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Case Report

Clinical, Radiological and Endocrinological Findings in a Case of McCune-Albright Syndrome

McCune – Albright Sendromu Olan bir Hastada Klinik, Radyolojik, Endokrinolojik Bulgular

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ABSTRACT

The McCune-Albright syndrome was described as a syndrome of polyostotic fibrous dysplasia, café au lait skin pigmentation, and autonomous endocrine hyperfunction in 1937. We report a 17-year girl with early menarche and accelerated growth for the past three years. The endocrinological examination showed slight rise of growth hormone with other hormones in normal range. The CT showed an expansive bony lesion over the left parietal area. The bone mass was excised with bone cement cranioplasty performed for the defect. Histology confirmed it was fibrous dysplasia. Although uncommon, this syndrome must be kept in mind in cases with bony abnormalities and extensive endocrinological workup done and treatment given for best results.

KEYWORDS: Café au lait, Fibrous dysplasia, McCune-Albright syndrome

ÖZ

McCune Albright sendromu 1937 yılında poliostotik fibröz displazi olarak tanımlanmış bir sendromdur. Bu sendrom deride kahverengi renklenme (Cafe au lait) ve otonomik endokrine hiperfonksiyon ile karekterizedir. Son 3 yıl içerisinde erken menarj ve erken gelişme yakınması olan 17 yaşında kadın hastaya çekilen bilgisayarlı tomografide sol parietal kemik genişlemiş olarak izlendi.Yapılan biyokimyasal incelemede büyüme hormonunun hafif bir şekilde yükseldiği görüldü, diğer hormon sonuçları normal sınırlarda bulundu. Parietal bölgedeki kemik lezyonun tamamı çıkarıldı, lezyonun çıkarılması sonucu oluşan boşluk metil-metakrilat ile kranioplasti yapılarak onarıldı. Histolojik incelemede fibröz displazi saptandı. Bu sendrom nadir olarak görülen fibröz displazi olmasına karşın, bu hastalarda endokrinolojik incelemeler yapılması hastalardan en iyi sonucun alınmasını sağlayacaktır.

ANAHTAR SÖZCÜKLER: Cafe au lait, Fibröz displazi, McCune-Albright sendromu, Deride kahverengi renklenme

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INTRODUCTION

The McCune-Albright syndrome was described as a syndrome of polyostotic fibrous dysplasia, café au lait skin pigmentation, and autonomous endocrine hyperfunction in 1937 (1,5). This syndrome manifest most commonly as precocious puberty and other endocrinological features like acromegaly, gigantism and hypercortisolism. Fibrous dysplasia in this syndrome mainly affects the long bones although any bone can be involved. These lesions can be aymptomatic or lead to disfiguring changes leading to gross cosmetic and functional abnormalities. Café au lait spots are large melanotic macules without any definitive underlying abnormality.

CASE REPORT

A 17-year-old girl presented with history of irregular menstruation, accelerated growth and progressively increasing swelling over the left parietal area for the last 3 years. Menarche was noticed at 13 years and the present menstrual cycle consisted of scanty irregular monthly bleeds occurring every three to five months. The family members had also noticed that she had grown taller than her older sisters over the last few years. Examination showed her to be taller by two inches from other family members with presence of a hard ill-defined smooth non-tender bony mass of 14 by 12 cm over her left parietal area (Figure 1). The overlying skin was moveable over the skull mass and a large café au lait patch was present in the right retroauricular area (Figure 2). There were no other musculoskeletal abnormalities in the rest of the body. The other siblings did not reveal any signs of neurofibromatosis.

The serum electrolyte, blood sugar and renal function tests were normal and the hormonal examination showed fasting growth hormone at 15 ng/ml (normal <5 ng/ml). Two consecutive samples did not reveal any abnormal levels of serum estradiol, follicle-stimulating hormone (FSH), or luteinizing hormone (LH) levels. The cortisol and thyroid hormone level were within normal range. Computed tomogram (C.T.) showed a large expansive bony lesion in the left parietal area with no intracranial extension (Figure 3). The centre showed sunray type of appearance with a cystic centre. The sella was normal. Ultrasound of the abdomen and pelvis did not reveal any underlying genitourinary abnormality.

Figure 1: *Picture showing the accelerated linear growth in this case.*

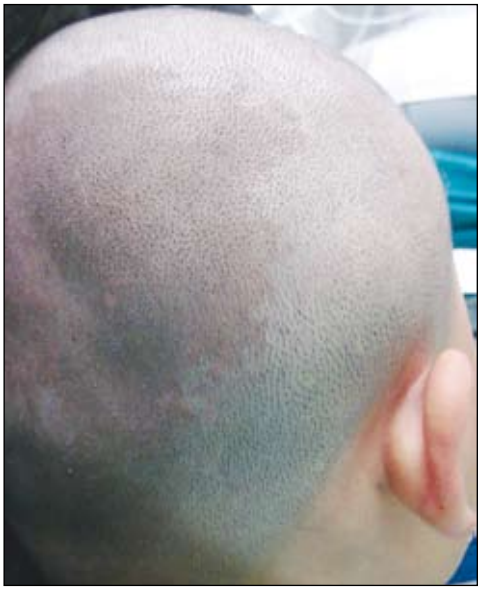


Figure 2: *The classical café au lait spot is seen in the right retroauricular area.*

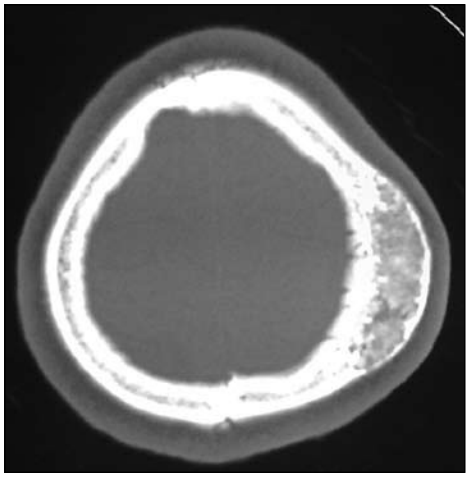


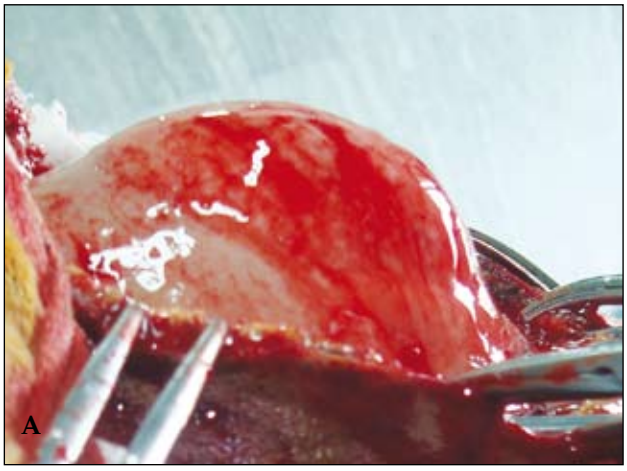
Figure 3: *CT showing the sunray type expansive bony lesion over the left parietal area. There was no intracranial extension.*

She underwent craniotomy and excision of the bony mass with a wide 2 cm margin followed by bone cement cranioplasty of the defect (Figure 4A,B). Intraoperatively there was no dural infiltration and the lesion although looking cystic on the CT was rock solid. A linear cut through the centre did not reveal any cystic defect. Postoperative CT showed good cover of the defect and there were no neurological sequel. The histopathology showed features of suggesting fibrous dysplasia (Figure 5). She is presently undergoing treatment with the endocrinologist for her hormonal abnormalities.

DISCUSSION

The classical triad of café-au-lait cutaneous spots, polyostotic fibrous dysplasia and endocrinopathies comprise the McCune-Albright syndrome. It is a rare disease with estimated prevalence between 1/100,000 and 1/1,000,000. The underlying genetic abnormality involves the mutation and activation of Gs protein alpha subunit coupling 7-transmembrane-domain receptors to adenylate cyclase that mediates the activation of adenyl cyclase and subsequent gland autonomous secretion. It is thus a rare proteiform disease due to postzygotic, somatic mutations at codon R201 of the GNAS1 gene resulting in cellular mosaicism (3). Precocious puberty is the commonest manifestation followed by thyroid disorders (8). Fibrous dysplasia that accounts for 7% of total benign bone tumors is also common in this syndrome (7).

The management is often multidisciplinary. Precocious puberty is managed medically in girls with the use of an anti-estrogen and in boys a combination of an antiandrogen with an aromatase inhibitor testolactone is preferable (6). Other drugs



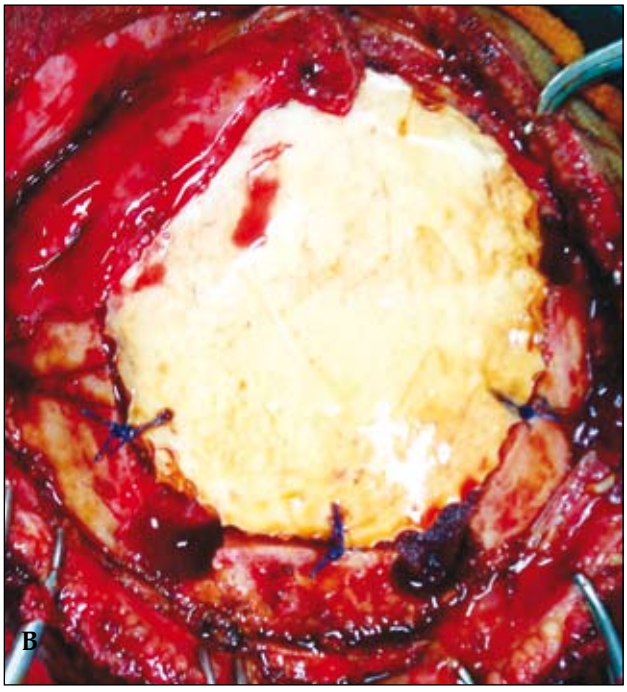


Figure 4: *Intraoperative picture showing the expansive bone mass* ***(A)*** *and the defect covered with bone cement* ***(B).***

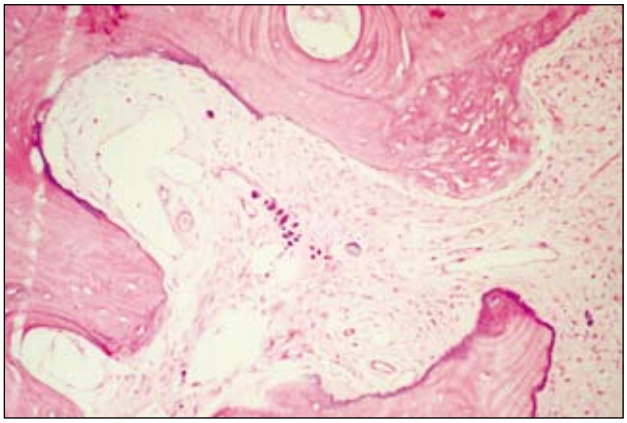


Figure 5: *Hematoxylin and Eosin stain showing bone canaliculi with absence of osteoblast in the periphery and fibroblast like cells with minimal extracellular matrix suggestive of fibrous dysplasia.*

like tamoxifen, fadrozole and ketoconazole have also been shown to be effective. Bisphosphonates have been used in the treatment of fibrous dysplasia to relieve bone pain and improve lytic lesions and Calcium, vitamin D and phosphorus supplements are also known to be beneficial in its management. Surgery is mainly indicated for symptomatic fibrous dysplasia and the procedure depends on the site, extent and relation to neighboring structures. Those involving the cranial base are the most difficult to manage. Hormonal abnormalities are best managed by an endocrinologist depending on the specific endocrinological manifestation. Malignant transformation is very rare. Two cases of thyroid cancer and one case of secondary osteosarcoma in fibrous dysplasia have been reported in literature (2,4).

There is yet no cure for this disorder and excellent outcome is achievable with medical treatment. Long term endocrinological follow up should be advised with early detection and treatment of the adverse effects of prolonged drug use.

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