International Journal of Health Sciences and Research
www.ijhsr.org ISSN: 2249-9571

Case Report

McCune-Albright Syndrome with Multiple Oral Manifestations- A Case Report

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ABSTRACT

McCune Albright syndrome is a rare disorder which constitutes three typical features: fibrous dysplasia, café-au-lait spots and endocrinal disorders. We report here a case of McCune Albright syndrome with all the three features and multiple oral manifestations. A 13 years old male patient presented with facial disfigurement since childhood. His physical examination revealed café-au-lait spots on right side of face and laboratory findings revealed endocrinopathies. No previously published case reports described multiple intra oral findings, this case report presented with expansion of maxillary and mandibular arch, macroglossia, enlargement of tongue papillae, painless soft tissue swellings on right buccal mucosa.

Keywords: McCune-Albright Syndrome, Fibrous Dysplasia, Café-au-lait spots, Oral manifestations, endocrinopathies, Fibro-osseous lesion.

INTRODUCTION

Fibrous dysplasia (FD) is a benign fibro osseous lesion in which normal substance of the medullary bone is replaced by fibro-osseous connective tissue, histologically characterized by varying degrees of osseous metaplasia. Considered hamartomatous, it results from mis-sense mutations in the gene (GNAS 1) coding for the α-subunit of the stimulatory G-protein in the guanine nucleotide binding, in chromosome 20q13.2-13.3 during embryogenesis. FD can be monostotic type (mutation occurs in only one bone constituting 80% of cases), polyostotic (several bones are involved), Craniofacial form (bones of craniofacial complex, involving sphenoid, zygomatic, frontonasal bones and the base of the skull involved) and McCune–Albright syndrome. McCune–Albright syndrome (MAS) is a sporadic, rarely occurring disease characterized by fibrous dysplasia of bone, café-au-lait skin pigmentation on the skin and endocrine disorders. [1] We present below a case of McCune Albright syndrome with a brief review of the same.

CASE REPORT

A 13 years old male patient presented with a chief complaint of facial disfigurement since childhood. Patient's parents gave a history of painless swelling being present since birth on right side of face which had gradually progressed to a size where it caused disfigurement of the face. Patient was diagnosed with lipoma on right cheek region for which patient underwent surgical excision at the age of 4.5 months. Swelling again appeared at the age of 7 years and a diagnosis of hamartoma was made for which patient underwent

another surgery. After 5 years patient again experienced increase in the size of the swelling leading to disfigurement of eye and ear. Patient also gave history of decreased hearing, diminution of vision in right eye and headache which was not associated with any aggravating and relieving factors. No significant relevant family history was present.

On clinical examination, a swelling measuring approximately 8x6x5cm noted over right side of face extending superoinferiorly from supraorbital region to submandibular region and anteroposteriorly from right lateral border of nose to parotid region. Skin over the swelling was intact. On palpation, the swelling was firm in consistency, non tender, non fluctuant with no local rise in temperature. A linear scar of approximately 7x2cm noted extending from right angle of mouth to right pre auricular area.

One large, asymmetric café au lait macule, brown in color with irregular borders was noted on right side of face on zygomatic and maxillary region. (Figure 1)

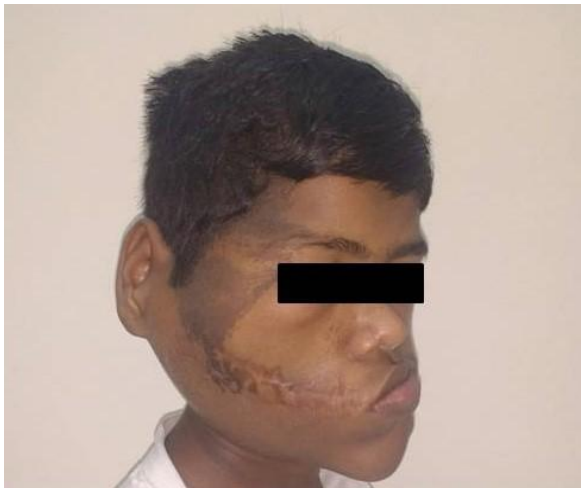

Figure 1- Facial disfigurement with Café-au-late macule

Lower third of face including lips and chin was found to be deviated to opposite side. Mouth opening was reduced and patient also gave history of difficulty in speech. Intra oral examination revealed two painless soft tissue swellings, non tender on palpation on right buccal mucosa; macroglossia with enlarged tongue papillae; expansion of maxilla and mandible on right side; spacing in maxillary and mandibular anterior teeth (Figure 2,3).

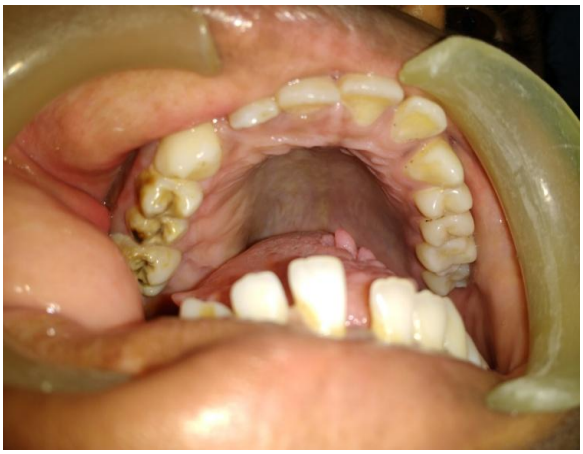

Figure 2- Soft tissue swelling, enlarged tongue papillae and expansion of maxillary arch

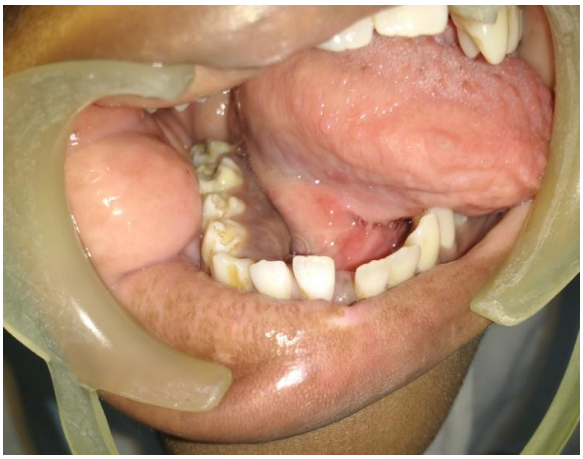

Figure 3- Expansion of mandibular arch

On whole body general examination patient appeared normal except testicular enlargement.

The laboratory findings related to hormones and electrolytes were as follows: serum prolactin, FSH, with normal values and LH, testosterone with decreased value; GH with increased value; T4, TSH with normal values and total T3 with slightly elevated value; PTH, creatinine, calcium, sodium, bilirubin total protein and urea with normal serum values, serum phosphate with elevated value; with normal hemoglobin level (13.2 gm %). Liver function test (SGOT, SGPT) were found to be within normal limits. Blood pressure was reported as 90/60 mmHg.

Panoramic radiographic findings were suggestive of irregular and sparse trabecular pattern; increased growth of right ramus of mandible, condylar and coronoid process, inferior displacement of mandibular canal, displacement of teeth, small and blunt teeth on right side, and also teeth on right side appeared magnified due to patient position and facial asymmetry (Figure 4).

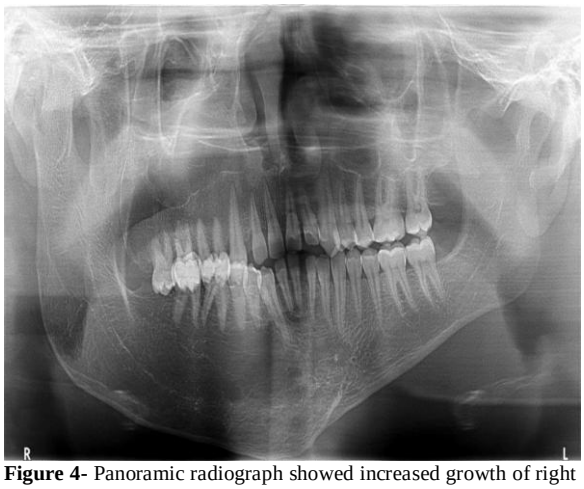

Figure 4- Panoramic radiograph showed increased growth of right ramus of mandible, inferior displacement of mandibular canal, displacement of teeth

Incisional biopsy of the skin lesion revealed hyperkeratosis, acanthosis, increased basal layer pigmentation and upward elongation of rete ridges in epidermis while dermis showed mild perivascular and periadnexal chronic inflammatory infiltrate composing of lymphocytes and plasma cells suggestive of epidermal nevus. Bone scan showed increased radiotracer uptake in the right orbital region, right maxilla and right temporal bone region.

CT head was performed for the patient and the findings were suggestive of thickening of the frontal bone on the right side, lateral and superior wall of the right orbit, right squamous temporal bone, zygomatic arch, superior wall of right maxillary sinus, coronoid process of right side of mandible with loss of cortico-medullary differentiation and its replacement by homogenous ground glass appearance, causing compression of the right maxillary sinus and right side nasal cavity suggestive of cranio facial deformity. Patient was referred to oral and maxillofacial surgery department for cosmetic recontouring.

DISCUSSION

FD is one of several fibro-osseous lesions that affect the maxillofacial region and three types of FD are: (1) Monostotic, involvement of one bone. (2) Polyostotic, in which multiple bones are involed.(3) MAS combining polyostotic FD, endocrinopathy, and cafe‑au‑lait spots.

In the present case, clinical features including multiple craniofacial bone involvement, café au lait macule on right side of face and endocrinopathies were suggestive of Macune Albright syndrome. Along with these extraoral and systemic features of macune Albright syndrome, this patient presented with intra oral manifestations including expansion of maxillary and mandibular arch, enlargement of tongue papillae, macroglossia and painless soft tissue swellings.

As on panoramic radiographs, the appearance of maxillofacial FD is variable, it becomes important for the clinician to be aware of all possible radiographic features and FD should also be taken into consideration in cases of lack of typical ground glass radiographic appearance. [2]

As observed in the present case, typical ground glass appearance was not present on the panoramic radiograph.

There is not a well established operative management of craniofacial dysplasia in MAS due to few case reports in long‑term follow‑up, with variable outcomes and timing of intervention. For mild deformities, careful follow-up of the patient during skeletal growth is recommended. Severe deformities require surgical remodeling of the affected bones which can be done for esthetic or functional purposes once the disease becomes dormant.

If needed, Shaping of the dysplastic bone can be repeated over time for postponing the radical surgery which consists of radical removal of dysplastic

bone and reconstruction with autologous bone graft. [3] The surgical approach of treatment depends on surgeon expertise and is not well-established, but the conservative approach with follow up is more adequate in McCune-Albright syndrome.

Most of the previous cases reported in literature about McCune Albright syndrome described only the typical extra oral features i.e. fibrous dysplasia, café-au-lait spots and endocrinopathy. Few reported cases showed oral manifestations associated with this syndrome such as Elif Ozsu et al in 2015 reported mandibular arch expansion and Aravinda K et al in 2013 reported expansion of both maxillary and mandibular arch. In this case, along with typical features of McCune Albright syndrome patient presented with oral manifestations also including expansion of maxillary and mandibular arch, enlargement of tongue papillae, macroglossia and soft tissue swellings. (Table 1)

AUTHOR	YEAR	EXTRA ORAL AND SYSTEMIC FINDINGS			ORAL MANIFESTATIONS				
		Café au Lait spots	*Fibrous dysplasia*	*Endo-Crinopathy*	*Expansion Of Maxillary arch*	*Expansion Of Mandibular arch*	*Enlargement of tongue papillae*	*Macro glossia*	*Soft tissue swelling*
Iulian raus et al [1]	2016				x	x	x	x	x
Elif Özsu et al [4]	2015				x		x	x	x
Lourenco R Et al [5]	2015				x	x	x	x	x
Jung-Hoon Noh et al [6]	2014	x			x	x	x	x	x
Aravinda k et al [7]	2013							x	x
Kollerova J Et al [8]	2013				x	x	x	x	x
Rustagi VT et al [9]	2011				x	x	x	x	x
Goswami M et al [10]	2009	x	x		x	x	x	x	x
Present report	2017								

Table 1 Showing various reports published in literature majority of which had only systemic findings.

CONCLUSION

McCune-Albright syndrome is a rare fibro-osseous lesion, associated with variable oral manifestations. Initial recognition of these manifestations may help facilitate better treatment and improve prognosis.

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How to cite this article: Mehra A, Karjodkar F, Sansare K et al. McCune-Albright Syndrome with multiple oral manifestations - a case report. Int J Health Sci Res. 2018; 8(2):297-301.

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