

# World Journal of *Clinical Cases*

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### EDITORIAL

- 307 Adjuvants to local anesthetics: Current understanding and future trends  
*Swain A, Nag DS, Sahu S, Samaddar DP*

### MINIREVIEWS

- 324 Treatment of sepsis: What is the antibiotic choice in bacteremia due to carbapenem resistant *Enterobacteriaceae*?  
*Alhashem F, Tiren-Verbeet NL, Alp E, Doganay M*

### CASE REPORT

- 333 Vertebroplasty and delayed subdural cauda equina hematoma: Review of literature and case report  
*Tropeano MP, La Pira B, Pescatori L, Piccirilli M*
- 340 Pseudotumoral acute cerebellitis associated with mumps infection in a child  
*Ajmi H, Gaha M, Mabrouk S, Hassayoun S, Zouari N, Chemli J, Abroug S*
- 344 Atlanto-axial langerhans cell histiocytosis in a child presented as torticollis  
*Tfifha M, Gaha M, Mama N, Yacoubi MT, Abroug S, Jemni H*

## Contents

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## Atlanto-axial langerhans cell histiocytosis in a child presented as torticollis

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### Abstract

Langerhans cell histiocytosis (LCH) is a rare condition mostly seen in children and adolescents. Eosinophilic granuloma (EG) is one of its three clinical entities and is considered as a benign osteolytic lesion. Many reports of patients with spine histiocytosis are well documented in the literature but it is not the case of atlantoaxial localization. We report here a new observation of atlantoaxial LCH in a 4-year-old boy revealed by persistent torticollis. He was successfully treated with systemic chemotherapy and surgery. Inter-body fusion packed by autologous iliac bone was performed with resolution of his symptoms. It is known that conservative treatment is usually sufficient and surgery should be reserved for major neurologic defects in spine EG. In atlantoaxial lesion, surgical treatment should be frequently considered.

**Key words:** Langerhans cell histiocytosis; Eosinophilic granuloma; Torticollis; Cervical spine

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**Core tip:** Langerhans cell histiocytosis (LCH) is a rare condition mostly seen in children and adolescents. Eosinophilic granuloma is one of its three clinical entities and is considered as a benign osteolytic lesion. Many reports of patients with spine histiocytosis are well documented in the literature but it is not the case of atlantoaxial localization. We report here a new observation of atlantoaxial LCH in a 4-year-old boy



revealed by persistent torticollis.

Tfifha M, Gaha M, Mama N, Yacoubi MT, Abroug S, Jemni H. Atlanto-axial langerhans cell histiocytosis in a child presented as torticollis. *World J Clin Cases* 2017; 5(8): 344-348 Available from: URL: <http://www.wjgnet.com/2307-8960/full/v5/i8/344.htm> DOI: <http://dx.doi.org/10.12998/wjcc.v5.i8.344>

## INTRODUCTION

Langerhans cell histiocytosis (LCH) is an uncommon disorder characterized by an abnormal accumulation of histiocytes<sup>[1]</sup>. It includes three clinical entities namely eosinophilic granuloma (EG), Hand-Schüller-Christian syndrome and Letter-Siwe disease<sup>[2]</sup>. It consists in various clinical manifestations from a single lytic bone lesion to multisystemic lesions with organ dysfunction<sup>[3]</sup>. EG is a benign osteolytic lesion that commonly affects the skeletal system in a unifocal or multifocal form<sup>[2]</sup>. Atlantoaxial involvement by LCH is very rare<sup>[4,5]</sup>, especially in a very young child<sup>[2,6,7]</sup>. The localization makes it difficult to diagnose. Neural deficit in spinal EG can be observed representing a life threatening condition<sup>[2,6]</sup>. The management is still controversial. We present, herein an unusual and rare case of atlantoaxial LCH with infiltrative mass involving the dens of C2 resulting in torticollis as the first symptom lasting for 3 wk in a 4-year-old boy. EG is discussed and the literature is reviewed.

## CASE REPORT

### Clinical presentation

A 4-year-old boy without significant medical history was admitted for limited neck motion for 3 wk. The physical examination showed an irreducible torticollis with analgesic attitude of cervical spine. The active and passive mobilization of the neck was painful and no motor or sensory deficit was detected. The general condition of the patient was good, the clinical examination did not show a tumoral syndrome and the neurological examination as well as skin examination and laboratory tests were normal.

### Imaging features

The magnetic resonance imaging (MRI) of cerebro-spinal cord uncovered an infiltrative mass involving the dens of C2 which is hypointense on T1 sequence and hyperintense on T2 sequence, extending to the surrounding soft tissues leading to an increase in C1-C2 space, without compression of the spinal cervical cord. Complementary CT showed fragmented dens with important C1-C2 dislocation (Figure 1).

### Histologic features

The odontoid and mass biopsy was performed by endoscopic guidance. Histological features were

consistent with inflammatory EG. The positivity of the immunostain by the antibody anti Ps100 and the antibody anti CD1a confirms the diagnosis of LCH (Figure 2).

### Treatment and evolution

Initial treatment was started prednisolone 40 mg/m<sup>2</sup> per day orally, with weekly reduction starting from week 4 and intravenous Vinblastine 6 mg/m<sup>2</sup> per week for six weeks. An external immobilization by a cervical collar was maintained during the entire period of chemotherapy.

The evolution was marked by a decrease in pain secondary to the active mobilization of the neck with a persistent passive analgesic position. The control radiologic MRI showed a displaced horizontal fracture of the dens responsible for a posterior wall recoil reducing cervical occipital hinge without intramedullary signal abnormality. The infiltrative process had regressed in size (Figure 3).

A posterior cervical arthrodesis was performed and the spine was stabilized with a metal lacing associated with tricortical iliac crest graft interposed between the posterior arch of C1 and C2. No neuro-vascular complications have been detected.

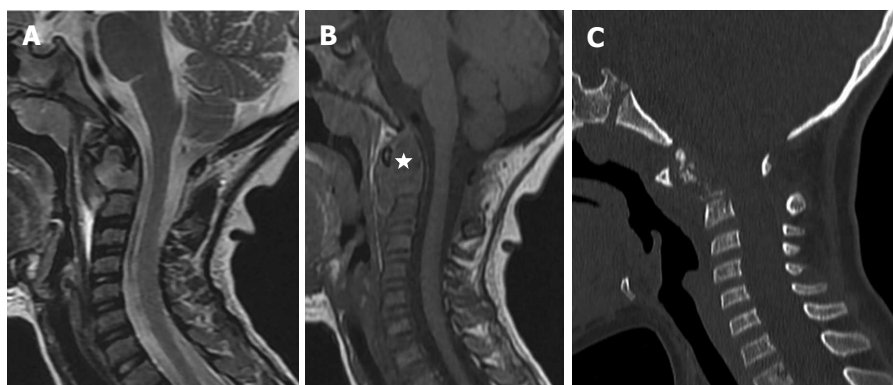
The patient is still under treatment consisting of prednisolone 40 mg/m<sup>2</sup> per day orally for five days in a week every four weeks and IV Vinblastine 6 mg/m<sup>2</sup> bolus every four weeks for 12 mo. Repeated CT scans revealed at 5 mo a consolidation of C2 fracture with moderate stenosis of the occipital hinge (Figure 4)

## DISCUSSION

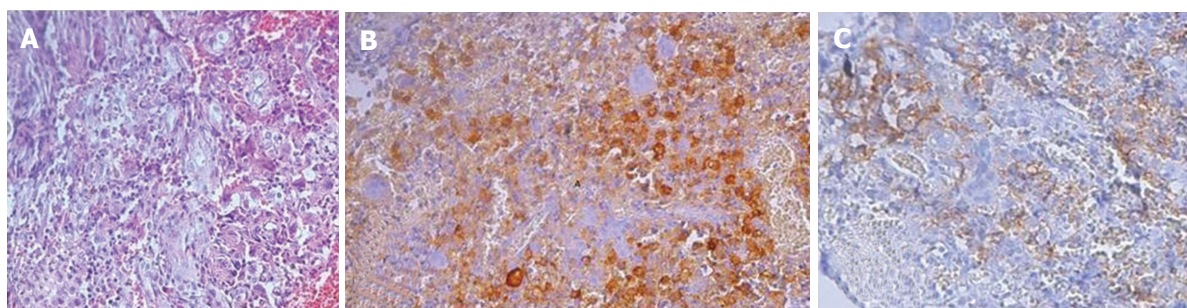
Spinal LCH commonly involves vertebral bodies, thoracic spine (54%) being the most common site of involvement followed by the lumbar (35%) and cervical spine (11%)<sup>[5]</sup>. Cervical vertebral involvement is exceedingly rare<sup>[6]</sup>. More than half of the cervical LCH lesions affect the C3-C5 vertebrae<sup>[4]</sup>. Atlantoaxial involvement by LCH is very rare<sup>[7]</sup>. Less than 15 cases have been reported in the literature. To our knowledge, it was the first Tunisian case of atlantoaxial LCH with odontoid process fracture reported in a 4-year-old child.

Our case present only torticollis as the first symptom, no other neurologic deficit was detected. In fact, pain, restricted range of motion or torticollis are the most common symptoms of cervical LCH<sup>[5]</sup>. However, the spinal destructive bony lesions etiology in children is extensive. Gaucher's disease, osteogenesis imperfecta, aneurysmal bone cyst, myeloma, tuberculosis, Ewing's sarcoma, osteogenic sarcoma, metastatic lesions, posterior fossa and cervical spinal cord tumors are part of the differential diagnosis of acquired torticollis with such bony lesions<sup>[5,7]</sup>.

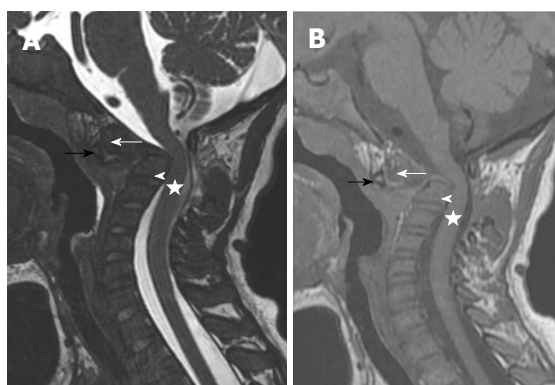
Loss of neural function is a rare occurrence with EG. It may result from vertebral collapse and impingement or, less frequently, from extradural extension of the lesion<sup>[5]</sup>. An asymptomatic case was also reported<sup>[2]</sup>.



**Figure 1** Initial cervical imaging: Sagittal FSET2 (A), SET1 magnetic resonance images (B) and Sagittal thin slice CT image (C). Infiltrative mass involving the dens of C2 hypointense on T1 and hyperintense on T2 sequence, extending to the surrounding soft tissues (star) leading to an increase in C1-C2 space. No compression of the spinal cervical cord. No signal abnormality nor rupture of the posterior longitudinal ligament spine. Complement CT showed fragmented dens with important C1-C2 dislocation.

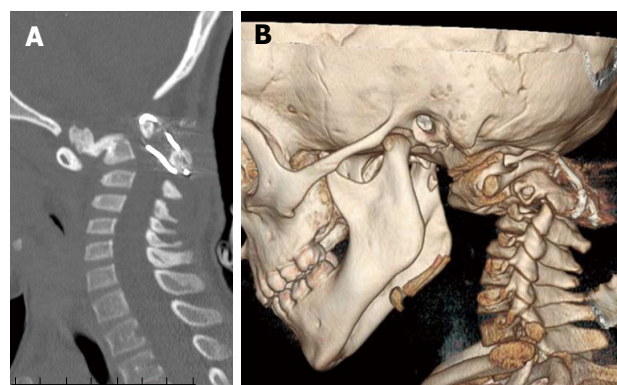


**Figure 2** Langerhansien histiocytosis histology. A: Inflammatory granuloma with eosinophils and histiocytes with circunvoluted nuclei; B: Positivity of the immunostain by the antibody anti Ps100; C: Positivity of the immunostain by the antibody anti CD1a.



**Figure 3** Six weeks follow-up after chemotherapy. Sagittal FSET2 (A), SET1 (B) magnetic resonance images: Displaced horizontal fracture of the dens responsible for a posterior wall recoil reducing cervical occipital hinge without intramedullary signal abnormality. Increase in the shrinkage of the cervical canal despite the regression of the infiltrative process. Black arrow: Anterior arch of C1; White arrow: The upper part of dens process; White arrowhead: The base of the dens; Star: Spinal cord.

Since the atlantoaxial lesion was the only one detected in our case, the biopsy of the spine lesion under endoscopic guidance established the diagnosis. If there were multiple lesions, the most accessible lesion would be the appropriate for biopsy to avoid open biopsy and prevent the possible vertebral growth plate damage<sup>[6]</sup>.



**Figure 4** Five months post-operative follow-up. Sagittal thin slice CT image (A) and volume rendering CT image (B): Consolidation of C2 fracture with moderate stenosis of the occipital hinge.

Our case fits into definitive diagnosis of LCH. The histological confirmation is required to establish LCH diagnosis<sup>[8]</sup>. The Writing Group of the Histiocyte Society identified three levels of confidence in the diagnosis of LCH. A definitive diagnosis, the third group, is established when histology is consistent with a diagnosis of LCH and the lesional cells are shown to express CD1a or to have intracytoplasmic Birbeck granules on electron microscopy<sup>[9]</sup>. Our patient presents a solitary spinal lesion. Spinal EG, in

the literature, is frequently associated with multiple skeletal lesion<sup>[3]</sup>. It is recommended that a technetium bone scan or a skeletal survey be performed early in the evaluation of every child with a suspected spinal lesion<sup>[3,5]</sup>.

Within the adult population, displaced type 2 odontoid process fracture can be treated operatively or non-operatively, depending on the patient age, co-morbidities, fracture pattern and displacement<sup>[2]</sup>. However, the management of such fractures in the pediatric population remains unclear especially in fracture spine due to LCH<sup>[10]</sup>. With such paucity of literature on this topic, it is unknown whether operative intervention associated with chemotherapy aid fracture union and functional outcome in young child.

In the present case, treatment consisted in chemotherapy (combination of oral prednisone and intravenous vinblastine). The effectiveness of this type of combination was demonstrated by LCH- I and LCH- II group in systemic LCH but the situation for spine EG remained unclear<sup>[1,9]</sup>. The infiltrative process has regressed in size with this association. The patient tolerated the treatment of chemotherapy with vinblastine well and showed no serious complications. The decision to continue treatment for one year was made.

Garg *et al*<sup>[3]</sup> report a spinal LCH in a child successfully treated without any chemotherapy use. The use of chemotherapy to treat solitary EG is still controversial, but it seems safe and effective in some studies. However, localized bone lesions with spontaneous regression are described<sup>[8]</sup>. The resolution occurred at a rate unaffected by the mode of treatment<sup>[6,8]</sup>.

Despite the absence of the observed neurologic deficit, the young age of the patient and radiologic assessment with displaced horizontal fracture of the dens and compromised spinal stability conducted to surgical excision followed by auto-graft fusion with satisfactory outcome in the present case. The use of surgery in such case is still controversial<sup>[1,3,6]</sup>.

Jiang *et al*<sup>[6]</sup> argue that mild neurologic deficit could be immobilized under strict observation and surgery should be reserved for major neurologic defects like myelopathy or monoparesis. The authors believe some intervention should be used to prevent possible LCH progress, especially in the C2 vertebral body. In this study, a protocol for the management of suspected LCH lesion of the cervical spine was suggested, the atlantoaxial LCH lesion is not included in this protocol<sup>[6]</sup>.

The radiotherapy was discussed but not applied for our patient. Some bone LCH lesions can be treated by radiotherapy alone<sup>[10,11]</sup>. However, a non successful radiotherapy performed on a 15-year-old girl presenting C1/C2 lateral LCH mass was reported<sup>[6]</sup>. Moreover, some authors support that radiotherapy might destroy the potential growth of the endochondral plates<sup>[6]</sup>. Further research on this topic is recommended.

Atlantoaxial LCH is rare. The diagnosis of the disease was made within a brief time limit with torticollis as the only clinic symptom. A delay in the diagnosis of this disease may lead to progressive neurological deterioration and increasing compression affecting largely the prognosis. Treatment modalities have changed over time depending on the clinical severity of the disease since it is quite varied. The combination of chemotherapy and surgical procedure seems to be effective in such lesion. This hypothesis needs to be improved from each other experiences.

## COMMENTS

### Case characteristics

A 4-year-old boy without significant medical history was admitted for limited neck motion for 3 wk.

### Clinical diagnosis

The physical examination showed an irreducible torticollis with analgesic attitude of cervical spine.

### Differential diagnosis

The spinal destructive bony lesions etiology in children is extensive. Gaucher's disease, osteogenesis imperfecta, aneurysmal bone cyst, myeloma, tuberculosis, Ewing's sarcoma, osteogenic sarcoma, metastatic lesions, posterior fossa and cervical spinal cord tumors are part of the differential diagnosis of acquired torticollis with such bony lesions.

### Laboratory diagnosis

Laboratory tests were normal.

### Imaging diagnosis

The magnetic resonance imaging of cerebro-spinal cord uncovered an infiltrative mass involving the dens of C2 which is hypointense on T1 sequence and hyperintense on T2 sequence, extending to the surrounding soft tissues leading to an increase in C1-C2 space, without compression of the spinal cervical cord. Complementary CT showed fragmented dens with important C1-C2 dislocation.

### Pathological diagnosis

The odontoid and mass biopsy was performed by endoscopic guidance. Histological features were consistent with inflammatory eosinophilic granuloma (EG). The positivity of the immunostain by the antibody anti Ps100 and the antibody anti CD1a confirms the diagnosis of langerhans cell histiocytosis (LCH).

### Treatment

Initial treatment was started prednisolone 40 mg/m<sup>2</sup> per day orally, with weekly reduction starting from week 4 and intravenous Vinblastine 6 mg/m<sup>2</sup> per week for six weeks. An external immobilization by a cervical collar was maintained during the entire period of chemotherapy.

### Related reports

Atlantoaxial involvement by LCH is very rare. Less than 15 cases have been reported in the literature. To our knowledge, it was the first Tunisian case of atlantoaxial LCH with odontoid process fracture reported in a 4-year-old child.

### Term explanation

LCH is an uncommon disorder characterized by an abnormal accumulation of histiocytes. It includes three clinical entities namely EG, Hand-Schüller-Christian syndrome and Letter-Siwe disease. It consists in various clinical manifestations



from a single lytic bone lesion to multisystemic lesions with organ dysfunction.

### Experiences and lessons

Atlantoaxial LCH is rare. A delay in the diagnosis of this disease may lead to progressive neurological deterioration and increasing compression affecting largely the prognosis. Treatment modalities have changed over time depending on the clinical severity of the disease since it is quite varied. The combination of chemotherapy and surgical procedure seems to be effective in such lesion.

### Peer-review

This is an interesting case report, which seems to provide readers with useful information. The manuscript is well written.

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