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Case Report

Radiology Section

Acquired Chiari Type 1 Malformation Secondary to Paget's Disease of the Bone: A Case Report

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ABSTRACT

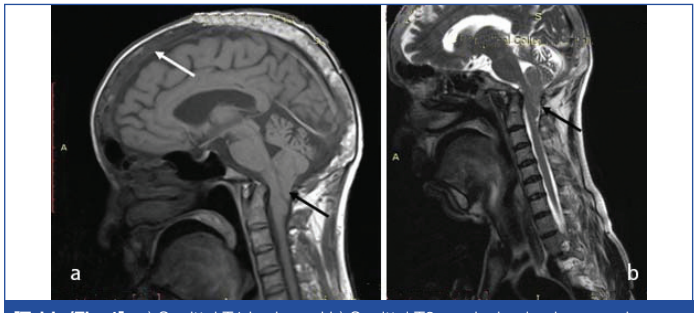
Chiari type 1 malformations are characterised by caudal descent of cerebellar tonsil into foramen magnum. Usually it is congenital. Paget's disease causing acquired chiari type 1 malformation is rare with only five reported cases in the literature. The diagnosis is primarily by Magnetic Resonance Imaging (MRI). Authors reported a case of acquired chiari type 1 malformation due to paget's disease in a 58-year-old male patient diagnosed by MRI and Computed Tomography (CT). The patient presented with complaints of headache, unsteady gait, slow movements with weakness in all the four limbs, difficulty in walking which aggravated since three days and stammering of speech. Initially a diagnosis of motor neuron disease was suspected and the patient was subjected for MRI examination of brain and spine. MRI revealed platybasia with cerebellar tonsillar herniation with calvarial thickening and multiple vertebral collapse. The CT of brain and spine was done to evaluate bones specifically in view of thickened cranial vault and multiple vertebral collapse on MRI which revealed features of paget's disease in the form of widened sclerotic bone with lytic areas and thickened trabeculae. Hence, diagnosis of paget's disease causing acquired chiari type 1 malformation was made which correlated with biochemical findings. The management is different in primary and acquired chiari type 1 malformation and hence it is important to diagnose this condition which is relatively rare.

Keywords: Cerebellar tonsil herniation, Computed tomography, Magnetic resonance imaging

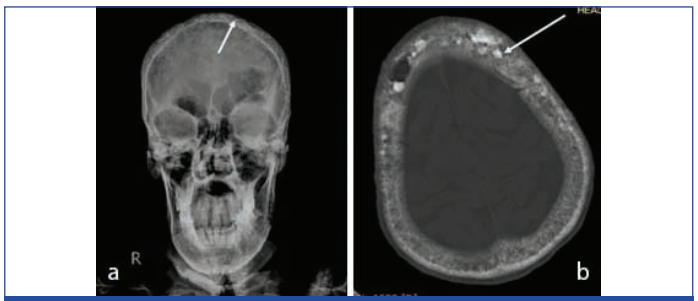
CASE REPORT

A 58-year-old male patient, presented with complaints of headache, unsteady gait, slow movements with weakness in all the four limbs, difficulty in walking for three months which aggravated since three days associated with stammering of speech. There was no significant past medical, surgical or family history. No evidence of increasing head circumference was noted. The symptoms were progressive in nature. Since, there was asymmetrical weakness with wasting and brisk reflexes, clinically, a diagnosis of motor neuron disease was made as motor neuron disease usually presents with these symptoms and brisk reflexes.

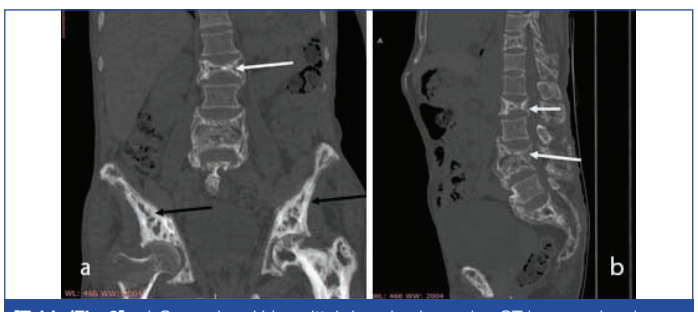
The patient was referred to the Radiology Department for Magnetic Resonance Imaging (MRI) of brain and spine. MRI of brain showed platybasia with cerebellar tonsillar herniation for about 10 mm below foramen magnum causing severe compression of cervical spinal cord [Table/Fig-1a,b]. The cranial vault looked abnormal on MRI and appears thickened [Table/Fig-1a]. Whole spine MRI showed no evidence of syringomyelia with multiple vertebral collapse. The patient did not gave any history of trauma. Computed Tomography (CT) of brain and spine was done to evaluate bones specifically in view of thickened cranial vault and multiple vertebral collapse on MRI. X-ray skull showed typical cotton wool appearance of skull [Table/Fig-2a]. The CT showed widened diploic space of skull [Table/Fig-2b] with multiple lytic-sclerotic lesions in most of the dorsolumbar vertebrae, pelvic bones and both femur with thickened trabeculae and expanded bones [Table/Fig-3a,b]. All these features and imaging findings were suggestive of paget's disease causing acquired chiari type 1 malformation. Blood investigations revealed elevated serum alkaline phosphatase (1400 IU/L) with normal calcium (9.2 mg/dL) and phosphate 3.8 mg/dL levels. The red blood cell count (5.7 million/μL), white blood cell count (7.2×10^9/L), haemoglobin levels (13 gm/dL), serum creatinine (0.9 mg/dL) and blood urea levels (18 mg/dL) were normal. The patient was given options of surgical management and since the patient refused,



[Table/Fig-1]: a) Sagittal T1 brain and b) Sagittal T2 cervical spine images shows herniation of cerebellar tonsil into foramen magnum compressing the cervical spinal cord (black arrows) with expanded cranial vault (white arrow).



[Table/Fig-2]: a) Plain X-ray Skull Anteroposterior view showing cotton wool appearance of skull (white arrow); b) Axial CT sections bone window of brain showing expanded diploic space with lytic and sclerotic areas (white arrow).



[Table/Fig-3]: a) Coronal and b) sagittal dorsolumbar spine CT images showing expanded bone with sclerotic areas involving pelvic bones (black arrows) with multiple compression fractures of dorsolumbar vertebrae (white arrows).

he was treated with bisphosphonates. Alendronate 40 mg daily for three months was given. On follow-up after three months, the serum alkaline phosphatase (600 IU/L) level has reduced with mild improvement in patient headache and unsteady gait. There were no adverse effects of bisphosphanates noted on follow-up.

DISCUSSION

Paget's disease causing acquired chiari type 1 malformation is rare with only five reported cases in the literature [1-5]. Paget's disease of the bone is a chronic skeletal disorder characterised by abnormal bone resorption and apposition with excessive bone remodelling. It can be asymptomatic or can cause various complications some of which can be debilitating [5]. The aetiology of the disease is largely unknown although there are theories of paramyxovirus infection.

There are three phases described in this disease which are lytic phase, mixed phase and blastic phase. In lytic phase osteoclastic resorption predominates, in mixed phase both osteoclastic and osteoblastic activity occurs with predominant osteoblastic activity and finally blastic phasic also called late inactive phase in which the osteoblastic activity declines [5]. Imaging plays essential role in the diagnosis of paget's disease of the bone. The common bones involved are lumbar vertebra, pelvic bones, femur and skull. Involvement of cranium can cause increasing head circumference due to bone expansion and remodelling, hearing loss, cranial nerve involvement, platybasia, basilar impression [5]. Our case had platybasia, basilar impression and expansion with sclerosis of cranial vault. Spine involvement can cause compression fractures like in our case, spinal canal stenosis, nerve root compression and kyphosis.

Chiari type 1 malformation refers to herniation of cerebellar tonsil below foramen magnum due to congenital malformations of posterior fossa. Recently there are many case reports describing chiari type 1 malformation acquired due to various causes. Chiari type 1 malformation due to paget's disease is extremely rare with only five reported cases in the literature to the best of authors knowledge [1-5]. This is the sixth reported case and first Indian case. The cause for chiari type 1 malformation due to paget's disease is due to softening of base of skull leading to platybasia, basilar impression with crowded posterior fossa because of bone overgrowth leading to herniation of cerebellar tonsil [1-3]. In the present case report the patient presented with symptoms of chiari type 1 malformation like headache, speech difficulty, progressive weakness of all four limbs and unsteady gait. Gait disturbance was common in 3 of the 5 reported cases like in our case with one patient presenting with only increasing head circumference as the complaint and other with otoneurological symptoms [1-5]. Among the previously reported patients, only one patient had evidence of syrinx [3].

Various causes for acquired chiari malformations have been described in the literature [6-12]. Broadly, it can be classified into three which are conditions which reduce the volume of cranial cavity like paget's disease, rickets, craniosynostosis, hyperostosis of skull, erythroid hyperplasia, conditions which increase the volume of intracranial contents like intracranial space occupying lesions, hydrocephalus, intracranial haemorrhages and conditions associated with Cerebrospinal Fluid (CSF) leak due to lumbar puncture, intracranial hypotension, post-traumatic, post-surgery and lumbar shunting [12]. Sometimes spontaneous intracranial hypotension likely due to spontaneous CSF leak also can cause acquired chiari type 1 malformation [12].

Surgical treatment like suboccipital decompression is the treatment of choice for congenital chiari type 1 malformation. However, acquired chiari type 1 formations are treated depending on the cause since so many conditions produce secondary chiari type 1 malformation [13]. Hence, imaging plays an important role in diagnosis and treatment. In the present case the ideal treatment would be craniectomy to reduce the intracranial tension however our patient refused surgical management. Hence, the only other option available for this patient was to treat the primary cause which is paget's disease with bisphosphonates.

CONCLUSION(S)

Chiari type 1 malformation due to paget's disease is extremely rare with only few reported cases. MRI is particularly helpful in establishing the diagnosis. Proper diagnosis of this entity is important as the treatment differs between primary and acquired chiari type 1 malformations.

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