Case Report

Congenital infiltrating lipomatosis of the face with temporomandibular joint ankylosis

ABSTRACT

Congenital infiltrating lipomatosis of the face (CIL-F) is characterized by unilateral diffuse infiltration of facial soft tissue by mature adipose cells with associated skeletal hypertrophy. The disease is also considered a subtype of partial hemifacial hyperplasia. We present a case of right hemifacial swelling with severely restricted mouth opening diagnosed with CIL-F associated with temporomandibular joint ankylosis which is very rare, and very few case reports of the same have been published. Computed tomography findings have been discussed in detail with review of literature.

Keywords: Ankylosis, computed tomography, hemifacial hyperplasia, lipomatosis, temporomandibular joint

INTRODUCTION

Congenital infiltrating lipomatosis (CIL) is a subgroup of lipomatous tumors, etiology of which is unknown. It was first described as a distinct clinical–pathologic entity by Slavin *et al*.[1] The disease is characterized by diffuse infiltration of facial soft tissue by mature adipose cells, osseous hypertrophy, and a high risk of recurrence after surgery. It is usually found at birth or early after birth.[2] There have been few reports on CIL involving the face, but to our knowledge, only two other reports have been published, wherein the disease is associated with underlying bony ankylosis of the temporomandibular joint (TMJ).[3,4]

CASE REPORT

A 35-year-old patient presented with chief complaints of severely restricted mouth opening for 1 year. The patient had severe difficulty in food intake and could eat only via small gap between the teeth on the right side. There was progressively increasing swelling of the face on the right side since childhood. The patient got operated to debulk the soft tissue in adolescence for cosmetic purpose but again started developing swelling. Unfortunately, operative or histology records were not available. The patient was not much concerned about her esthetic appearance, but her main concern was her inability to open mouth and difficulty in food intake.

On examination, swelling was soft in consistency, extending from the preauricular region to the midline and from below the lower eyelid to the right submandibular region. Linear postoperative scar was seen in the lower part of the lump. Preauricular swelling was hard on palpation. Downward deviation of the right corner of the mouth with ipsilateral hypertrophied lips and deviation of the mouth toward contralateral side were seen [Figure 1]. All blood reports, chest X-ray, and abdominal ultrasound were normal. Neurological examination did not reveal any abnormality. No other cutaneous lesions were seen. Orthopentogram revealed the presence of marked expansion and remodeling of bones forming the right TMJ with associated TMJ ankylosis [Figure 2].

Ruchi Gupta[1,2], Sailesh Kumar Mukul[3], Prem Kumar[2], Amit Kumar[3]

Departments of [1]Radiology, [2]Radiodiagnosis and [3]Dentistry, AIIMS, Patna, Bihar, India

Address for correspondence: Dr. Ruchi Gupta, Department of Radiology, AIIMS, Phulwari Sharif, Patna - 801 507, Bihar, India.
E-mail: drruchigupta28@gmail.com

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Computed tomography (CT) scan was done using 256-slice scanner. There was enlargement of foramen ovale, greater palatine foramen, sphenopalatine foramen, inferior orbital foramen, and nasolacrimal duct on right side. [Figure 3]. CT revealed fatty infiltration of the cheek, masticator space, parotid gland, and muscles including masseter, medial and lateral pterygoid, and temporalis muscle on right side. Fatty infiltration was also seen in the upper and lower lips and right half of the tongue [Figure 4]. Volume-rendered image revealed bony ankylosed mass involving the right TMJ [Figure 5].

Surgical excision of ankylosed mass was done along with temporalis muscle interposition by the department of oral maxillofacial surgery [Figure 6]. The main purpose of surgery was to remove the ankylosed mass so that the patient could open her mouth and eat properly. The patient's esthetic appearance was not of primary concern, and the patient was counseled for another staged surgical procedure for esthetic correction.

DISCUSSION

Slavin *et al*. had first reported three cases of infiltrating lesions of adipose tissue involving the face in 1983.[1] They described the following histomorphological findings: infiltration of

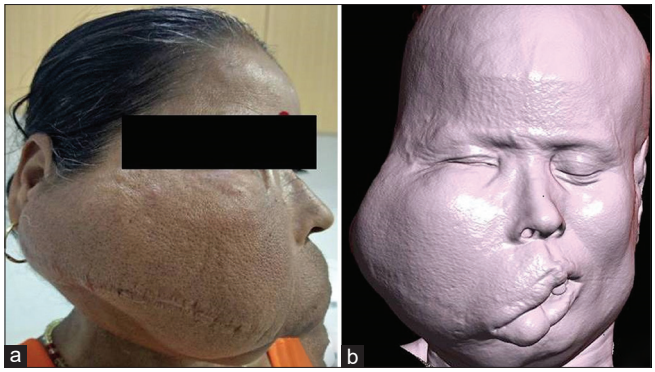

Figure 1: (a) Side profile of the patient showing right hemifacial swelling extending from the preauricular region to the upper and lower lips on the same side. Linear postoperative scar is seen in the lower part of the swelling. (b) Shaded surface display image showing right hemifacial swelling, ipsilateral enlargement with drooping of the lips, and deviation of the mouth toward contralateral side

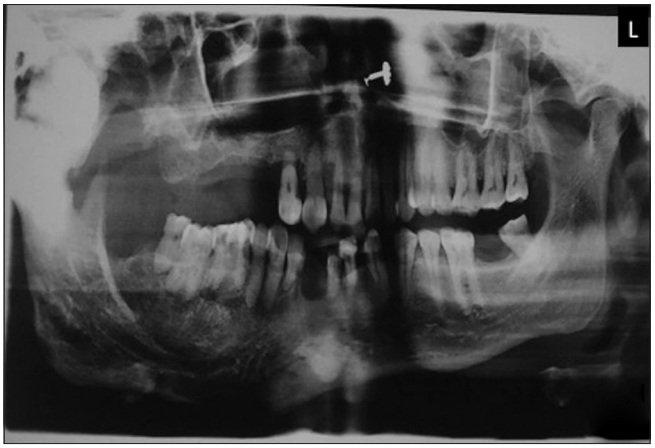

Figure 2: Orthopentogram shows right bony temporomandibular joint ankylosis, lack of structural organization, and loss of temporomandibular joint space with large mass of bone extending toward the skull base and into the genial notch. Left temporomandibular joint appears normal

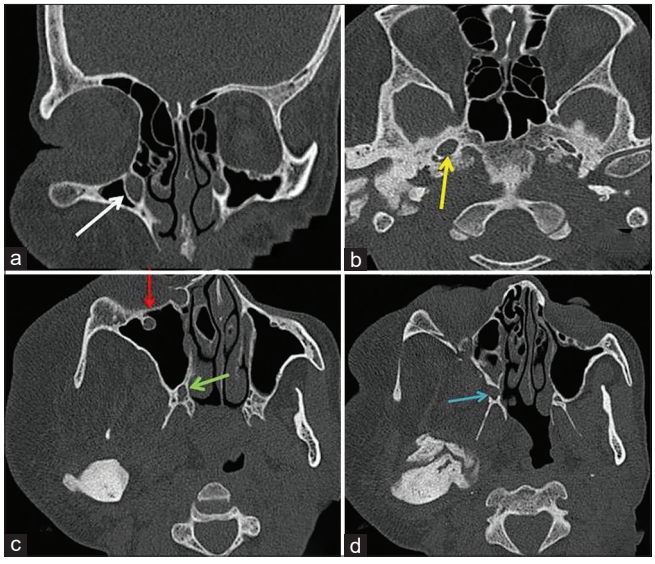

Figure 3: (a) Coronal computed tomography reformatted image (bone algorithm) shows enlarged right nasolacrimal duct (white arrow), (b) axial computed tomography image (bone algorithm) shows enlarged right foramen ovale (yellow arrow), (c) enlarged right inferior orbital foramen (red arrow) and greater palatine foramen (green arrow), and (d) enlarged right sphenopalatine foramen (blue arrow)

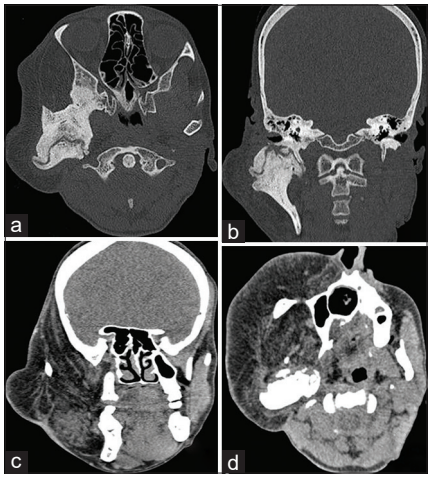

Figure 4: (a) Axial and (b) coronal computed tomography images (bone algorithm) show marked expansion with new bone formation, remodeling with articular surface irregularity of the right condylar head and temporal bone forming the glenoid fossa with the right temporomandibular joint ankylosis. There is an expansion with sclerosis of the right greater wing of the sphenoid bone and (c) coronal and (d) axial computed tomography images (soft tissue window) show fatty infiltration of the right side of the cheek, masticator space, buccal space, parotid gland, masseter, medial and lateral pterygoid muscles, upper lip, and right half of the tongue

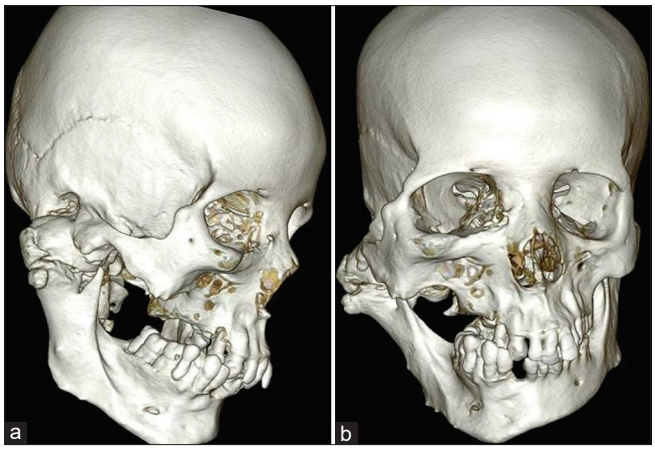

Figure 5: Volume-rendered images (a) and (b) show hypertrophied-ankylosed bony mass involving the right temporomandibular joint with loss of structural organization and preserved sigmoid notch. On the right side, the upper molar and premolar teeth are absent. Rest of the teeth are malaligned

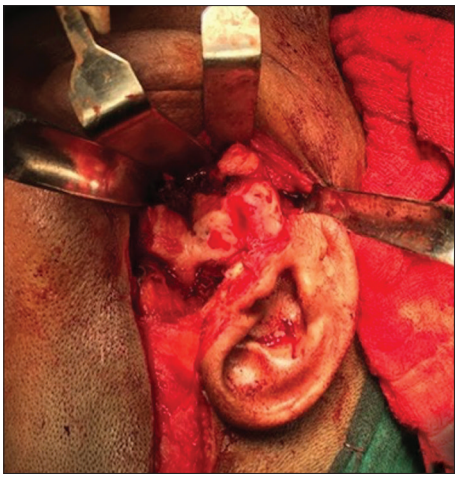

Figure 6: Intraoperative image of the patient showing excision of the ankylosed bony mass

fat into the adjacent soft tissue and hypertrophy of the underlying skeleton, absence of malignant characteristics and lipoblasts, presence of fibrous elements, increased number of vessels with unifocally thickened muscular walls, and increased number of nerve bundles of variable sizes with focal fibrosis.

The clinical and CT findings of this study were correlated with Sahai *et al*. and Keramidas *et al*. studies.[3,4] Diffuse right hemifacial fatty infiltration associated with right TMJ involvement in the form of TMJ ankylosis with marked bony expansion and remodeling was seen in both studies due to which there was a severely restricted mouth opening consistent with TMJ ankylosis Type II.[5] In our study, there is also enlargement of ipsilateral foramen ovale, inferior orbital foramen, nasolacrimal duct, greater palatine foramen, and sphenopalatine foramen. Ringrose *et al*. described that the enlargement of cranial and facial skeleton foramina accompanies the hyperplasia of facial bones.[6] Foraminal enlargement is used to differentiate congenital hemifacial hyperplasia (HFH) from other conditions that cause overgrowth of the facial skeleton.[7]

Meckel *et al*. had first described HFH in 1822.[8] The disease is characterized by marked unilateral development of bony and soft tissues of the face. HFH was classified into true HFH (THFH) and partial HFH (PHFH) by Rowe *et al*.[9] They described THFH as unilateral enlargement of all viscerocranial structures, and PHFH does not include enlargement of all structures. Bou-Haidar *et al*. suggested that congenital hemifacial lipomatosis may be considered as a subtype of PHFH.[10] However, lipomatosis was a dominant feature in their report with the lack of significant muscular hyperplasia. However, in our case, both muscular and skeletal enlargements were equally present.

Differential diagnosis, in this case, included lipoma, liposarcoma, lipoblastomatosis, and congenital overgrowth syndromes causing progressive HFH.[2] Lipoma is an encapsulated tumor with thin fibrous strands. However, in our case, the fatty infiltration was diffuse and seemed to be unencapsulated. Lipoblastomatosis was excluded since it occurs exclusively in infancy and early childhood and shows the presence of lipoblasts and signet ring cells. The presence of bony hypertrophy and remodeling is a chronic process; hence, malignancy was also excluded by the clinical presentation of long history. Liposarcoma was also excluded by the absence of lipoblasts and pleomorphism on histopathology. Cutaneous signs are usually present in diseases such as infiltrating angiolipoma or facial angioma (Klippel–Trenaunay syndrome and Sturge–Weber syndrome) and neurofibromatosis with its typical café au lait spots.[11] Once these differentials were ruled out, then the diagnosis of CIL was made based on clinical presentation, classical CT findings, associated cranial foraminal enlargement, and histopathological findings.

In the early years, authors suggested wide and early surgical excision.[1,12] Recent literature supports a more conservative approach with the addition of temporary measures such as liposuction and elevation of the drooping upper lip.[4]

CONCLUSION

CIL of the face, a subtype of PHFH, is characterized by diffuse infiltration of facial soft tissue by mature adipose cells, the diagnosis of which is established the diagnosis of which is established by clinical presentation, imaging studies and histopathologically and histopathologically and

characterised by bony hypertrophy, diffuse hypertrophy of the soft tissues of the cheek, underlying muscles, ipsilateral enlarged hemitongue, facial skeletal foraminal enlargement, and macrodontia. Surgical debulking of soft tissue is the treatment of choice; however, frequent relapses mar the eventual success of surgery. In case of associated TMJ ankylosis, the removal of ankylosed mass is the treatment of choice.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given her consent for her images and other clinical information to be reported in the journal. The patient understands that name and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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