peer reviewed **CASE REPORT**

McCune-Albright syndrome: a case report and review

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ABSTRACT

McCune-Albright syndrome is a rare condition. Patients may present with polyostotic fibrous dysplasia, have café-au lait spots on the skin and evident endocrinopathies, the most common being precocious puberty which is probably an early diagnostic sign. This case report is of a 15 year-old male who presented for dental treatment and was diagnosed with the syndrome. Radiological examination revealed characteristic bony dysplasia in his mandible and maxilla. Periodic radiographic evaluations, including cone-beam computed tomography (CBCT), were performed over the next three years. The images showed the expected slight recession of his bony lesions.

Keywords fibrous dysplasia, polyostotic lesions, cone beam computed tomography

LAY ABSTRACT

This is a case report of a male teenager who presented with dental problems and was found to have irregular tissue in his jaw and also creamy-brown spots on his skin. His pathology presented challenges to management and treatment options.

CASE REPORT

In late 2009, a 15 year-old male presented at an oral health centre. He was short and stocky and seemed to have a slight scoliosis. His stature and weight were below the 50% percentile for boys of his age (Figure 1). He had an open bite malocclusion and was referred by an orthodontic department for routine panoramic (PAN) and cephalometric (CEPH) radiographs for assessment. The radiographs showed the maxillary canines to be in severely ectopic positions, with the crowns mesial to the roots of the lateral incisors. Multiple coalescent radio-opacities with irregular borders were seen within the mandible. The maxillary canines were in positions inimical to orthodontic movement and in early December 2009 these teeth were removed by a maxillo-facial and oral surgeon. At surgery, the surrounding tissue was observed to be of a consistency unfavourable to tooth movement. No attempt was made to excise any of the lesions, but some of the fibrous tissue was removed as a biopsy. Histological examination confirmed that the tissue was a benign fibro-osseous lesion. However, one relies on radiological patterns on images to classify the type. Widespread irregular radio-opacities are indicative of a fibrous dysplasia as opposed to an ossifying fibroma which is discrete and well-defined. Osseous dysplasias on the other hand present with totally different radiological patterns; none of which resemble a fibrous dysplasia.

The patient was referred for an endocrinological assessment in January 2010. This assessment, combined with the radiological patterns of fibrous dysplasia, confirmed the diagnosis of McCune-Albright syndrome (MAS). From 2013 to 2017 PAN radiography was done for periodic assessments of the lesions (Figure 2). A cone beam computed tomography (CBCT) scan was taken in January 2016 to investigate the extent of the lesions and to assess the possibility of planning further orthodontic treatment to correct the malocclusion (Figure 3). A second CBCT scan was undertaken in February 2017 to determine the feasibility of placing dental implants in the maxilla. At this radiographic session, the patient reported that he had 'hard bony lumps' on both humeri and a stiffness of his left shoulder. These symptoms were in keeping with MAS. The report of the oral radiologist indicated that the lesions were a form of fibrous dysplasia (FD) and that extensive orthodontic movement was not advised. Treatment was thus limited to aligning the maxillary permanent incisors, whilst the deciduous maxillary canines were retained. The placement of implants was contra-indicated. A follow-up CBCT scan done in November 2018 showed that radiolucencies were seen within some of the radiopaque lesions. This suggested a 5-10% regression in these lesions (Figure 4). Other lesions, however, appeared to be static with no indication of change. The report at this stage confirmed that placement of implants could not be considered.

The patient is a healthy and active individual currently engaged in academic studies. He is satisfied with the current status and is determined to live his life to the full.

DISCUSSION

Fibrous dysplasia (FD) is a benign skeletal disorder in which normal bone is replaced with woven bone and fibrous tissue creating growing lesions, resulting in bone expansion and asymmetry.[1-3] Radiologically, FD lesions present as radiopaque masses with a 'ground glass' appearance.[2,4] (Figures 2, 3, 4). The condition can be monostotic (affecting one bone), or polyostotic (affecting many bones). The skull and jaws, followed by the femur and tibia, are common sites.[2,3,5]

The maxilla is more frequently affected, resulting in displacement of teeth and malocclusion. FD can occur as a separate abnormality, but if it occurs in conjunction with skin hyperpigmentation (café-au-lait spots) and/or endocrinopathies then MAS is the most likely diagnosis.[1,3]

MAS was first identified in 1937 by McCune and Albright, as a rare, sporadic genetic disease.[3,6,7] Positive diagnosis requires the presence of all three described manifestations: polyostotic FD (skeletal), café-au-lait spots (skin), and endocrinopathies (endocrine).[1,3,4,6-9] The prevalence of MAS is estimated to be between 1:100 000 and 1:1 000 000 in the general population with no ethnic differences in the frequency of the disease.[1,4,7]

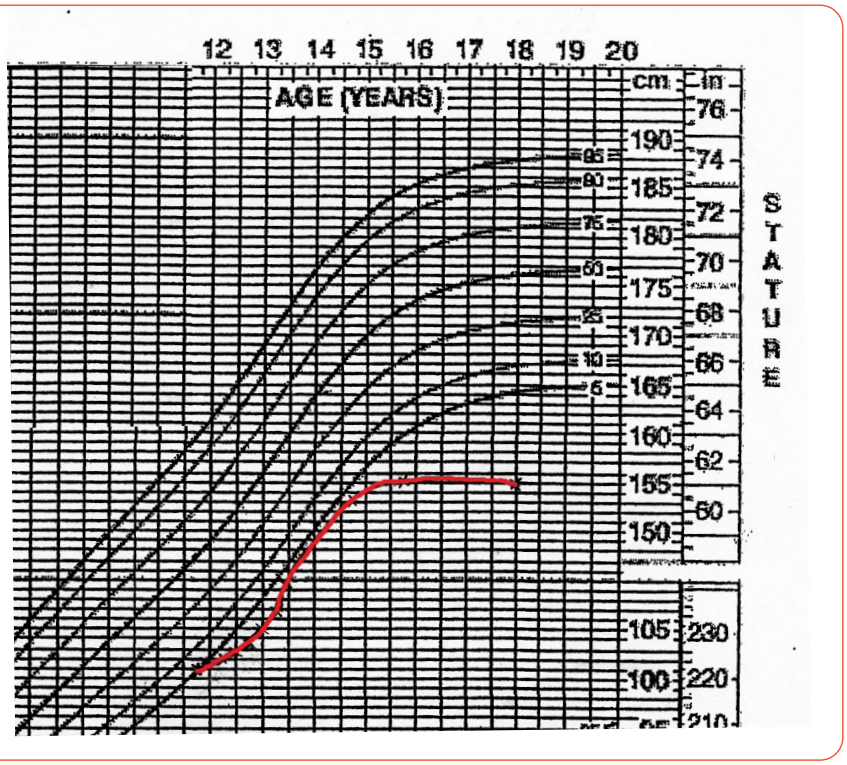

Figure 1. Growth chart showing the patient's height being well below the 50% percentile range.

Precocious puberty (PP) is the most common endocrine symptom and is believed to be the presenting clinical sign of MAS.[4,7] Females are affected more frequently than males; PP may become apparent later in life in males compared with females.[4,7,8] The abnormal activation in females of granulosa or theca cells of the ovary can result in a precocious increase of endocrine function; in males it is only the somatic mutation of the Leydig cells that will result in a PP finding.[7] The studies of Utrianien et al[1] and Wang et al[3] showed that all patients with MAS had skin hyperpigmentation (café-au-lait spots) and that recognition could point to the diagnosis being possible at a younger age before the onset of PP. Utrianien et al[1] advise that a diagnosis of MAS should be considered in patients who have widespread café-au-lait changes because a delay in the diagnosis may have considerable consequences, but this remains a diagnostic challenge.[1]

In childhood, MAS management usually focusses on the treatment of FD and PP.[10] A study conducted by Wang et al[3] found that the use of bisphosphonate therapy (BPT) in adults and children resulted in a

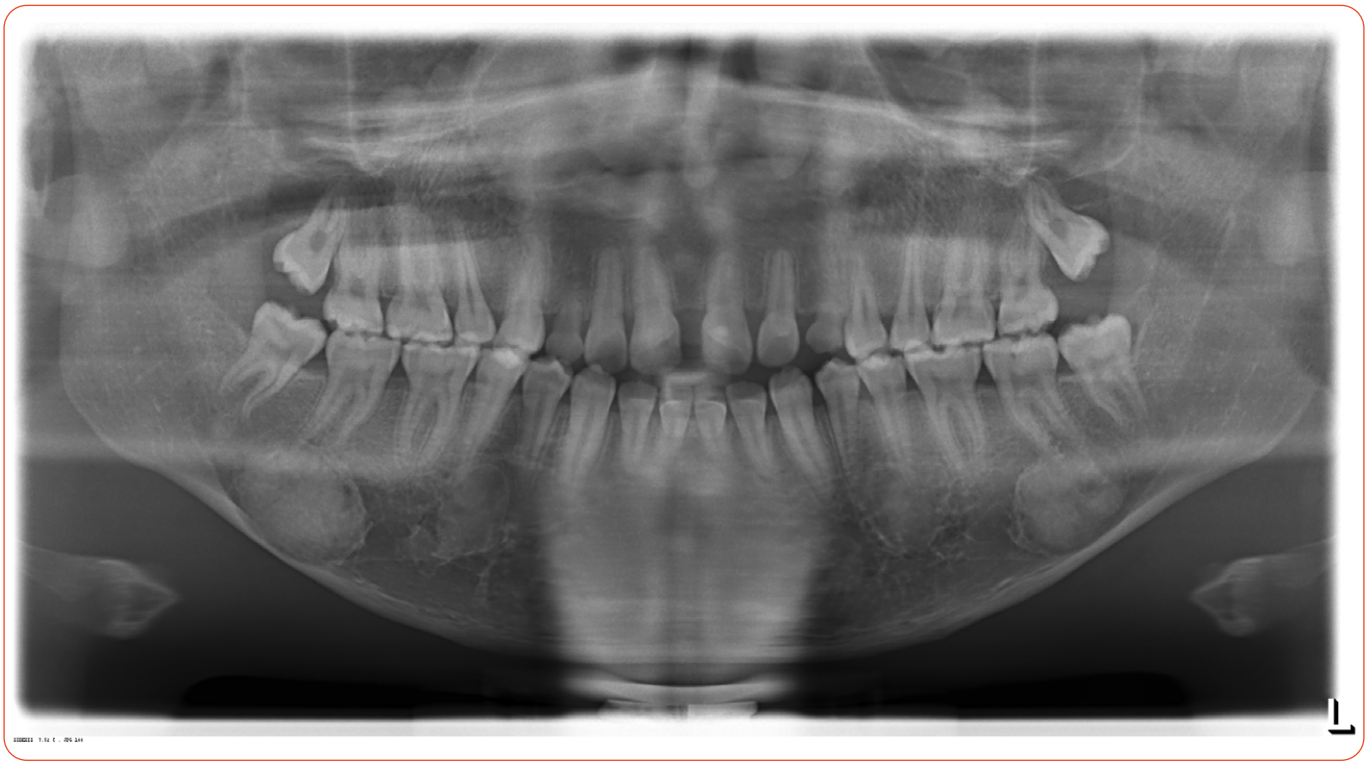

Figure 2. A panoramic x-ray taken in 2013 showing fibrous dysplasia lesions in the mandible.

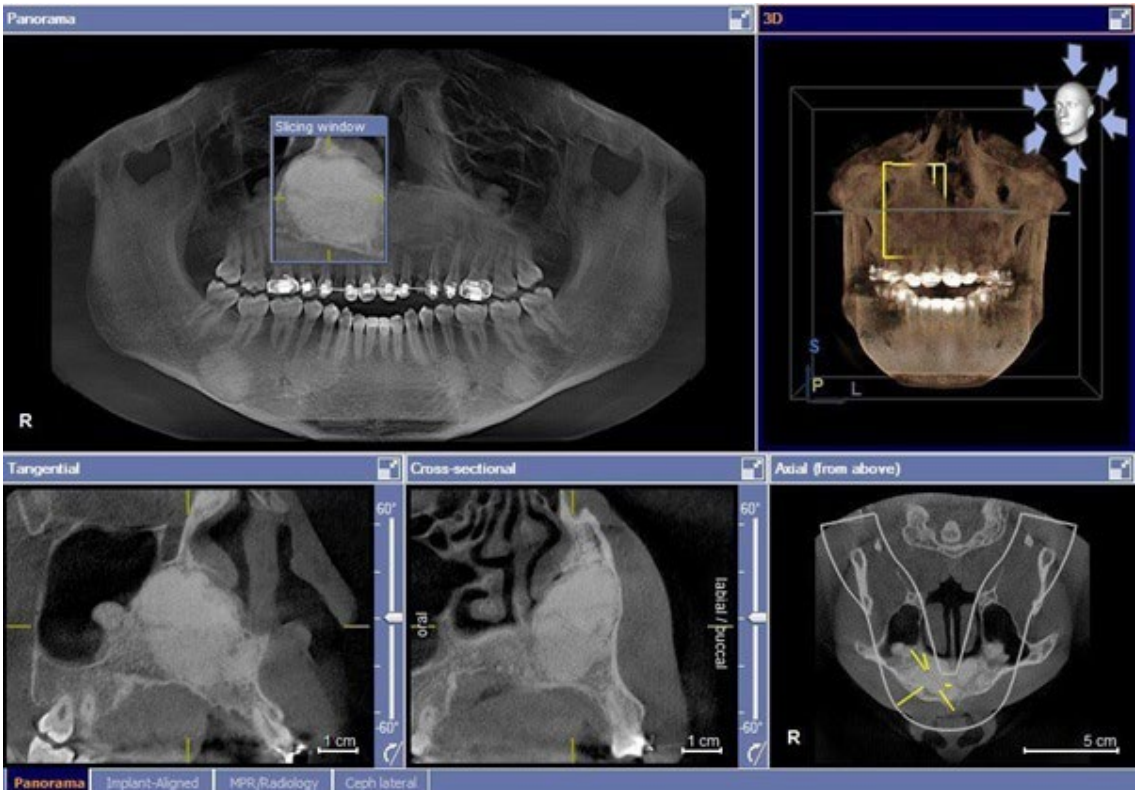

Figure 3. A screenshot of the CBCT taken in 2016 displaying the largest maxillary fibrous dysplasia lesion.

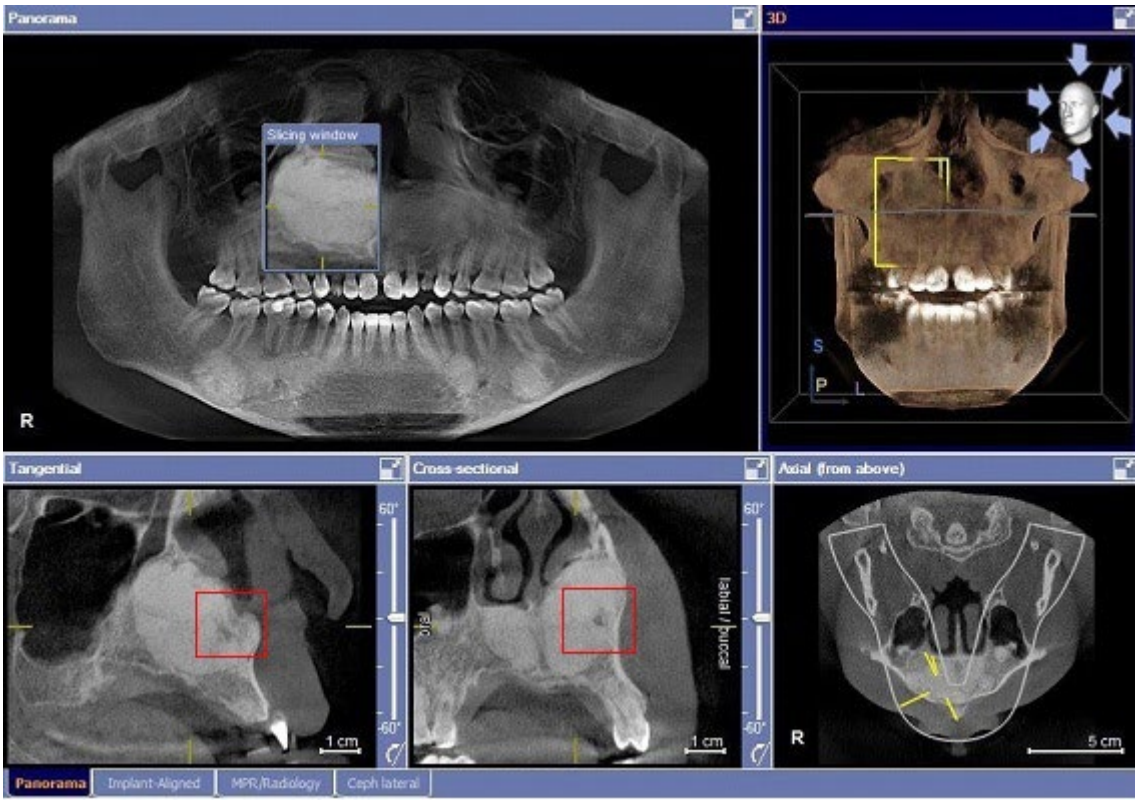

Figure 4. A screenshot of the CBCT taken in 2018 showing regression in the large maxillary fibrous dysplasia lesion (in red boxed area), when compared to the 2016 scan in figure 3.

decrease of bone turnover markers and a diminution of the incidence of pain and fracture. They compared the effects of pamidronate (old BPT) and zoledronic acid (new BPT) and reported no significant differences.[3] BPT was used intermittently for some participants in a study to treat chronic generalised bony pain, with positive results.[10]

In adulthood the focus is on issues relating to endocrine hyperfunction.[10] FD lesions usually stabilise during late adolescence or early adulthood. Occasionally they can grow into adulthood and may recur later in life. Adults with MAS experience problems with pain, mobility challenges and deformities.[1-3,10] These deformities can be linked to acromegaly, which is caused by an excess of the growth hormone.[1,9,10] This is a common manifestation affecting about 20-30% of MAS patients,[1,6] and is probably more frequent in males and almost always associated with craniofacial FD.[6] In adulthood, pain may increase due to skull deformities that are aggravated by acromegaly.[10] There have been reports of cancerous development in patients with MAS.[10]

CONCLUSION

The identification of the syndrome has exerted a considerable effect on the orthodontic management of the patient. Should the condition show remission and recovery of bone in the area, then there may be two choices. Firstly, remove maxillary deciduous canines and move the first premolars mesially to serve, after recontouring, as canines and plan to place implants to carry premolar crowns in the original sites of the first premolars. The second option is to accept the current occlusion, maintain observation, and consider treatment to close his open bite. That would almost certainly require orthognathic surgery, which may be contraindicated.

It seems that not much is known about the consequences of attempting orthodontic movement of the roots of teeth into and through the lesions of FD. The most sensible and pragmatic approach would be to avoid any stimulus which may precipitate further loss of bone. The advice to wait until there is clear evidence of stabilisation of the lesions should be respected, but even then, the assessment of how much tooth movement is indicated and whether this would involve an invasion of the fibrous tissue must be taken into account. Sequential radiographs, as were performed in this patient, are indicated to enable monitoring of the lesions. In this case, the first intimation of MAS was identified in routine pre-orthodontic radiological assessment. Subsequent monitoring by repeated PAN and CBCT meant that the condition could be tracked. In most cases some regression of the lesions is seen.[1,2,10] This was the position in this case, although not yet at a conspicuous level. Consultation with and guidance by an endocrinologist is advisable, especially should there be any indication of the development of acromegaly and/or other complications.

CONFLICT OF INTEREST

None to declare.

CONTRIBUTIONS OF THE AUTHORS

AA (Wits) did the radiography and review of literature. WE (Wits) provided information of orthodontic treatment and assisted in the write-up of the case report.

INFORMED CONSENT TO PUBLISH

Permission was obtained from the patient to use his records for this case report.

ETHICS APPROVAL

Approval was granted by the Human Research Ethics Committee of the University of Witwatersrand, Johannesburg. Certificate number – M190495.

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