

The Mc Cune - Albright Syndrome: A Case With Giant Mucocele

Mc Cune - Albright Sendromu: Dev Mukoselli Bir Vaka

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Abstract: McCune-Albright syndrome is a specific category of fibrous dysplasia in which the majority of cases feature craniofacial involvement. Mucoceles sometimes accompany this rare condition. This report describes a patient with McCune-Albright syndrome who had a giant mucocele within the dysplastic fibrous lesion in her frontal bone.

Key Words: Fibrous dysplasia, McCune-Albright syndrome, mucocele

Özet: McCune-Albright sendromu, kraniyofasiyal bozuklukların sıklıkla birlikte olduğu fibröz displazinin özel bir alt grubudur. Bu nadir görülen sendrom mukosel ile ilişkili olabilir. Bu makalede, McCune-Albright sendromunda, fibröz displazi ile birlikte olan dev mukosel vakası tartışılmıştır.

Anahtar Kelimeler: Fibröz displazi, McCune-Albright sendromu, mukosel

INTRODUCTION

Fibrous dysplasia is a developmental anomaly of bone. The craniofacial bones are often affected. In such cases, fibrous dysplasia may develop inside or outside the skull, resulting in cosmetic issues and neurological complaints that include vision loss, deafness, and hemiparesis (1,2,3,4,5). Surgical treatment is recommended for cosmetic reasons and to preserve or restore neurological function.

McCune-Albright syndrome represents a special category of fibrous dysplasia in which the majority of cases have craniofacial involvement. The characteristic features of the syndrome are

café-au-lait skin spots, early sexual development, and fibrous dysplasia (1,2,4,5). McCune-Albright syndrome accounts for 3% of all cases of fibrous dysplasia. It is extremely rare for a patient to exhibit all the main traits of this syndrome (1,2,4,5). We present a case of McCune-Albright syndrome in which a giant mucocele formed within the dysplastic fibrous tissue.

CASE REPORT

A 25-year-old woman was admitted to our clinic with progressive cranial shape deformity and generalized tonic-clonic seizures of 2 years' duration. The patient had a history of early sexual development at age 9. Physical examination

revealed craniofacial asymmetry and bilateral exophthalmia, as well as café-au-lait spots on the legs and neck. The patient's neurological evaluation was normal. Cranial computerized tomography demonstrated enlargement of the frontal bones (Figure 1). T₂-weighted magnetic resonance images showed hyperintense and

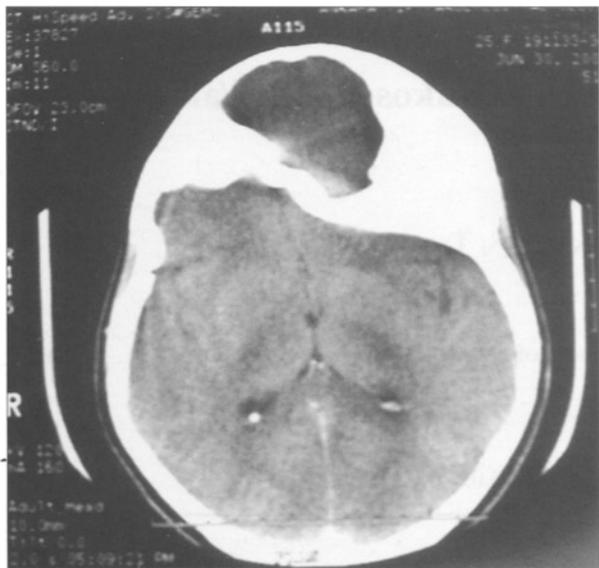


Figure 1: A computerized tomography image demonstrates the expanded frontal bone containing a mucocele.

hypointense cyst formation resembling a mucocele. This lesion was located within the expanded frontal bone, and the expansion suggested fibrous dysplasia (Figure 2). The patient's hormone profile revealed increased serum prolactin (67.31 mIU/ml), decreased serum total testosterone (6.37 ng/ml), and decreased serum cortisol (1 µg/ml).

The combination of early sexual development, café-au-lait pigmentation, endocrinological abnormalities, and fibrous dysplasia suggested the diagnosis of McCune-Albright syndrome. Cortisone replacement therapy was initiated and surgery was scheduled.

The operation included a large frontal craniotomy and removal of a mucocele. The mucocele was located in dysplastic fibrous tissue that had invaded the inner table of the frontal bone. The frontal sinus was cranialized. Because the patient refused the use of synthetic cranioplasty

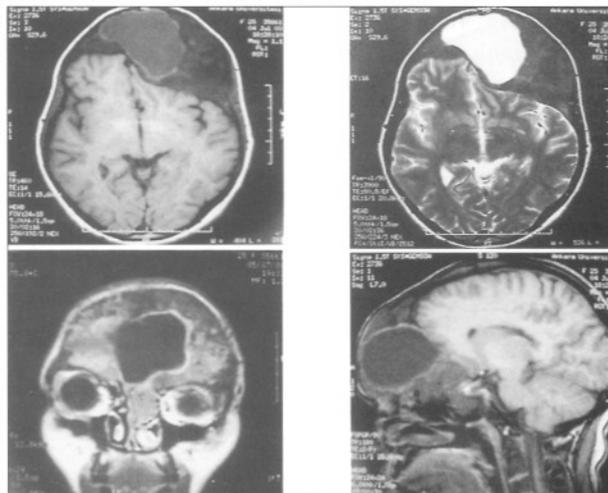


Figure 2: A T₁-weighted magnetic resonance image (upper left) shows the patient's frontal bone expanded by fibrous dysplasia. A T₂-weighted image (upper right) demonstrates hypointense and hyperintense cyst formation suggesting a mucocele within the dysplastic tissue. Coronal and sagittal T₁-weighted images are shown at the lower left and right, respectively.

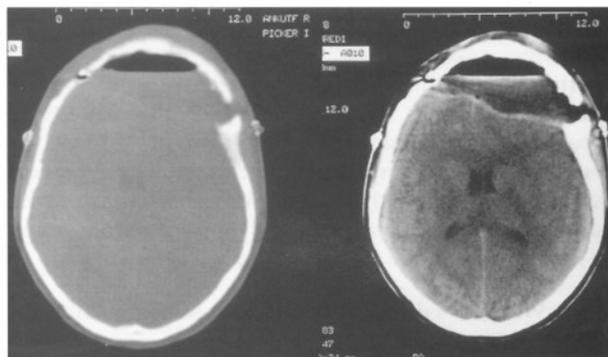


Figure 3: Postoperative control computerized tomography confirmed complete removal of the dysplastic fibrous tissue.

material, the reconstructed frontal bone was replaced in the correct anatomical position. Histopathological examination of the diseased bone tissue confirmed the presence of fibrous dysplasia. The patient was discharged in good condition on postoperative day 6.

DISCUSSION

McCune first described this condition in 1936, and Albright followed in 1937. Consequently, the name McCune-Albright syndrome was applied (2). This syndrome is an inherited disease of unknown

cause. Studies indicate that many types of endocrinopathy are associated with this syndrome, including hyperthyroidism, changes with pituitary adenoma, Cushing's syndrome, hormonal shifts in autonomous adrenal hyperplasia or hypoplasia, and hyperprolactinemia (1,2).

Recent research suggests that a mutation in exon 8 of the *Gs α* gene might cause early sexual development by activating the signal-transduction pathways in early embryogenesis. In our case, it is likely that sexual precocity was the patient's first manifestation of McCune-Albright syndrome, but this was overlooked by her parents and physicians.

Many reports have documented multiple serious endocrinological abnormalities and other significant organ-system pathologies that can be lethal in patients with this syndrome (1,2,5). Although the reports to date have described endocrinological abnormalities of hyperfunction, our patient exhibited below-normal serum cortisol and total testosterone levels. It should be emphasized that thorough endocrinological assessment of these patients is essential, especially when surgery is planned. In addition to minimizing surgical risk, many of these patients' endocrinological problems respond well to medical therapy.

Fibrous dysplasia occurs in two forms, monostotic (70%) and polyostotic (30%), and a mixed type in which both types are seen in McCune-Albright syndrome (1). Monostotic refers to single bone involvement, frequently the craniofacial bones or ribs. This type of dysplasia generally ends with the onset of puberty. In some cases, the monostotic form progresses to the polyostotic form in the third decade (1,2). The polyostotic form is characterized by multiple bone lesions, which are usually located on the same side of the skeleton and mainly affect the lower extremities and facial bones (1,2,4,5). Recurrent bone fractures and extremity deformities are pathognomonic for the polyostotic form. Although the majority of fibrous dysplasia that accompanies McCune-Albright syndrome is polyostotic, the frontal bone was affected in our case. Also, there was no history of recurrent bone fractures.

The combination of fibrous dysplasia and mucocele is considered to be extremely rare, and was first described by Brich et al. in 1979 (1,2). Authors have suggested that such mucoceles develop from obstruction of a sinus ostium caused by problems such as osteoma, fibrous dysplasia, and chronic inflammation of the sinus mucosa (1,2,3). It is proposed that the mucocele slowly distends due to mucous secretion from epithelial cells within the encapsulated mucosa (3). The lesion is thought to gradually enlarge over time within the soft tissue, or it may destroy a thin section of inner table of bone and expand into the cranium. Fibrous dysplasia is abnormal fibrous tissue that fills the osseous medullar cavity, and a thin shell of normal cortical bone almost always remains (1,2,4,5). This thin shell of normal bone may limit the size of any accompanying mucocele. In our case, we suggest that the mucocele might have extended into the dysplastic fibrous tissue because this tissue is more forgiving than the thin shell of normal cortical bone that was present.

As mentioned, some reports have documented vision loss in patients with McCune-Albright syndrome. All these cases were attributed to fibrous dysplasia, but we suggest that neurosurgical decompression of the optic nerve could preserve vision in this situation.

This article describes the rare finding of a giant mucocele located within dysplastic fibrous tissue in a patient with McCune-Albright syndrome. Multiple types of endocrine hyperfunction, café-au-lait pigmentation, and fibrous dysplasia are the major diagnostic features of this syndrome. All patients diagnosed with fibrous dysplasia, especially those who exhibit abnormal pigmentation or endocrine alterations, should be carefully evaluated for endocrinological abnormalities. Early diagnosis of this syndrome will likely prevent further endocrinological and/or the development of visual problems.

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