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CASE REPORT

Coexistence of Chiari malformation type I and isolated hemihypertrophy in a 15-year old girl: a case report

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ABSTRACT

Hemihypertrophy is the asymmetrical growth of one or more parts of the body, and might be either isolated or a component of various syndromes. Coexistence of isolated hemihypertrophy and Chiari malformation type I are very rarely. In this study, our objective was to present a 15 years old girl case with isolated hemihypertrophy and Chiari malformation type I in the light of literature data.

Key Words: Hemihypertrophy, Chiari malformation type I, child, syndrome

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INTRODUCTION

Hemihypertrophy is the asymmetrical growth of one or more parts of the body, and might be either isolated or a component of various syndromes. The exact etiology of isolated congenital hemihypertrophy is unknown. The disease is often sporadic; however, few cases of familial inheritance have been described [1]. Clinical importance of hemihypertrophy arises from the higher incidence of abdominal tumors including Wilms tumor in children compared to the general population [2].

Chiari malformation type I (CMI) is a congenital malformation characterized with herniation of brain stem or cerebellar tonsils from foramen magnum and different degrees of descent to the caudal. The exact etiology of CMI is unknown, although several hypotheses have been proposed. Coexistence of this malformation and isolated hemihypertrophy has been reported very rarely [3].

In this manuscript, our objective was to discuss in the light of literature a 15 years old girl with isolated hemihypertrophy and Arnold Chiari malformation type I.

Case Report

Fifteen years old girl patient presented to our clinic with complaints of vision loss and hypertrophy of left part of the body compared to the right. Medical history revealed that the latter complaint was present since birth and complaints of reduced vision, persistent headache and neck pain emerged in the last 3 months. Physical examination revealed that weight was 65 kg (90 p) and height 163 cm (75 p). Left upper and lower extremities were hypertrophied compared to the right. Symmetrical measurements of the extremities showed hemihypertrophy: Largest circumference of left femur, right femur were 68 and 60 cm, respectively, and circumferences of left arm and right arm were 29.5 and 27.5 cm, respectively (Figure 1). Mental development was normal, pupils were normo-isocoric, eye movements were intact, and cerebellar tests were normal. Examination of other systems proved no abnormality and laboratory tests including complete blood count, liver and renal function tests, complete urine analysis, abdominal and renal ultrasonographic examinations were normal. Doppler examination of left upper and lower extremities demonstrated that venous structures were open, blood flow was intact with no significant edema, and increased muscle thickness on the left compared

to the right. Magnetic resonance imaging (MRI) of the brain demonstrated that cerebellar tonsils were herniated approximately 22 mm from foramen magnum to the spinal cord (Chiari malformation type I) and corpus of the corpus callosum was thinned (Figure 2). An intramedullary syrinx cavity of 3 mm in largest diameter was determined in spinal MRI between the levels of T1-T9 (Figure 3). Normal results were obtained in echocardiography, vertebral artery colored Doppler USG and carotid artery colored Doppler examinations. No abnormalities were determined in genetic analyses. A refractive error of 10/10-6/10 was determined in ocular examination, and she was followed at three-month intervals, without any clinical and laboratory finding of intracranial pressure or concomitant disease at the outpatient clinic for 12 months.

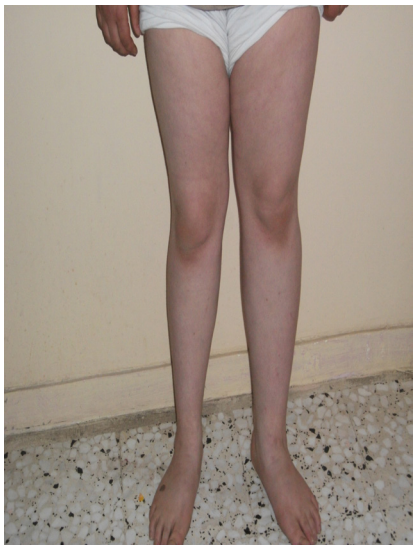

Figure 1. Appearance of isolated hemihypertrophy in a 15-year old girl.

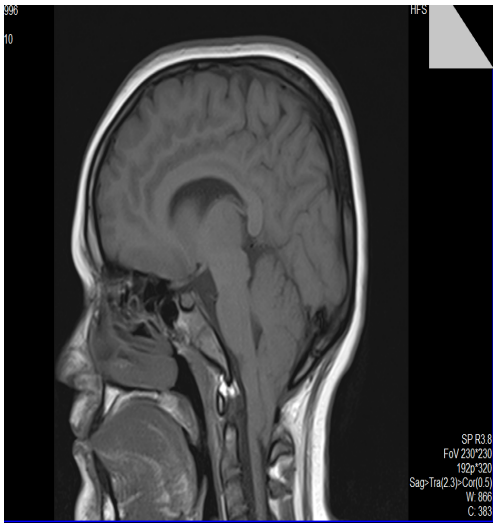

Figure 2. Sagittal T1W MRI of the brain showing the Chiari malformation type I (cerebellar tonsils extend 22 mm below foramen magnum).

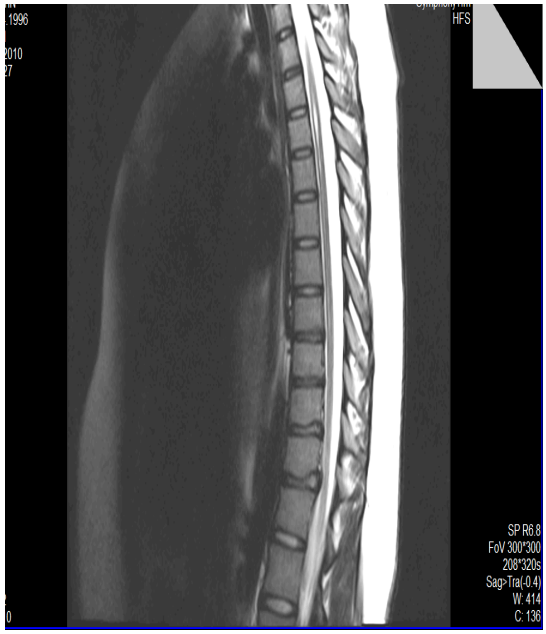

Figure 3. Sagittal T2W MRI of the spine showing the syrinx in thoracic spinal cord.

DISCUSSION

Hemihypertrophy has been classified as complex involving the majority of half of the body, simple involving a single extremity, and hemifacial involving one side of the face [4]. Hemihypertrophy might be isolated or a component of several syndromes including Beckwith Wiedemann, Proteus, Russell-Silver, Neurofibromatosis type 1 and Klippel-Treaunay-Weber [2,5,6]. Mental retardation might be observed in 15-28% of patients with hemihypertrophy [7,8].

Our case presented with isolated hemihypertrophy of the left arm, leg and body in addition to Arnold-Chiari type I malformation. No genetic syndromes were determined in the case. Abdominal USG and cardiological examinations, and mental development were normal.

The exact etiology of isolated hemihypertrophy is unknown; the condition is often sporadic with rare reports of familial cases. In the study of Heilstedt et al. [1] performed in Tunis, two cases of autosomal dominant familial isolated hemihypertrophy have been reported. However, there were no history or physical examination findings suggesting familial inheritance in our case.

Clinical importance of hemihypertrophy arises from the higher incidence of abdominal tumors including Wilms tumor, adrenocortical carcinoma, hepatoblastoma, and less frequently rhabdomyosarcoma, neuroblastoma and pheochromacytoma in children compared to the general population [2].

No tumors were determined in the detailed physical examination and laboratory examinations performed in our case. Follow-up at periodical intervals was initiated since the patient was at greater risk of tumor development compared to the general population.

CMI is a congenital abnormality characterized with herniation of brain stem or cerebellar tonsils from foramen magnum and their caudal descent at different degrees. The exact etiology of CMI is unknown, although several hypotheses have been proposed. The potential pathogenetic mechanism of CMI is considered to arise from a defect in mesoderm of posterior fossa and cerebellum therefore herniates from the incompetent cavity that forms from this mesoderm [9,10]. Several different symptoms of cerebellar, brainstem and spinal cord pathologies have been described and attributed to this malformation since its first description [9,11,12]. This abnormality may manifest itself with obscure and various symptoms including head and joint aches, vertigo, hypoesthesia, and weakness in extremities. The diagnosis is often established with difficulty and therefore it is often delayed. Patients consist mostly of older children or adults, and are often asymptomatic [9,13]. The association between an isolated hemihypertrophy and CMI has first been described in 1976; since then only a few papers reported this association which is related with genetic syndromes in almost all cases [13-15].

Our case presented with non-specific complaints including chronic headache and pain in the extremities. Left-sided hemihypertrophy was determined in physical examination. Brain MRI demonstrated CMI and servico-thoracal MRI demonstrated an intramedullary syrinx cavity of 3 mm of diameter between the levels of T1-T9. In the study by Pettorini et al. [3] reported that coexistence of CMI and syringomyelia in a 9-year old girl with hemihypertrophia whose clinical signs resolved after surgery. Moreover, Tubbs et al. [13] described the tonsillary ectopia (approximately 15 mm) in 2 patients with hemihypertrophia. Authors reported that these subjects did not have syringomyelia and were followed for years with no complications.

Our case differed from that of Pettoroni et al. [3] with an age of 15 and no need for surgery. Additionally, presence of syringomyelia was an important difference of our case from those of Tubbs et al. [13]

In conclusion, the findings of this case suggest that potential coexistence of CMI should be kept in mind in cases of isolated congenital hemihypertrophy. Therefore, the central nervous system and spinal cord imaging can be performed in these patients.

Declaration of Conflicting Interests: The authors declare that they have no conflict of interest.

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