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CASE REPORT

Recurrent torticollis secondary to Langerhans Cell Histiocytosis : A case report

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Torticollis is a common clinical sign encountered by pediatricians and orthopaedic surgeons in a wide spectrum of childhood conditions ranging from benign to life-threatening. We report the case of a child with recurrent torticollis caused by Langerhans Cell Histiocytosis (LCH). The patient was a 1-year-old boy with recurrent torticollis, followed by a painless swelling over the right temporal bone. The diagnosis was confirmed by an open biopsy of the calvarial lesion. As LCH is a very rare cause of torticollis it was not considered in the initial differential by the primary care physicians and the diagnosis was delayed about 4 months. The patient received chemotherapy with steroids and etoposide for 52 weeks. He showed complete regression of the sign and imaging tests at the end of treatment were normal. No relapse of symptoms occurred during a follow-up period of 2 years. The rarity of this disease as well as the site and form of presentation are emphasised to alert physicians for an early diagnostic evaluation, which is important to prevent neurological lesions and other late complications.

INTRODUCTION

Acquired torticollis, at any age, is not a distinct entity, but rather a clinical sign of an underlying disorder. Benign tumours that can involve the cervical spine should always be considered in the differential diagnosis of acquired torticollis. Although pathologically and physiologically benign, they can be clinically malignant if their surgical accessibility or risk of metastasis and recurrence places the neural structures at high risk (*11, 13*). The purpose of this report is to alert physicians to the possibility of benign entities such as LCH whenever a child is found to have torticollis.

CASE REPORT

A 1-year-old boy presented to the Paediatric clinic of our hospital complaining of cervical pain (painful cervical motion) and torticollis on the left side, of 3 days duration. His medical history revealed another episode of torticollis four months

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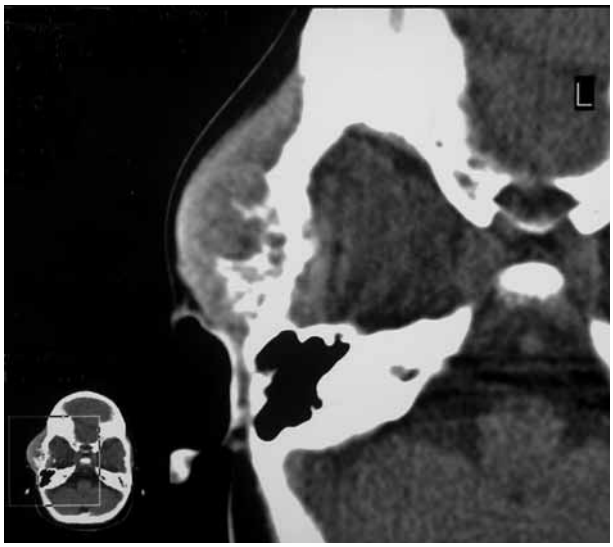

Fig. 1. — CT scan of the right temporal bone lesion

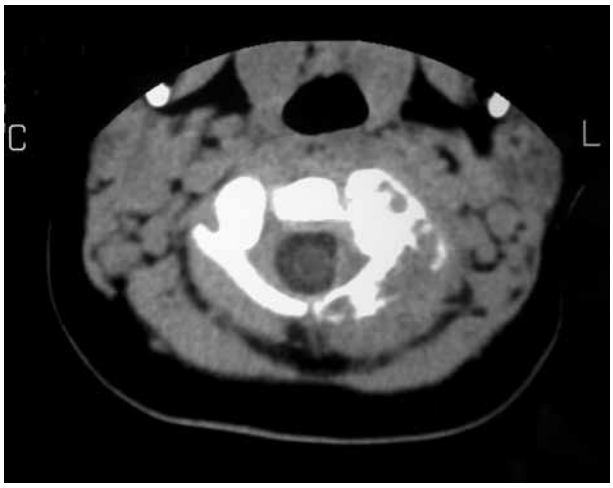

Fig. 2. — CT scan of the cervical spine showing the lesion in the C1 vertebra

before. At that time he presented to his pediatrician with fever, painful cervical motion and torticollis on the right side. That episode was attributed to inflammation of the adjacent neck tissues after an upper respiratory tract infection. He received an antibiotic for ten days and torticollis gradually disappeared.

There was no history of trauma, surgery in the head or neck or exposure to medication or drugs. On physical examination the diagnosis of torticollis was verified without evidence of craniofacial asymmetry or short neck with a low posterior hairline indicating a bony cervical spine anomaly. Ocular torticollis was excluded. The pharynx was free of signs of inflammation and retropharyngeal swelling. Enlarged lymph nodes in the neck and tenderness of the sternocleidomastoid muscle were absent. In contrast, point tenderness over the cervical spine indicated an underlying lesion at the C1-C2 region. A painless swelling was noted over the right temporal bony. He was afebrile and the respiratory and gastrointestinal tract were free of symptoms. The liver and the spleen size were within the normal ranges for his age. The neurologic examination showed no weakness or sensory deficits.

Blood cell count disclosed a white blood cell count of 9420/mm^2, haemoglobin 11.5 gr/dl and platelets 415000/mm^2. Liver and kidney function and serum electrolytes were normal.

Standard radiograph was difficult to obtain due to his age and the painful cervical motions. Computed tomography of the skull showed :

a) A solitary calvarial lesion of the right temporal bone that extended through both the inner and outer tables with soft tissue involvement (fig 1).
b) A spinal lesion in the arch of the first cervical vertebra which appeared wider. Involvement of the posterior elements such as the spinal cord and the epidural fat was obvious. The transition zone between normal and abnormal bone was poorly defined while reactive sclerosis was absent (fig 2).

To exclude the diagnosis ofneuroblastoma, an MIBG scan was performed and this was normal. An isotope bone scan showed increased scintigraphic activity in the right temporal bone and the left arch of C1. At the abdominal ultrasound the liver and the spleen were within the normal range for his age and the chest radiograph showed no pulmonary lesions.

An open biopsy of the temporal lesion was performed. The diagnosis of LCH was made on the basis of morphology and immunocytochemistry

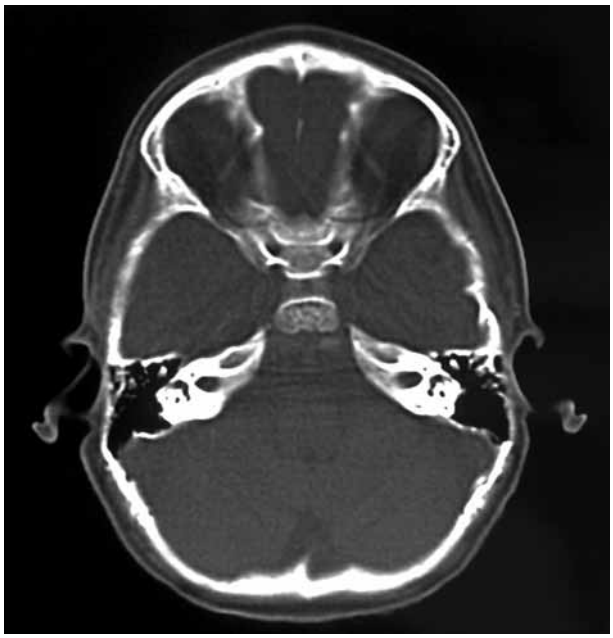

Fig. 3. — Calvarial CT scan two years later demonstrates complete healing of the temporal bone lesion.

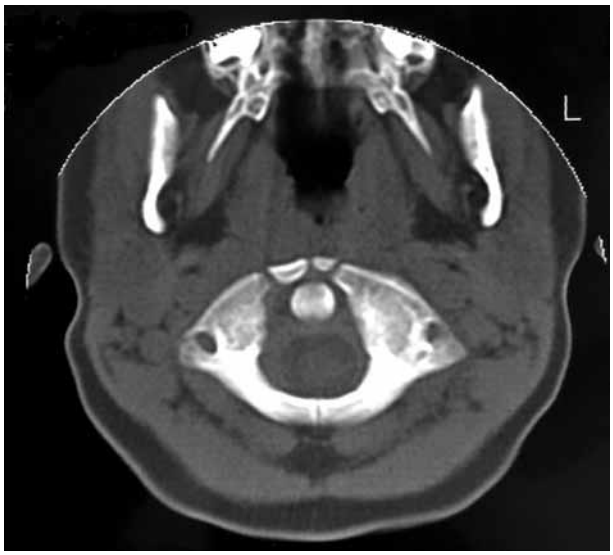

Fig. 4. — Cervical spine CT scan two years later demonstrates complete healing of the cervical spine (C1 vertebra) lesion.

including extensive staining with CD1a and S100. The diagnostic evaluation was completed with a skeletal survey, bone marrow aspirate and biopsy, and blood and urine osmolality. All the above parameters were normal.

According to LCH accepted criteria, our case characterised as single system disease-multiple site (multiple bone lesions) and as intermediate risk group (age < 2 years without organ dysfunction). He received initial therapy (1-8 week) with etoposide (150 mg/m^2 iv twice weekly, day 1 and 4 for the first 2 weeks, and 150 mg/m^2 once weekly for another 6 weeks) and dexamethasone (10 mg/m^2 daily for the first 2 weeks, 5 mg/m^2 daily for another 2 weeks, 2.5 mg/m^2 daily for another 2 weeks, 1.25 mg/m^2 daily for another week). Steroids were then tapered and stopped during week 8 followed by maintenance treatment (9-52 weeks) with etoposide 150 mg/m^2 iv, every second week (week 9-52) and steroids : dexamethasone pulses every second week, 10 mg/m^2 for 3 days, week 10-52. During both the induction and maintenance therapy he received trimethoprime-sulfomethoxazole at the dose of 15 mg/kilogram/day.

Our patient's response was evaluated according to the protocol predetermined intervals when the disease was judged to be either active or inactive. After 6 weeks of treatment, our patient achieved regression (active disease but improving) and was judged as responder. Re-evaluation at the end of therapy showed resolution (no active disease). Calvarial and cervical spine CT scan 2 years after diagnosis demonstrated complete healing of temporal and cervical lesions (fig 3, 4). On the last evaluation (3 years after diagnosis), the patient was free of symptoms.

DISCUSSION

Torticollis, the term used for the clinical finding of the head tilted toward a shortened sternocleidomastoid muscle and the chin rotated toward the opposite site, is a common clinical sign in children and may be congenital or acquired. In infancy, congenital causes such as muscular (congenital muscular torticollis), neurologic and bony abnormalities of the cervical spine predominate (*4*). In children, however, acquired torticollis due to trauma, infection and tumours is more prevalent (*2, 11*). The presentation of congenital and acquired torticollis is similar but in contrast to congenital

torticollis ; the child with the acquired type, is usually older, and the onset of torticollis is more acute.

Our patient's age at presentation, medical history, and physical examination findings excluded congenital torticollis and some causes of the acquired type such as trauma, upper respiratory tract infection, retropharyngeal abscess, myositis, cervical adenitis, ocular motility disorders, dystonic reaction, Sandifer syndrome and benign paroxysmal torticollis (*2, 11*).

Other rare causes of the acquired type, such as calcification of the intervertebral disk, vertebral artery abnormalities, cervical spondylodiscitis and benign or malignant tumours were also considered (*5, 6, 7, 8*). Imaging studies excluded the majority of them except from the benign tumours such as the Langerhans cell Histiocytosis (LCH) and the malignant counterpart metastatic neuroblastoma or Ewing sarcoma. Further evaluation with MIBG scan and an isotope bone scan almost excluded all the above malignant tumours while LCH diagnosis remained as more possible (*10, 13*). The diagnosis was confirmed with the histological evaluation of the temporal bone lesion. A granuloma of many types of haemopoietic cells such as T- and B-lymphocytes, neutrophils, eosinophils and "ordinary" (phagocytic) histiocytes was found enriched in dendritic cells of Langerhans type as characterised by positive staining for CD1a (*9*).

LCH is the most frequent form of childhood histiocytosis. Its frequency has been estimated to be around one case in 25,000 children per year.

In 1987 the Writing Group of the Histiocyte Society recommended a division of the histiocytic disorders in childhood into three classes : Langerhans cell histiocytosis (Class I), haemophagocytic syndromes (Class II) and malignant histiocytic disorders (Class III). Class IV comprises some very unusual forms, among which the less uncommon is the so-called sinus histiocytosis with massive lymphadenopathy (Rosai-Dorfmann disease) (*9*).

LCH may present as :

a) Single system disease-single site, single bone lesion, isolated skin disease, solitary lymph node.
b) Single system disease-multiple site, multiple bone lesions, multiple lymph node involvement
c) Multisystem disease, multiple organ involvement, with or without organ dysfunction (*1*).

Untreated LCH can have a variable course resolving spontaneously or disseminating with grave if not fatal course. Age and organ dysfunction are strong prognostic indicators (*3*).

A good risk group was defined as older patients without organ dysfunction. The second was an intermediate risk group under 2 years of age without organ dysfunction and the poor risk group was the third, made up of similarly young patients but with organ dysfunction (*3*).

According to the above criteria, our patient was classified in the intermediate risk group (age < 2 years with a single system-polyostotic disease).

Osseous lesions have been observed in 80-100% of cases in various series of LCH patients either in isolation or as a part of multisystem disease. The skull, the long bones, and then the flat bones are most frequently involved. The feature and possible complication(s) of the osteolytic lesion of LCH remain local : for instance, when the ear region is involved, the mastoid bone appearance may mimic mastoiditis, periorbital involvement may lead to proptosis, and vertebral LCH, as in our case, produces bony collapse with painful swelling causing torticollis.

Some skull lesions, as in our case, are not only lytic but may have an accompanying mass that impinges on the dura. The lesions of the facial bones or anterior or middle cranial fossa with intracranial tumor extension are part of a CNS-risk group, which has a threefold increased risk for developing diabetes insipidous and an increased risk of other nervous system (CNS) disease (*1*).

According to the new LCH-III study, for patients with multiple bone lesions belonging to "CNS-risk" group as described above, a short treatment course with only a single agent (e.g., prednisone) is not sufficient, and relapses commonly occur. Treatment with vinblastine and prednisone for 6 to 12 months is recommended.

Our patient received etoposide and prednisone according to the treatment protocol of the "First

International HLH Study 1994". He also received trimethoprime - sulfomethoxazole. This antibiotic combination has been used successfully at the dose of 12 to 15 mg/kg for 1 to 3 months in cutaneous disease and children with single-system disease, whereas children with a multisystem disease had a variable response (*12*).

According to the Histiocyte Society criteria for active or inactive disease on each response definition, our patient has the better response, having continuous regression of the disease with complete resolution at the end of therapy.

CONCLUSION

Acquired torticollis, at any age, is not a distinct entity, but rather a symptom that can be related to different pathological mechanisms ranging from simple to life-threatening conditions. Among other possible causes, Langerhans cell histiocytosis should be considered in the differential diagnosis of acquired torticollis. Imaging studies at the time of presentation often reveal additional lesions of the underlying disease, and are very helpful for the diagnosis and the treatment plan without any delay.

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