of centers of referral in the world may inevitably lead to misdiagnosis.

Diagnosis of ECD relies on established radiological and histological criteria; MRI is the modality of choice when evaluating the different CNS manifestations, Drier *et al.*, recommend performing systematically a cerebro-medullary MRI on ECD patients [6]. The histological diagnostic criterion is met providing that typical ECD histiocytes are found in the examined lesion. These histiocytes are non-Langerhans' foamy histiocytes, which lack Birbeck granules, nested within a polymorphic granuloma, fibrosis or xanthogranulomatosis. Immunohistochemical staining is positive for CD68 and negative for CD1a- and S100- [2].

The spinal cord lesion is rare [7, 8], at our knowledge it has been observed in only eleven patients including our case, most of them with a complete spinal cord compression syndrome. Furthermore, few reports of epidural lesions exist, and only six describe the characteristic MRI findings. Capparos-Lefebvre *et al.*, described a patient with a complete block extending from cervical spine to T12 [9, 10]. In our case, we had two block processes at T4 to T7 and from T12 to L2 [11]. Thus, ECD should be considered as a differential diagnosis of multiple sclerosis, gliomas, meningioma and other neurological diseases.

Table 1: Summary of reported patients with epidural spinal involvements of Erdheim-Chester disease

Author	Year	Presentation	Physical exam	Radiographic findings	T1-weighted MRI	T2-weighted MRI	Treatment	Outcome
Albayram <i>et al.</i> , [12]	2002	43-year-old man with 2 weeks leg weakness and 2 months upper back pain.	Gait spasticity and hyperreflexia both legs. Diminished sensation below T1 level.	MRI: Epidural and subdural soft tissue masses causing cord compression T1-T6.	Epidural lesion hypo intense to cord without enhancement. Subdural lesion isointense to cord.	Epidural lesion hypo intense to cord without enhancement. Subdural lesion hypo intense cord.	T1-T6 decompressive laminectomy	N/A
Allmendinger <i>et al.</i> , [13]	2010	87-year-old man with low back pain.	N/A	MRI: Epidural lesion of T10, L3, and S1 vertebrae.	Hypo intense.	Isointense.	Radiotherapy and interferon a. therapy.	Died 2months after diagnosis.
Babu <i>et al.</i> , [9]	1997	22-year-old man with 4 months feet numbness and gait difficulty	Spastic paraparesis (2/5) with sensory level T6.	MRI: Epidural mass extending from T5- T11.	N/A	N/A	T5-T12 laminectomy with radiation (8Gy).	Neurologically intact at 4 years.
Caparros-Lefebvre <i>et al.</i> , [10]	1995	74-year-old woman with paraplegi, ataxia and bladder incontinence.	N/A	CT scan: Large lesion in epidural space at L1.	N/A	N/A	N/A	Died of aspiration pneumopathy.
Curgunlu <i>et al.</i> , [14]	2003	43-year-old man with several months' progressive weakness, feet numbness, spasticity and muscle wasting lower limbs.	Weakness of hip, knee flexors, and knee extensors. Increased reflexes in both extremities. Normal sensory exam.	MRI : Multiple epidural masses between T1 -T9, T11-L1.	N/A	N/A	T1-T5 laminectomy and decompression, palliative therapy.	Recurrence and systemic progression.
Pego-Reigosa <i>et al.</i> , [11]	2000	50-year-old woman with 5 months paresthesia and weakness of the lower extremities.	Spastic paraparesis (4/5) and bilateral Babinski responses.	CT myelogram: Extradural mass at T3 and T9.	N/A	N/A	Surgical resection.	N/A
Shimada <i>et al.</i> , [8]	2007	47-year-old man with lesions of the spinal dura on autopsy.	N/A	N/A	N/A	N/A	N/A	Died 3.5 years after initial presentation of disease.
Takeuchi <i>et al.</i> , [15]	2012	56-year-old man with weakness of the left side and paresis.	N/A	MRI: Cervical intramedullary lesion.	Hypo intense	Hypo intense	Surgical resection.	N/A
Tzoulis <i>et al.</i> , [16]	2012	31-year-old woman 10 years of slowly progressive gait difficulty.	Spastic paraparesis with extensor plantar responses and normal sensory function.	MRI: Diffuse, noncontrast enhancing lesion cervical and thoracic vertebrae with intramedullary lesion T4-T6	Inhomogeneously hyperintense.	N/A	Interferon-alpha.	At 1.5 years after diagnosis, no signs of disease progress.
BrianY. Hwan [17]	2014	25-year-old woman with 3 weeks worsening bilateral lower extremity numbness and paresthesias and difficulty with ambulation	4+/5 strength lower extremities. T5 sensory level with - 50% decreased sensation to light touch.	MRI: enhancing T1 /T2-weighted Ventral epidural Mass at C5-T1 Compressing cord.	Hypo intense	Hypo intense	Surgical resection.	At 3 months, regained baseline neurologic function and returned to work.
This case report	2019	35 years old women, 3 months before her admission, weakness of two legs, and sensitive disturbance	Paraparesis (2/5) with sensory level T6, and hyperreflexia both legs with bilateral Babinski responses	MRI showed an epidural and dura mater infiltration by pathologic processes at T4 to T7 and from T12 to L2,	Hypo intense	Hypo intense	Laminectomy and fusion	After one year no recurrence

N/A = not applicable

Seven cases in the literature were described with a dorsal localization (Table 1); Spinal manifestations of ECD in our patient were at the dorsal and lumbar levels, described as linear infiltration of the dura and the anterior epidural space causing spinal cord compression in MRI, and our intervention allowed a decompression with fusion and direct histological examination with genetic search complement.

CONCLUSION

In summary, Erdheim Chester disease being multisystem disease, multidisciplinary approach would help in better patient care and management. Moreover, the medullary lesions should be considered in ECD manifestation even if is infrequent.

DECLARATIONS

ETHICS APPROVAL AND CONSENT TO PARTICIPATE: Not applicable.

AVAILABILITY OF DATA AND MATERIALS:

Data sharing not applicable to this article as no datasets were generated or analyzed during the current study.

COMPETING INTERESTS: The authors declare that they have no competing interests.

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AUTHORS' CONTRIBUTIONS

ZAKARIA CHANDIDE TLEMCANI: Conceptualization, Writing original draft, & editing. IMADEDINE SAHRI: review & editing. HOUSNI ABDERRAHMANE: Writing, review & editing. LAAGUILI JAWAS: Review & editing. Abad CHERIF EL ASRI: Review & editing. MILOUDI GAZZAZ: Supervision, Validation, & review. All authors read and approved the final manuscript.

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