Acquired Torticollis as Presentation of Langerhans Cell Histiocytosis: Two Contrasting Cases

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Abstract
Langerhans cell histiocytosis (LCH) is a rare disorder that primarily affects young children. Because LCH can affect multiple organs, particularly bone and skin, it presents with a wide range of symptoms. This article reports two different cases with acquired torticollis as the primary complaint. Acquired torticollis is an atypical and less common presentation of LCH. These case reports highlight the importance of identifying a cause for acquired torticollis, which includes LCH in the differential diagnosis. Adequate diagnosis of LCH is important for timely initiation of appropriate treatment to prevent complications and long-term consequences.

Introduction

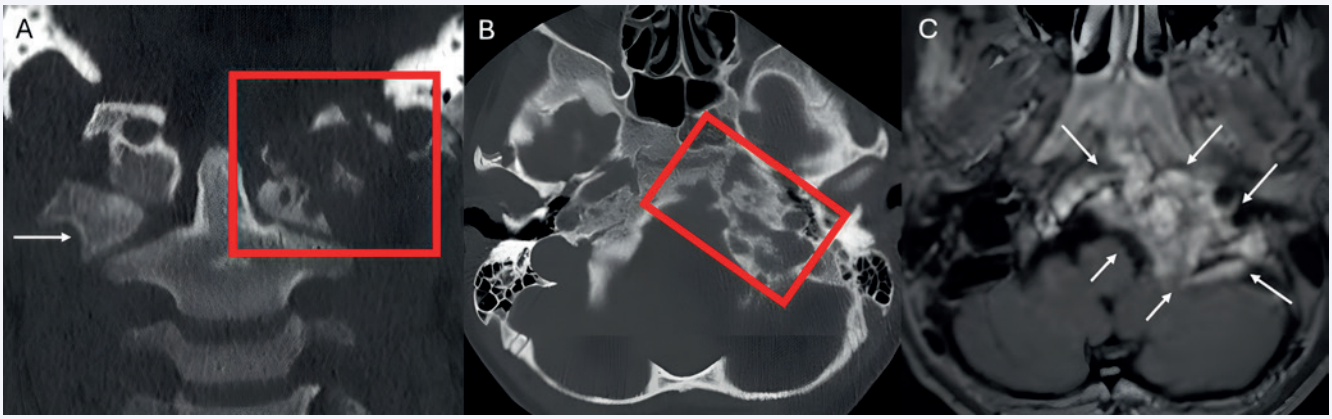
Langerhans cell histiocytosis (LCH) is a rare disorder, mostly affecting children aged one to three years old, but it can occur at any age. It involves pathological single- or multi-organ accumulation of immature myeloid precursor cells resembling Langerhans cells of the skin and mucosa. LCH affects about five children per million annually, but is likely underreported due to its diverse presentation, diagnostic challenges, and self-limiting localized forms (1).

LCH can be localized or multi-organ and most frequently affects the bones, skin and pituitary gland, but can also involve the lungs, lymph nodes, central nervous system (CNS), bone marrow, liver, or spleen (1, 2).

Bone is most commonly affected in children, especially the skull, resulting in pain or swelling (1). Skin lesions include seborrheic-like rashes, eczematous patches, papules, or mucosal ulcers (1, 2). Posterior pituitary involvement often leads to central diabetes insipidus, while anterior pituitary involvement is rare (1). Beyond these common presentations, other symptoms may include gastrointestinal, pulmonary, or lymphatic issues (2).

The presentation of LCH with torticollis as the primary sign is uncommon (3). Acquired torticollis, wich is often indicative of an underlying pathology, may result from trauma, nerve palsy, sternocleidomastoid spasm, infection and rotatory atlanto-axial subluxation (4). LCH is rarely considered in the initial differential diagnoses of torticollis.

FIGURE 1: CT skull base and cervical spine (**A,B**). Coronal reconstructions of the upper cervical spine (**A**) show lytic destruction of the left occipital condyle and the lateral mass of the atlas (red square) and a right sided atlanto-axial and –occipital lateral luxation (white arrow). Axial reconstructions of the skull base (**B**) show aggressive lytic destruction of the left posterior part of the clivus and the petrous apex of the mastoid (red square). On MRI axial T1-weighted images with Gadolinium (**C**) shows avidly enhancing soft tissue in the areas of bony destruction (*white arrows*).



This report describes two children with acquired torticollis who were later diagnosed with craniospinal LCH. Each case involved a unique mechanism of torticollis.

Case 1

The first patient is an eight-year-old boy with no notable clinical or family history. He initially presented with left-sided neck pain occurring after a fall, without torticollis. Two weeks later, he returned with a respiratory infection, accompanied by persistent neck pain. At this time, torticollis with lateral flexion of the neck to the right and painful left-sided lymphadenopathy were observed. No B-symptoms were present. Antibiotics, a cervical collar, and physiotherapy did not alleviate the pain or torticollis. Due to persistent torticollis, an x-ray and magnetic resonance imaging (MRI) of the cervical spine were performed, showing no abnormalities. Ultrasound revealed bilateral lymphadenopathy. Therefore, reactive torticollis caused by a respiratory infection with lymphadenopathy was retained as a working diagnosis.

However, two months later both the torticollis and lymphadenopathy persisted. At this point, our patient showed a lateral flexion of the neck to the right and an apparent rotation of the chin to the left. This position is known as 'cock robin posture', prompting further investigation into an atlanto-axial subluxation as the cause of the torticollis. Subsequently, a dynamic cervical computed tomography (CT)-scan was performed, which confirmed the subluxation and additionally showed significant osteolysis extending into the left occipital condyle, clivus and lateral mass of the atlas. A subsequent MRI demonstrated associated soft-tissue masses in the area of bony destruction, along with two additional comparable lesions in the right parietal bone. These radiologic findings are showed in Figure 1. Based on the location and radiologic characteristics multifocal LCH was suspected, with bone metastases of an unknown primary tumor as the main differential diagnosis. The diagnosis of a single-system multifocal LCH was confirmed by a CT-guided biopsy. Based on the confirmation of the diagnosis, a course of vinblastine and prednisolone was initiated, four months after the initial presentation. Subsequent maintenance therapy was continued for 24 months, resulting in a significant reduction of the lesions. Currently, three years after his initial presentation, the patient remains in clinical remission, with only radiological follow-up required

Case 2

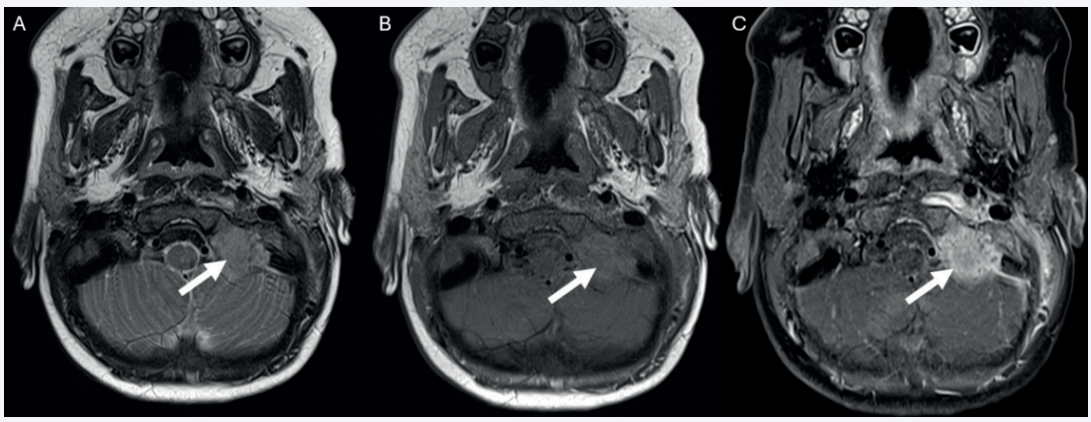
The second patient is a boy without significant medical history, who presented at the age of 19 months with a head tilt to the left. Clinical examination showed torticollis towards the left side, with both lateral flexion and rotation of the chin to the left, and right-sided cervical lymphadenopathy. Initial laboratory investigations showed a slightly elevated C-reactive protein (CRP) value, without any other abnormal findings. A CT-scan and MRI of the skull and neck were performed, showing an area of osteolysis at the level of the left posterior skull base and left occipital condyle with an accompanying soft-tissue component. The MRI-findings in this patient are showed in Figure 2. Additionally, an edematous aspect of the left half of the tongue was seen on MRI, likely correlated with a hypoglossal paresis. The diagnosis of unifocal LCH was ultimately confirmed by CT-guided biopsy. Although the lesion in the second patient was unifocal, systemic treatment with vinblastine and prednisolone was initiated due to the presence of torticollis and hypoglossal nerve compression. Given an adequate decrease in lesion size under this therapy, it was decided to discontinue chemotherapy six months after initiation and continue radiological follow-up. At present, nearly two years after the initial presentation, the patient remains in clinical and radiological remission, with near-complete resolution of the primary lesion and no ongoing treatment.

Discussion

Acquired torticollis is a potential manifestation of many different underlying conditions, and assessment requires simultaneous appreciation of the most common and most threatening differential diagnoses. Among the most common causes are an inflammation of the sternocleidomastoid muscle, neck trauma and acute infections such as upper respiratory tract infections or cervical adenitis. It is crucial to be aware of the life-threatening infections such as a retropharyngeal abscess or Lemierre syndrome, which necessitate prompt diagnosis and intervention. More infrequently, neoplastic diseases, such as CNS or bone tumors, as well as several neurological diseases such as myasthenia gravis can result in torticollis. In addition, torticollis can have ocular causes, such as superior oblique muscle weakness, strabismus and congenital nystagmus, and some miscellaneous causes, specifically gastroesophageal reflux, juvenile rheumatoid arthritis and drug-induced dystonia (4). Therefore, torticollis should not be considered a diagnosis, but rather a sign of an underlying condition.

With a prevalence of five children per million per year, LCH is a very rare disease (1). Bone lesions are the primary manifestation of LCH, with the skull being the most commonly affected region (61%) (3). Based on scattered case reports, only 4.7% of children with LCH involving the head and/or cervical spine present with torticollis (5).

FIGURE 2: MRI brain shows a T2-iso- to slightly hyperintense (**A**), T1-iso-intense (**B**), avidly contrast enhancing, (**C**) mass antero-inferiorly in the squamous part of the occipital bone extending into the left occipital condyle (*white arrows*).



The diagnosis of LCH is often suspected on the basis of imaging studies (typically CT or MRI) that are suggestive of the disease, but histopathological confirmation of the lesions is required for a definitive diagnosis. Due to the wide variety of organ systems involved, radiologic features vary widely. Histopathologic examination reveals uncontrolled proliferation of Langerhans-type cells, typically accompanied by numerous eosinophils, which led to the previous designation eosinophilic granulomas.

Generally, patients with unifocal single-system LCH can be managed with watchful waiting or with local therapies, as these patients usually have a good prognosis. For the multifocal osseous disease, systemic therapy is generally indicated, as these patients have a much higher risk of recurrence and progression. Systemic treatment in the pediatric population generally consists of a combination of prednisolone and vinblastine, as was the case in our two patients (6). Although the lesion in the second patient was unifocal, systemic therapy was chosen in the context of torticollis and hypoglossal canal compression with resulting hypoglossal nerve palsy.

We presented two patients with LCH-related torticollis, with distinct clinical manifestations. Our first patient presented with rotatory atlanto-axial subluxation as the underlying cause of the torticollis, a novel aspect reported only in one previous case report (7). Clinically, he presented with the typical 'cock robin posture' with flexion contralateral to the lesion and rotation of the chin to the side of the injury, due to a spasm of the contralateral sternocleidomastoid muscle (8). The major risk of missing an atlantoaxial dislocation is the progression to acute spinal cord damage (9).

In contrast, our second patient presented with a 'typical' torticollis, with flexion to the side of the lesion, likely due to a compressive effect of the lesion on the sternocleidomastoid muscle. However, the patient we describe is notably younger than most previously reported cases of torticollis associated with LCH.

Despite sharing the etiology of LCH, our patient's clinical differences have distinct implications for diagnosis and management.

Through this case report, we aim to emphasize that both typical and atypical torticollis can be initial symptoms of craniospinal LCH, regardless of age. Swift recognition and treatment are paramount to halt disease progression and prevent subsequent complications such as diabetes insipidus, growth retardation and secondary malignancies (10).

Moreover, in patients presenting with atypical features of torticollis, further investigation is mandatory, as we have demonstrated with the distinctive 'cock robin posture' as a hallmark of a rotatory atlantoaxial subluxation, a specific cause of torticollis that can be seen in bone lesions of LCH (7).

Conclusion

Langerhans cell histiocytosis (LCH) is a rare disease with a broad spectrum of clinical presentations. In this manuscript, we describe two patients who presented with torticollis as the primary sign of LCH, but there were distinct differences in clinical presentation. One patient presented with a 'cock robin posture' indicating a rotatory atlantoaxial subluxation, whereas the other patient presented with a typical torticollis as a result of compression of the sternocleidomastoid muscle. These cases illustrate that both typical and atypical torticollis can be the first symptom of craniospinal LCH. Therefore, LCH must be included in the differential diagnosis of all forms of acquired torticollis to ensure proper diagnosis, treatment and prevention of complications.

The authors have no conflicts of interest to declare with regard to the topic discussed in this manuscript.

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