Special Issue Article "Craniosynostosis"

Case Report

Lambdoid Diastasis Subsequent to Multisuture Craniosynostosis and Intracranial Hypertension in Osteopetrosis

Amirhosein Nejat, Zohreh Habibi and Farideh Nejat*

Department of Neurosurgery, Children's Medical Center, Tehran University of Medical Science, Tehran, Iran

ARTICLE INFO

Received Date: June 25, 2020
Accepted Date: August 10, 2020
Published Date: August 12, 2020

KEYWORDS

Osteopetrosis
Craniosynostosis
Diastasis
Lambdoid

Citation for this article: Amirhosein Nejat, Zohreh Habibi and Farideh Nejat. Lambdoid Diastasis Subsequent to Multisuture Craniosynostosis and Intracranial Hypertension in Osteopetrosis. SL Clinical Medicine: Research. 2020; 3(1):117

Corresponding author:
Farideh Nejat,
Department of Neurosurgery, Children's Hospital Medical Center, Tehran University of Medical Sciences, Iran, Tel: +98-912-1494064; Fax: +98-21-66930024;
Email: nejat@sina.tums.ac.ir

ABSTRACT

Osteopetrosis is a rare disorder which is characterized by failure of osteoclasts to resorb bone minerals leading to thick and dense but fragile bone. Craniosynostosis is rare in patients with osteopetrosis which aggravates the severity of skull deformity and intracranial hypertension in these patients. Association of craniosynostosis with diastasis of open suture is uncommon. Here we report two patients with osteopetrosis and multisuture craniosynostosis who had lambdoid suture diastasis. The radiologic feature and the cause of this rare structural malformation of skull are discussed.

INTRODUCTION

Osteopetrosis is a rare autosomal recessive disorder which involves bone remodeling by increasing the thickness and density of cranial bones especially in the skull base. This disease is characterized by the failure of osteoclasts to resorb bone minerals with impaired bone building and remodeling. Because of an osteosclerotic bone abnormality the entire skeleton becomes dense which is the radiologic hallmark of this disease. The condition has several subgroups which are different in their onset of symptoms, severity and associated extra-skeleton manifestations [1]. Craniosynostosis, or early fusion of one or more cranial sutures, is a rare abnormality that can be found in osteopetrosis with thick skull. A rare feature of osteopetrosis can manifest as a disproportionate skull shape due to associated metabolic disorder [2]. Here, we report two children with osteopetrosis, multi-suture synostosis, and lambdoid suture diastasis who were referred to us for the progressive pulsatile bulging of occipital area. The radiologic characteristics and the cause of this rare structural malformation of skull in these patients are described.

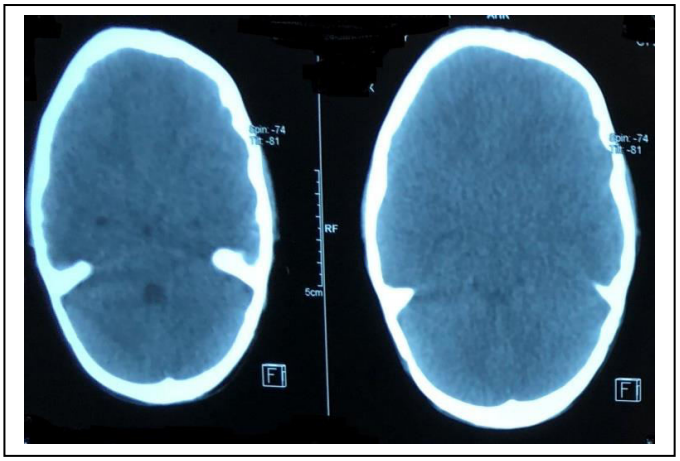
CASE PRESENTATION

Case 1

A 6-year-old boy was referred to our clinic due to headache and visual loss, which was progressive during last few months. He was the first child of a healthy couple who had noticed his blindness since last year, after then the diagnosis of osteopetrosis was made during evaluation of blindness with detection of abnormal skull thickness and optic foramen stenosis. The parents noticed a strip of soft tissue at posterior part of his skull about 6 months ago. The patient had no light perception with optic atrophy in ophthalmological evaluation but his general condition, mental state and motor exam

were otherwise normal. His skull was elongated anteroposteriorly in a dolichocephalic shape with head circumference of 56cm. There were some engorged dilated veins in the scalp. There was an about 2-cm diastasis of both lambdoid sutures along with a pulsatile posterior fontanel in touching the diastatic part.

Brain Computed Tomography (CT) showed typical findings of osteopetrosis including thickening of the cranial vault, foraminal stenosis, and non apparent paranasal sinuses and mastoid air cells. No intra-parenchymal abnormality was found except for sulcal effacement due to high intracranial pressure. Increased skull thickness was associated with closure of all sutures except for both lambdoid which developed diastasis (Figure 1).



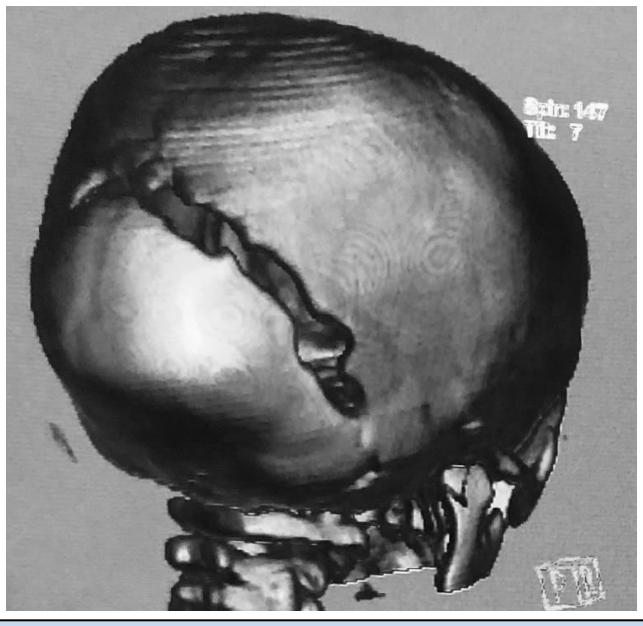


Figure 1: Brain CT scan of patient one shows thick skull with sulcal effacement (A) and closure of all sutures except for lambdoid that found obvious diastasis (B).

The patient underwent multiple strip craniotomy (including one frontal and three biparietal strips) and skull reconstruction. Dura mater was very tense but after craniotomy became relaxed. Calvarium was thick especially at vertex, with decreased internal diameter. With drilling and finally bone flap fixation and reconstruction, total cranial volume was increased [3]. Headache improved within a few weeks during post-operative period and no new problem was found in 3 year follow up. Brain CT scan 6 months after surgery revealed improved sulcal effacement and decreased severity of diastasis (Figure 2).

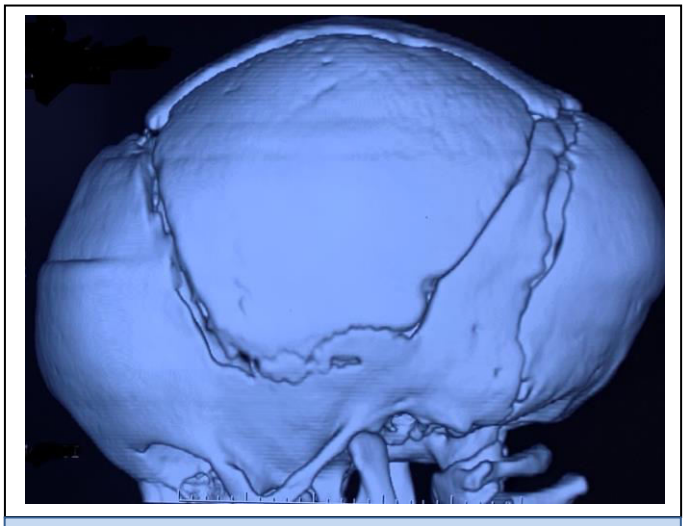


Figure 2: Postoperative 3D CT scan shows the incisions of craniotomy and confirms decreased severity of diastasis.

Case 2

A 5-y-old girl with osteopetrosis was admitted in our ward due to headache, occipital pulsatile area and progressive visual loss. The condition temporarily improved after ventriculoperitoneal shunting for hydrocephalus but was followed by exacerbated headache and visual blurring following shunt failure. Her younger brother had similar history of osteopetrosis associated with blindness and craniosynostosis who was operated by expansile craniotomy. Physical examination revealed papilledema and bulging of occipital area and a pulsatile band of soft tissue at lambdoid suture. Head circumference was 44 cm which ranked below the 3rd percentile for age.

Brain CT scan showed thick calvaria, metopic and bicoronal craniosynostosis, and sulcal effacement associated with

Lambdoid Diastasis Subsequent to Multisuture Craniosynostosis and Intracranial Hypertension in Osteopetrosis. SL Clinical Medicine: Research. 2020; 3(1):117.

widening of lambdoid suture (Figure 3). Optic foramina were still open on three-dimensional reconstructed CT.

The patient underwent the same craniosynostosis surgery by expansile craniotomy as case 1to increase total cranial volume. The symptoms improved in a few weeks after surgery and the vision remained stable during 2 year follow up. She was planned for bone marrow transplantation to stop disease progression and avoiding further foraminal stenosis to save vison.

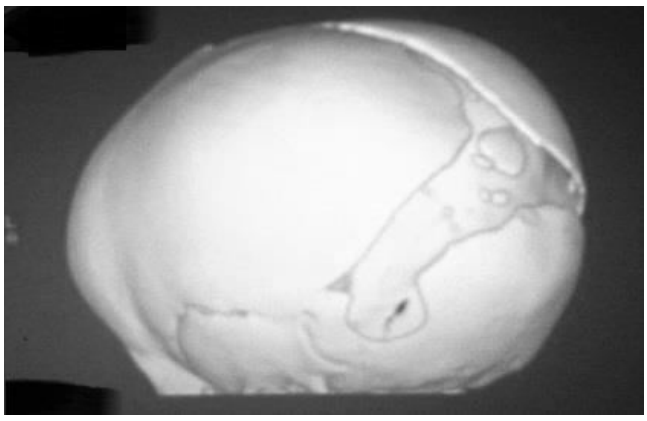


Figure 3: Brain CT scan of patient two confirms closure of metopic and bilateral coronal sutures associated with lambdoid diastasis.

DISCUSSION

Craniosynostosis is a relatively common disorder in pediatric neurosurgery practice (1 in 2500 live births) which can involve one or more sutures [4]. Multi-suture synostosis, which can be seen in both syndromic and non-syndromic craniosynostosis, is more associated with high intracranial pressure compared to single suture synostosis. Skull deformity can be caused by decreased growth around closed sutures and compensatory growth of open sutures to amend intracranial hypertension. Bicoronal synostosis is the most common suture involved in syndromic craniosynostosis but in our cases metopic and sagittal sutures were also involved [2,5]. The common features of the involved calvarium in multi-suture synostosis include skull remodeling, thinning of bone, silver beaten appearance, acrocephaly, microcephaly and even focal acquired encephalocele subsequent to brain herniation through the skull defect secondary to severe bone thinning [2,4]. Diastasis of open sutures is not a known feature observed with multi-suture craniosynostosis.

There are three discrete clinical classes of osteopetrosis including infantile, intermediate, and adult onset types which are classified according to age and clinical features. Benign osteopetrosis is an inherited autosomal dominant disease in adult with good long-term survival. Infantile and intermediate types are autosomal recessive. Infantile type usually results in death during the first decade of life due to bone marrow suppression and subsequent severe anemia, bleeding diathesis, infection and growth retardation. Bone marrow transplantation is the treatment of choice in this subtype [1,6].

Osteoclast dysfunction in osteopetrosis and impairment in bone modeling and remodeling lead to more brittle bone in spite of making thick bone. Increased thickness and bone mass specially in cranial base cause diverse degree of neurological deficit due to cranial nerve compression including deafness, visual loss, facial nerve paralysis and venous drainage impairment resulting in increased intracranial pressure and subsequent hydrocephalus or pseudotumor cerebri [1,7].

Although osteopetrosis is a disorder comprising variable molecular abnormalities and clinical features, all forms have osteoclasts dysfunction. But if symptoms occur in a child, it is severe and concomitant with many complications and morbidities [1,2]. The two children reported here had the intermediate type with an odd presentation of lambdoid diastasis in craniosynostosis and associated high intracranial pressure. High intracranial pressure occurs subsequent to imperfected venous drainage of the brain at level of foramen of jugular and decreased intracranial capacity due to excessive thickening of skull [3,7]. Craniosynostosis with involvement of several sutures and limiting the normal growth of skull is another cause of intracranial hypertension in these cases.

Any open suture provides cranial growth in the vertical axis to that suture to compensate the growth restriction along the nearby closed suture, but diastasis instead of skull growth is a rare phenomenon in compensatory response of non-synostotic sutures [3]. Diastasisis not routinely observed during compensatory growth of cranium in multi-suture craniosynosotosis of non metabolic origin [2]. Lambdoid diastasis in two patients with osteopetrosis in which bone formation and resorption do not occur in a normal manner might be subsequent to metabolic disorder in these children .It

Lambdoid Diastasis Subsequent to Multisuture Craniosynostosis and Intracranial Hypertension in Osteopetrosis. SL Clinical Medicine: Research. 2020; 3(1):117.

can be assumed that a proportion of high intracranial pressure in these two cases would occur in the metabolic problem of underlying disease, due to decreased intracranial volume caused by thick calvarium and smaller internal diameter of skull. This condition together with limited skull growth in craniosynostosis would exacerbate the entire scenario, leading to abnormal diastasis in open lambdoid sutures which was the main complaint of parents of these two patients. In usual craniosynostosis the nearby open sutures provide growth of the remaining skull in vertical direction to normal sutures [3]. It causes different kinds of skull deformity subsequent to compensatory growth of open sutures. Disturbance in bone turnover in these two cases might lead to abnormal soft tissue growth at the level of open lambdoid suture as the only compensatory response to limited skull growth at multiple closed sutures, which accompanied by intracranial pressure, pushed away the bones alongside the open lambdoid sutures and provided the diastasis.

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