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CASE REPORT / OLGU SUNUMU

Unilateral absence of sphenoid wing in a Neurofibromatosis type 1 patient: imaging findings

Neurofibromatozis tip 1'li hastada sfenoid kanadın tek taraflı yokluğu: görüntüleme bulguları

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ABSTRACT

Neurofibromatosis type 1(NF-1) or von Recklinghausen disease is the most common of the neurocutaneous syndromes. It is characterized by pigmented cutaneous lesions and tumors of central nervous system. By the way, it could affect all the systems of the body. Sphenoid wing abnormalities are very rare and often associated with NF-1 so, they are considered pathognomonic. We report an unusual case NF-1 with unilateral absence of sphenoid wing and findings of multi-detector computed tomography and magnetic resonance imaging. *J Clin Exp Invest 2013; 4 (3): 364-366*

Key words: Neurofibromatosis type 1, multi-detector computed tomography, magnetic resonance imaging, sphenoid dysplasia.

ÖZET

Nörofibromatozis tip 1 (NF-1) diğer adıyla von Recklinghausen hastalığı nörokutanöz sendromların en sık görülenidir. Pigmente cilt lezyonları ve santral sinir sistemi tümörleri ile karakterizedir. Buna ek olarak tüm sistemleri etkileyebilmektedir. Sfenoid kanat anomalileri nadir görülür ve genellikle NF-1 ile ilişkilidir. Bu nedenle genellikle patognomonik kabul edilir. Bu yazıda sfenoid kanat yokluğuyla karakterize nadir bir NF-1 olgusunu, çok kesitli bilgisayarlı tomografi ve manyetik rezonans görüntüleme bulgularıyla beraber sunduk.

Anahtar kelimeler: Nörofibromatozis tip 1, çok kesitli bilgisayarlı tomografi, manyetik rezonans görüntüleme, sfenoid displazi

INTRODUCTION

Neurofibromatosis type 1 (NF-1) is an autosomal dominant disorder which is estimated to occur in one in 3,000 live births. It is the most common of the phakomatoses that includes variety of congenital abnormalities of the skin, nervous system, bones, endocrine glands, and sometimes other organs. Sphenoid dysplasia occurs in 5-10% of cases with NF-1 and absence of a sphenoid wing is very rare. Furthermore, sphenoid wing abnormalities is often considered pathognomonic for NF-1 [1,2].

We report a rare case of NF-1 with unilateral absence of sphenoid wing and findings of multi-detector computed tomography (MDCT) and magnetic resonance imaging (MRI).

CASE REPORT

A-4-year old girl was admitted to hospital with history of multi-organ trauma in 2004. Physical examination revealed no abnormalities due to trauma but revealed cafe-au-lait spots with variety size on his arms and chest. After investigation, the diagnosis of NF-1 was confirmed. In 2008, the patient was admitted to hospital with ptosis of right eyelid and swelling of soft tissue around the right eye. The patient was accepted for radiological evaluation in our clinic with suspicious for periorbital cellulitis. Orbital MRI was performed to show abnormalities of the soft tissue such as inflammation, infections. Axial MRI images showed enlargement of middle cranial fossa, increased soft tissue in the right periorbital area (Fig 1). Additionally, the enlarged right superior orbital fissura and anterior herniation of temporal lob towards to the apex of orbita was found. Sagittal T2 weighted images demonstrated multiple hyperintense focus which are typical lesions of NF-1 at the periventricular region (Fig 2), and after contrast injection heterogeneous enhancement at the right periorbital region was found. The contrast enhancement of soft tissues near bone was suspicious for infections of bone.

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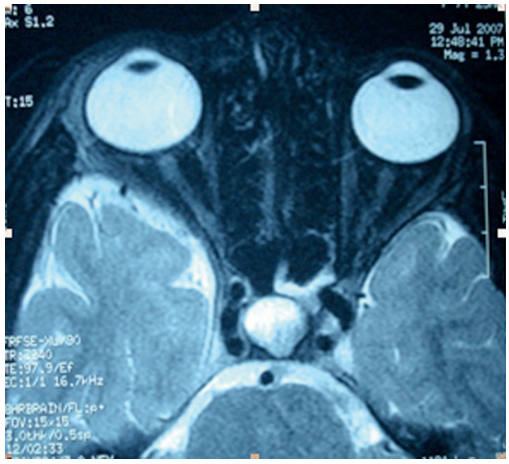

Figure 1. Axial T2 weighted MR image demonstrated enlarged right middle cranial fossa

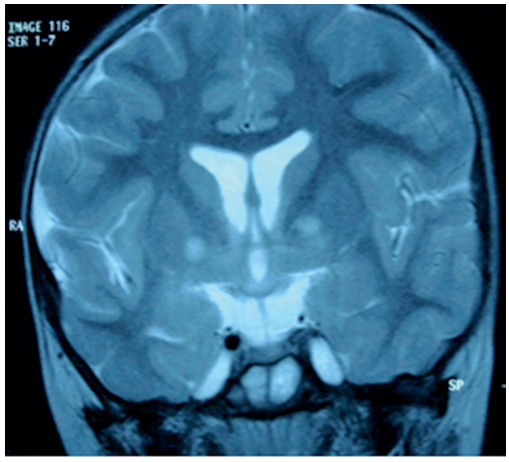

Figure 2. Sagittal T2 weighted image showed multiple hyperintense focus at the periventricular region

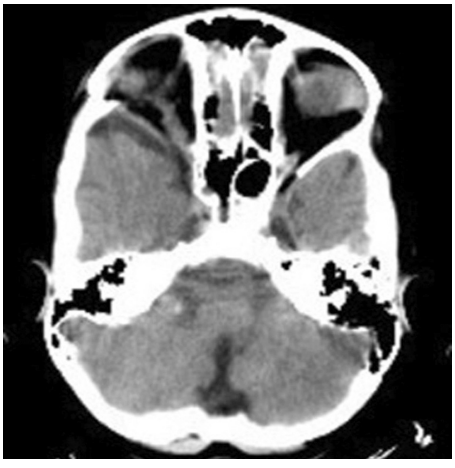

Figure 3. Axial MDCT image showed absence of right sphenoid wing

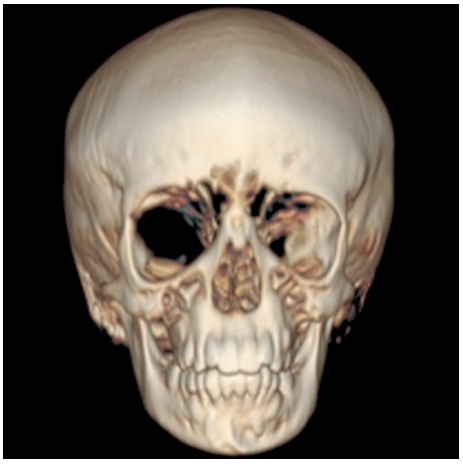

Figure 4. The anterior volume rendered 3D CT image showed details of a bony defect in the right orbital region

To investigate bone abnormalities and evaluate infectious process such as osteomyelitis MDCT was performed and volume rendered 3D CT images were obtained. Axial MDCT images showed absence of right sphenoid wing (Fig 3) and no sign of infection. The anterior volume rendered 3D CT image showed details of a bony defect in the right orbital region (Fig 4).

DISCUSSION

Neurofibromatosis type 1 (NF1), or von Recklinghausen disease, is a common hereditary disease which has a variety of localized or, more frequently, systemic manifestations throughout the thorax, abdomen, pelvis, and extremities. The typical clinical picture of NF-1 includes multiple circumscribed areas of hyperpigmentation in skin which are called café au lait spots and neural tumors of various types. The diagnosis is based on the clinical symptoms. The diagnostic criteria consists of café au lait spots, iris hamartomas (Lisch nodules), axillary freckling, bone dysplasia, affected first degree relatives, and multiple central nervous system tumors such as optic nerve gliomas, neurofibromas, and plexiform neurofibromas. Occasionally, neurofibromas are found incidentally at radiological imaging, because affected patients usually have very mild cutaneous symptoms [2,3]. Skeletal abnormalities are scoliosis, sphenoid wing dysplasia, bony distortion and local cystic or erosive change. Bone abnormalities in NF-1 are progressive and may lead to serious clinical consequences also be resistant to treatment [4]. Furthermore, bone lesions such as severe scoliosis affect prognosis of NF-1 and may reduce life expectancy [5]. Sphenoid wing is one of

the most frequently involved skeletal sites. Although sphenoid wing dysplasia has been observed in between 5 to 10 percent of patients with neurofibromatosis, the absence of a sphenoid wing is very rare. Hence, sphenoid wing abnormalities are often considered pathognomonic [6-8]. Sphenoid wing dysplasia is often detected in asymptomatic individuals. Clinically, sphenoid bone abnormalities cause soft tissue and orbital abnormalities such as exophtalmus, ptosis. Awareness of this process prevent further investigation and wasting time [1,2].

MRI could demonstrate regional signal hyperintensities. They are also named as unidentified bright object (UBO). They are found on T2-weighted MR imaging studies in 40% to 93% of children with NF-1 [9]. MRI could show the other abnormalities of central nervous system such as neurofibroma, plexiform neurofibroma and optic glioma. However, MRI is not able to identify bone abnormalities. Although MDCT has risk of radiation, it is able to scan a large area in a few minutes. MDCT is capable of showing skeletal abnormalities better than MRI. Reconstructions and 3D images demonstrate more detailed information with higher image quality [10].

In conclusion, NF1 is one of the most common genetic disorders, radiologists should be familiar with its diagnostic imaging manifestations such as absence of sphenoid wing. Also, in patients who has already had the diagnosis of NF-1, the physician should be aware of bone changes such as sphenoid dysplasia and associated soft tissue abnormalities.

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