



Imaging of advanced craniofacial fibrous dysplasia associated with McCune-Albright syndrome: A case report

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ABSTRACT

The fibrous dysplasia of bone is painless, benign, and slowly progressive bone lesion. It may rarely become aggressive by compression of adjacent organs or malignant transformation. This disease falls within the scope of a McCune-Albright syndrome in less than 7 % of cases. The authors reported the case of a 25-year-old woman, living in the countryside, who suffered from a severe craniofacial fibrous dysplasia which has begun since her childhood with no medical care. Her clinical picture was that of McCune-Albright syndrome, and CT scan revealed advanced dysplasia with endocranial compression.

1. Introduction

Fibrous dysplasia of bone is a benign, a congenital, and a non-communicable disease caused by the GNAS mutation encoding the Gsa protein. Its prevalence is less than 1/2,000 [1,2]. It affects both sexes equally and is characterized by a benign proliferation of fibrous tissue in the bone marrow. Bone lesions are usually unique or multiple and can affect all osseous structures with a high frequency of maxillofacial lesions.

The McCune-Albright syndrome is a rare disease characterized by skeletal lesions, skin hyperpigmentations, and hyperfunctional endocrinopathies. It represents less than 7 % of bone dysplasia and its prevalence is estimated at between 1/100,000 and 1/1,000,000 [3,4]. It slowly evolves but might be aggressive in exceptional cases or lead to malignant transformation in 0.9–4% of cases [5,6].

2. Clinical observation and imaging findings

A 25-year-old woman living in the countryside was received at the CT scan Department for a major craniofacial abnormality which has emerged since her childhood associated with severe headaches. This patient had never been at any health care center before this abnormality. Her clinical history showed that she was normally born without cranial issue. The cranial and facial lesions were gradually developed since the early childhood with early menarche (endocrinopathy). The physical examination revealed a small size, a diffuse hyperpigmentation of the skin, a frontal bone hump, and two parietal bone humps on the

head (Fig. 1). The visual and auditory acuities were normal.

The CT scan showed a diffuse thickening of the cranial vault with an osteocondensation of "frosted glass appearance" (Fig. 2). The osseous frontal abnormalities were both condensing and lytic with multifocal cortical ruptures in the endocranium, periosteal reactions, and extradural intracranial extension of increased tissue density after injection of contrast medium. The mixed imaging features suggested a malignant transformation of the disease (Fig. 3). The diploe was thickened. The considerable osseous frontal hypertrophy was responsible for a compression of the frontal cerebral parenchyma underscored by the deletion of the cortical furrows. The maxillary bone, mandible, temporal bones, facial sinuses, and skull base bones had lytic lesions with a blown appearance of the bones (Fig. 4). The significant bone hypertrophy had almost led to the total stenosis of the facial sinuses. The ethmoid cells were normal with no tooth extraction. Three dimensional reconstructions have been useful in assessing craniofacial dysmorphism (Fig. 1). The cervical spine and the osseous structures of both shoulders also showed similar diffuse osteolysis features. This polyostotic fibrous dysplasia associated with skin patches and endocrinopathy was diagnosed as McCune-Albright syndrome within this patient. Unfortunately, she died of cardiac arrest in the following days after the performance of imaging. Her sudden death hampers us from performing a bone biopsy to confirm the malignant transformation of bone dysplasia.

3. Discussion

Although the fibrous bone dysplasia is uncommon, its aggressive

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Fig. 1. Three-dimensional CT SCAN reconstruction shows frontal and parietal bone humps. Notice maxillary hypertrophy.

forms are well-documented. According to the literature data, the frequency of the aggressive forms of the fibrous bone dysplasia varied from 0.9 % to 4 % with a multifocal predominance. Their association with McCune-Albright syndrome is rare as described in our case [5,6].

3.1. Epidemiological and clinical characteristics of aggressive craniofacial dysplasia

In the literature, this aggressive tendency was generally observed after more than 20 years of the disease onset [4]. However, rare cases of

malignant transformation have been reported in adolescents [6,7]. Our patient who lived in the countryside had not received any medical care since her childhood because of her family's financial issue and of the bad cultural perception of deforming disease regarding as curses. The long delay of the clinical management of the patient's disease would probably responsible for her considerable craniofacial deformation. The maxillary bone was described in the literature as the main site of malignant transformation, followed by the mandible, and zygoma in the case of aggressive dysplasia, [3,4].

In our case, the frontal bone underwent a cortical rupture of the

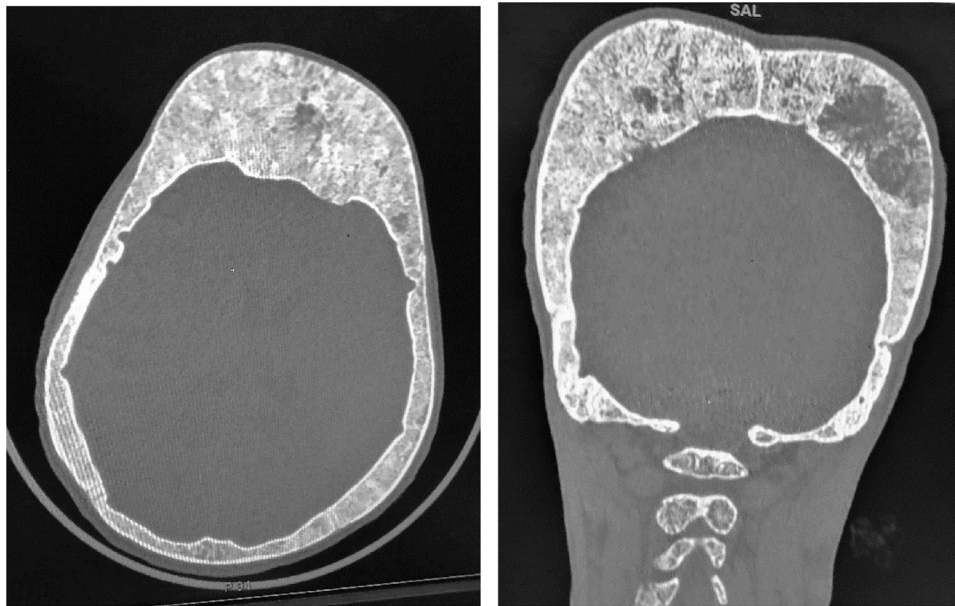


Fig. 2. Axial CT scan with coronal reconstruction showing heterogeneous condensing hypertrophy in frosted glass of the diploe of the cranial vault bones.

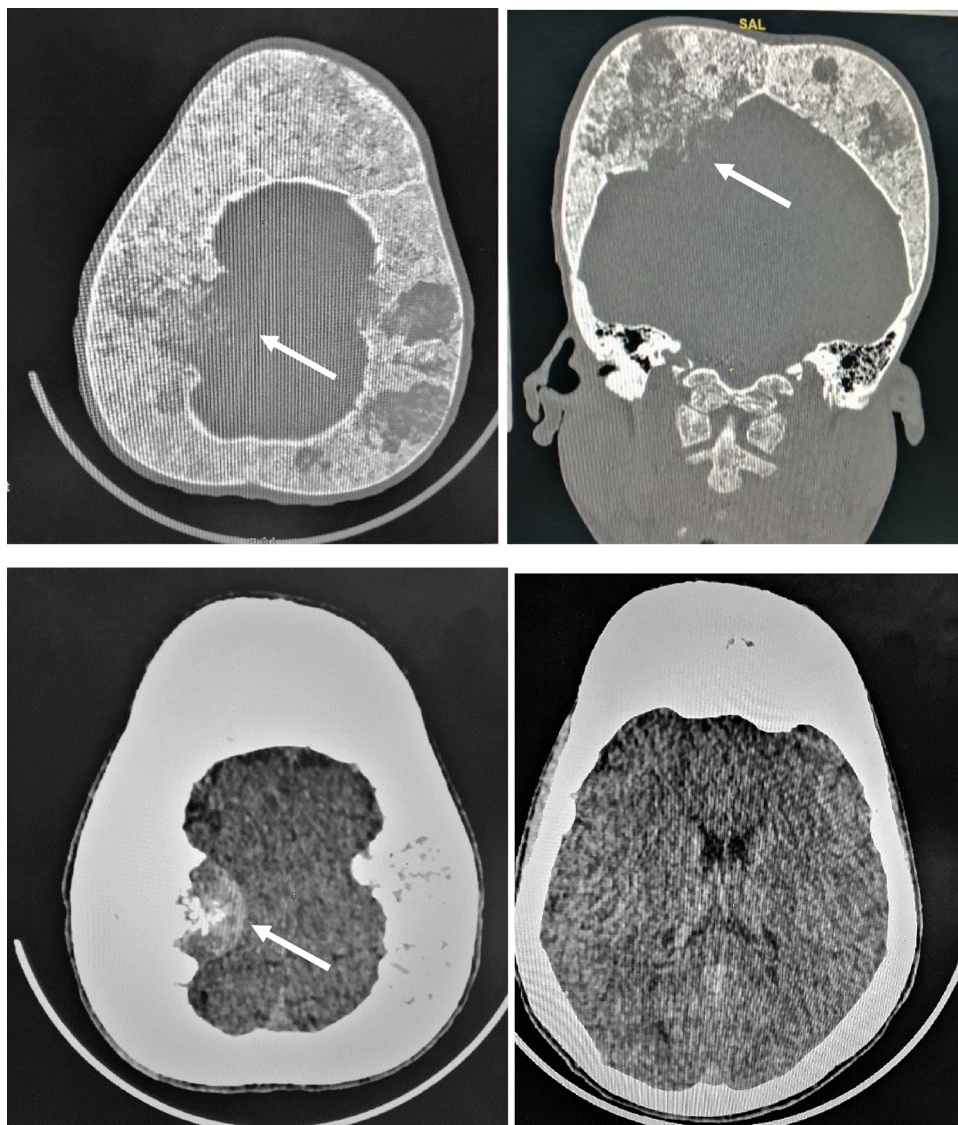


Fig. 3. Axial CT scan with coronal reconstruction showing signs of radiological aggressiveness with cortical rupture and extradural extension of lesions (arrow).

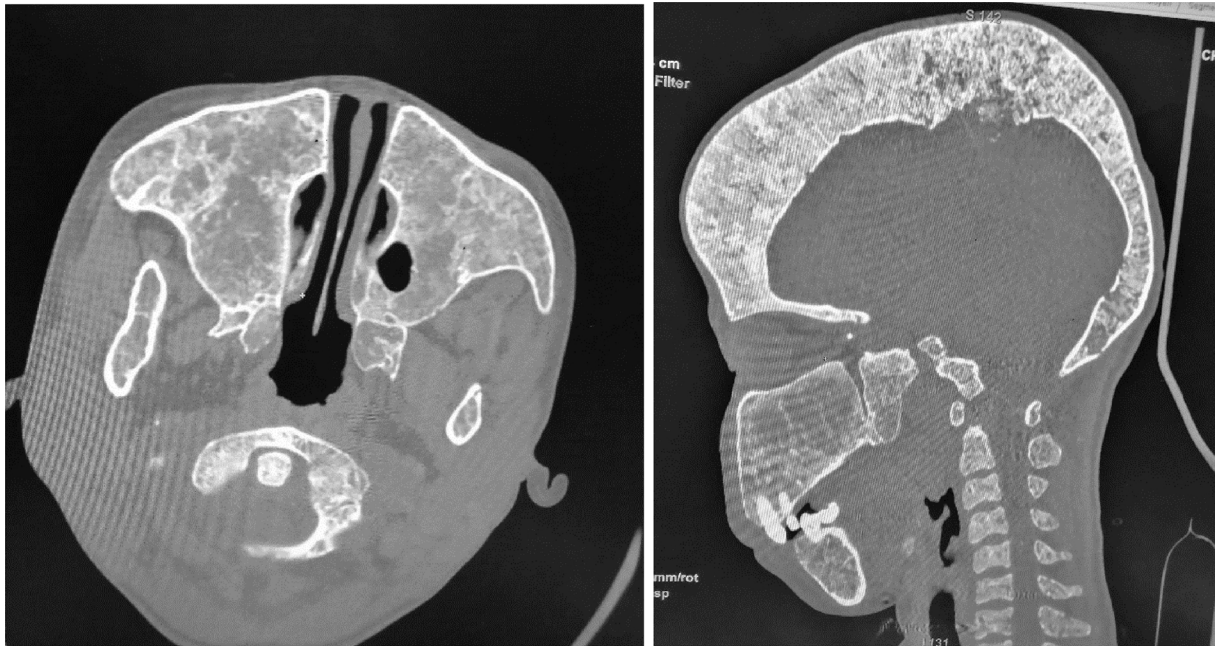


Fig. 4. Axial CT scan with sagittal reconstruction showing diffuse lesions of the facial bones with condensation of the maxillary and sphenoidal sinuses.

inner table with an extradural extension of the lesion. The clinical signs predicting this aggressive transformation were the bone swelling with unaesthetic craniofacial deformation, followed by increased pain and severe headache [5,8,9]. However, a few cases of asymptomatic malignant transformations or early malignant forms have been reported [4]. The etiopathogenesis of aggressive forms remains unknown. The role of radiotherapy as a risk factor of malignant transformation remains controversial, although some studies have shown a significant rate of malignant transformation in patients with bone dysplasia who received radiotherapy (39 %) [7]. Therefore, the use of radiotherapy should be codified in people with fibrous bone dysplasia.

3.2. Histopathologic characteristics and therapeutic approach to the aggressive form of fibrous bone dysplasia

Typically, the histologic examination of the biopsy of the suspected bone disease is only necessary in unusual or suspected cases of malignancy. The risks and benefits of the biopsy should be clearly explained to patients [2]. Osteosarcoma is the predominant histologic type, followed by fibrosarcoma and chondrosarcoma. Interestingly, at least 2 co-existing histological types have been identified in malignant tumours in a very few cases [10–12]. According to the Fibrous dysplasia (FD)/McCune-Albright Syndrome (MAS) International Consortium Management Guidelines [2], the objective of staging craniofacial lesions was to define and to record the extent, the distribution, and the impact of fibrous dysplasia on the craniofacial skeleton. After anamnesis and physical examination, the following tests are recommended when they are clinically relevant: objective assessment of facial asymmetry by clinical and three-dimensional photography and psychological impact assessment [29]. The imaging assessment encompasses series of facial radiographs and 1-mm or less thickness of sections CT scan. If the craniofacial lesions are adjacent to the valuable structures or nerve pathways, the patient should be transferred to the appropriate medical specialties centers. Our patient had extensive craniofacial impairment except the cranial nerves. Adjuvant treatments, such as radiotherapy and chemotherapy, allow local control of the disease by stabilizing it

and increase the overall survival of patients in cases of malignant transformation. The radiological evaluation includes a series of facial radiographs, a thin section CT scan of 1 mm or less in thickness. If the craniofacial lesions are adjacent to the relevant nerve structures or pathways, consideration should be given to transferring the patient to the appropriate specialties. Regarding to prognosis, the treatment outcomes of these patients mostly remain unsatisfying. Local tumor recurrences and distant metastases are the major causes of the death of the patients [2,4].

4. Conclusion

Advanced craniofacial fibrous dysplasia is caused by late management in a context of limited access to health cares. CT Scan is used for exhaustive lesion assessment. At this stage, the disease prognosis is poor.

Declaration of Competing Interest

The authors declare that they have no conflict of interest.

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