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CONGENITAL INFILTRATING LIPOMATOSIS OF THE FACE – AN UNUSUAL CASE OF FACIAL ASYMMETRY

ARVIND ANAND [1], VAGISHA DAHIYA [2] and PARNIT KALRA[2]

[1] Professor, Department of Radiology, School of Medical Sciences and Research, Sharda University, Greater Noida, India.
[2] Junior Resident, Department of Radiology, School of Medical Sciences and Research, Sharda University, Greater Noida, India.

Abstract

A young female patient was referred to the department of Radiology for CECT scan. The patient had marked facial asymmetry along with a soft, painless swelling over the right side of the face progressively increasing in size. The unilateral macroglossia and diffuse swelling on right side of the face showed fatty infiltration on CT imaging. These features are classical of Congenital Facial Infiltrating Lipomatosis. The disease is sporadic in nature. Adipose tissue is embryologically derived from the neural crest, as are the membranous bones (orbits, mandibles, zygomatic bones) The infiltration appears inhomogeneous, with a feathery appearance diffusely infiltrating, non-encapsulated lesion of fat attenuation. Our case showed involvement of the right zygoma and orbit.

Keywords: Infiltrating Facial Lipomatosis, Facial Asymmetry, Macroglossia, Case Report.

INTRODUCTION

Congenital diffuse infiltrating lipomatosis of the face is an uncommon clinical entity, characterized by unilateral aggregation of non-encapsulated, mature lipocytes which infiltrate the surrounding tissues [1, 2].The term lipomatosis refers to extensive and abnormal accumulation of fatty tissue which is not limited by tissue planes, most often seen in the trunk or extremities, and very rarely involving the head and neck [3].Gradual enlargement of the lesion and the resultant facial asymmetry are highly specific clinical features for the condition [5]. The concerns are mainly aesthetic, with no neurological deficits reported in association. Effective management remains a challenge, due to the high recurrence rate following surgical removal [8]. Although first described in 1983 by Slavin et al [12], very few cases have been reported in medical literature, owing to the low incidence of the condition [13].

Case Report

A 22 year old female patient was referred to the department of Radiology for CECT scan. The patient had marked facial asymmetry along with a painless swelling over the right side of the face, present since birth which was gradually increasing in size.

The general physical examination and the vitals were unremarkable.

Physical examination revealed a prominence of the right cheek, with upwards displacement of the right eye. On oral examination, marked prominence of tongue on the right side was noted along with hypopigmentation and few mucosal protuberances (Image 1).

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On palpation, the right sided tongue mass was soft, non-compressible and non-pulsatile. It was non-tender with no evident local rise of temperature. Oral mucosa was normal. Sensations were intact with no motor abnormality. No cervical lymphadenopathy was present.

A CECT scan of the face was performed on a 128-slice

multi-detector CT scanner (OPTIMO WIPRO GE HEALTHCARE). The sections at inferior plane revealed diffuse and unencapsulated fatty infiltration of the tongue, giving a feathery appearance (Image 2); and also in the subcutaneous plane over the right side of the face (Image 3).The superior sections at the level of retromolar trigone revealed hemi hypertrophy of the right side of the tongue with prominenent convexity at superior margin. Hypodensity of fat attenuation (HU~60) was seen which was limited to the right side of the tongue with a sharp line of demarcation at the midline. (Image 4).

Hypodensity of fat attenuation is also seen in the right parapharyngeal space medial to the neck of the mandible (Image 5, 6).

The fat planes with adjacent gingival buccal structures were maintained and the underlying vascular structures were normal.

The volume rendered images showed facial asymmetry with relative bony hypertrophy on the right side involving the zygoma and lateral wall of right orbit (Image 7, 8).

No abnormal uptake of contrast was noted on contrast-enhanced images.

The above CT findings were consistent with the clinical features of the patient.

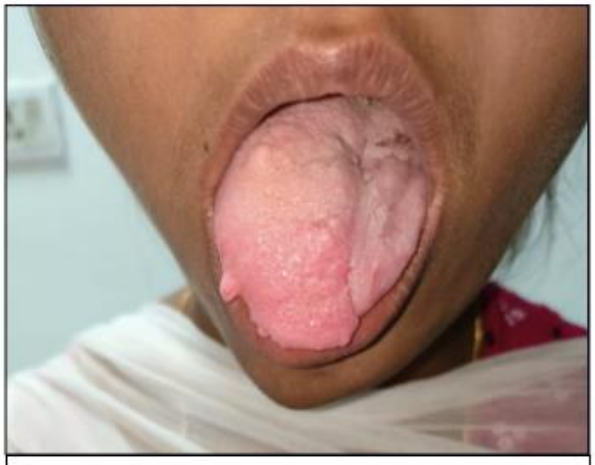

Image 1: Marked prominence of tongue on the right side.

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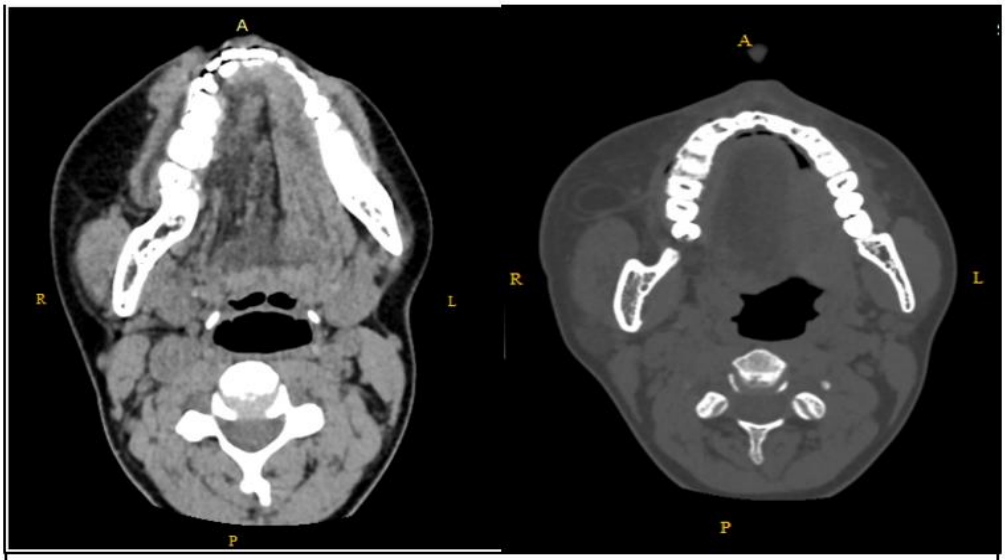

Images 2 and 3: Diffuse and unencapsulated fatty infiltration giving feathery appearance seen in the subcutaneous plane over the right side of the face

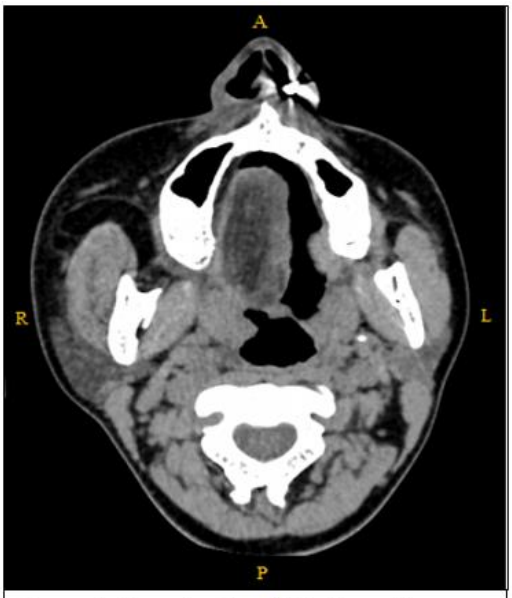

Image 4: Hypodensity of fat attenuation (HU~60) seen limited to the right side of the tongue with a sharp line of demarcation at the midline.

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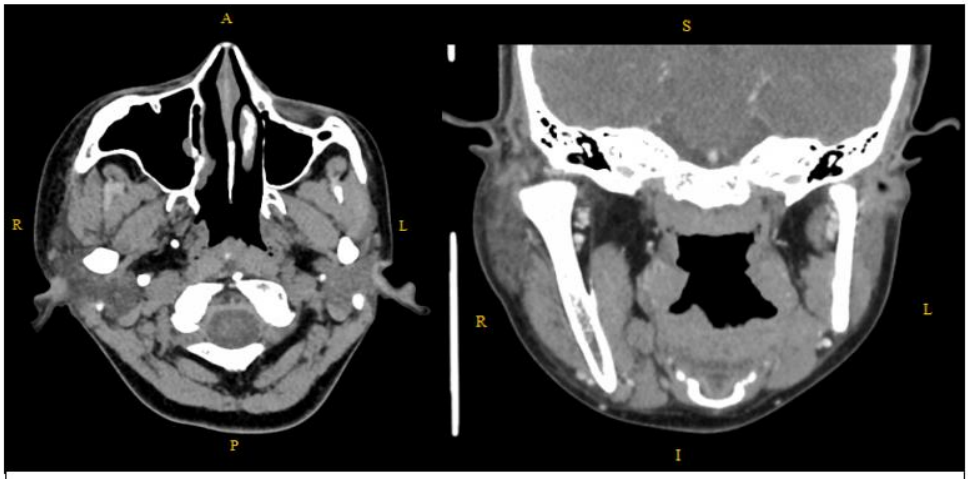
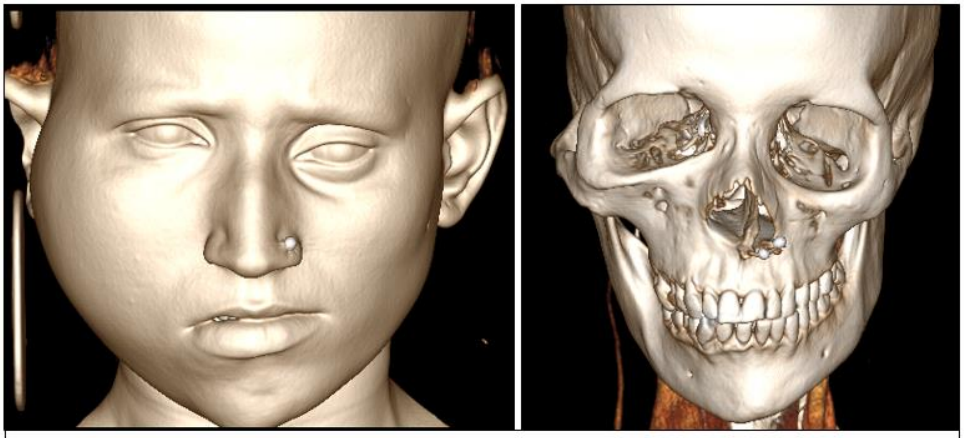


Images 5 and 6: Hypodensity of fat attenuation in the right parapharyngeal space medial to the neck of the mandible.



Images 7 and 8: Volume rendered images showing facial asymmetry with relative bony hypertrophy on the right side.

DISCUSSION

Lipomas are common benign tumours of adipocytes which are mesenchymal in origin, presenting as well-circumscribed, encapsulated doughy masses often in the subcutaneous plane [1]. Lipomatoses are extensive, poorly circumscribed, benign proliferations of mature adipocytes. These are most often found to involve the neck, pelvis, limbs or the trunk [1, 2, 3].

Congenital Facial Infiltrating Lipomatosis (C-FIL) is described as a congenital, non-hereditary condition wherein the histopathological features seen are a non-encapsulated mass of mature adipocytes, fat infiltration in surrounding muscles and soft tissue, absence of malignant characteristics with no lipoblasts, presence of fibrous elements, and increased numbers of nerves and vessels [10].

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As opposed to liposarcoma and lipoblastomatosis which contain immature adipocytes, facial infiltrating lipomatoses show infiltration of normal tissue by mature adipocytes. Infiltrating lipomatosis has been reported on the cheek, lip, floor of the mouth, buccal sulcus, tongue, and parotid gland [4].

A case of facial infiltrating lipomatosis commonly presents with a diffuse large swelling on one side of the face that is soft, painless and progressively increasing in size. Unilateral hypertrophy of soft tissues of the face, most commonly the cheek is seen, secondary to underlying fat infiltration and skeletal overgrowth. Other possible findings include macrodontia and early eruption of deciduous and permanent teeth on the affected side, macroglossia, and mucosal neuromas which are seen as protuberances on the tongue and buccal mucosa. Thus clinical examination of the oral cavity is warranted in a case of suspected C-FIL [3,4,5].

The disease is sporadic in nature, and while the exact etiopathogenesis is not well understood, somatic mutations in phosphatidylinositide-3-kinase (PI3K) signaling pathway have been identified in affected tissues [9]. The disease may be considered a neurocrisotpathy.

Adipose tissue is embryologically derived from the neural crest, as are the membranous bones (orbits, mandibles, zygomatic bones) which are often contiguously involved [10].

Other possible causative mechanisms include trauma, metaplasia, chronic irradiation, congenital cytomegalovirus infection, and Chromosome 12 abnormalities [11].

Radiological imaging findings include hypertrophy of facial bones and soft tissue swelling.

Computed tomography typically shows a diffusely infiltrating, non-encapsulated lesion of fat attenuation. The infiltration appears inhomogeneous, with a feathery appearance given by the intervening fibrous tissue component.

Ipsilateral and asymmetrical hypertrophy of the underlying bony and adjacent soft tissue structures is also often seen.

Magnetic resonance imaging is highly specific for identification of fat tissue and in identifying the exact extent of the infiltration. Infiltration of unencapsulated fat in subcutaneous or adjacent soft tissue is seen, with characteristic T1- and T2-hyperintense signal that suppresses at fat-suppressed sequences. [1, 2, 3, 4, 5, 7, 11, 12, 13].

Our diagnosis of Congenital Facial Infiltrating Lipomatosis was based on the clinical symptoms, presentation of the patient and classical CT imaging findings. The patient was unwilling for MRI and hence we are unable to present our MRI findings.

The differential diagnoses include the Proteus syndrome, a rare sporadic condition which is characterized by atypical skeletal growth, with features consisting of asymmetrical limb growth, epidermal nevi, parotid or ovarian tumors, and disregulated adipose tissue manifesting as lipomas or regional lipohypoplasia [6]. Encephalocutaneous lipomatosis is another differential, which is characterized by lipomas of ectodermal tissues including skin, eyes and central nervous system [7].

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As the infiltration is extensive and often involves multiple indispensable facial structures, a complete surgical resection is rarely achievable, leading to a high recurrence rate [8]. Debulking surgical procedures are mainly performed to improve the cosmesis of the patient, and eradication is not the primary goal. Liposuction may also be done for aesthetic reasons. Malignant transformation of infiltrating lipomatosis has not been documented, thus a conservative approach for the management of these patients is being increasingly employed [8,11,13].

Citations

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