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Infiltrating Lipomatosis Of The Face In A Temporofacial Localisation: A Case Report And Literature Review

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Abstract
Infiltrating lipomatosis of the face (FIL) or Congenital Infiltrating Lipomatosis of the Face (CLIF) is a rare clinico-pathological entity characterised by congenital hypertrophy of the face in which mature adipose tissue infiltrates soft tissue structures on one side, with muscle involvement and associated bone hyperplasia in the other side. This causes considerable facial asymmetry and can alter essential facial functions such as swallowing, chewing, vision and breathing. Due to the diffuse infiltration and the involvement of important facial structures, complete surgical excision is often impossible, with a high recurrence rate.
We present a case of infiltrating congenital lipomatosis of the face who had swelling on the right side of the face since birth.

Keywords: *Congenital, lipomatosis, facial asymmetry, PIK3CA,*

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I. Introduction

Infiltrating lipomatosis of the face (FIL) represents a significant challenge in clinical management due to its rare occurrence and complex presentation. The condition is characterized by the infiltration of mature adipose tissue into facial soft tissue structures, often leading to considerable asymmetry and functional impairment. Surgical intervention, typically in the form of reduction surgery, is the primary treatment modality, although complete excision is rarely possible due to the diffuse nature of the infiltration and the involvement of critical facial structures. The recurrence rate post-surgery is notably high, emphasizing the need for ongoing clinical evaluation and management. Advances in genetic understanding have identified mutations in the PIK3CA gene as a significant pathogenic factor, which opens potential avenues for targeted therapies in the future. The complexity of FIL requires a multidisciplinary approach to optimize patient outcomes and manage the physiological and psychological burden imposed by this condition.

II. Case Report

A 49-year-old woman, married and mother of two children, without any personal or significant family medical history, was hospitalized for the gradual swelling of the right hemiface since birth, resulting in facial asymmetry.

The clinical examination revealed facial asymmetry and swelling on the right side of the face extending to the temporal region. The skin above the swelling was normal and there was no discoloration. On palpation, the mass was of soft, non-sensitive, non-pulsatile, non-compressible with unclear demarcations. It measured approximately 12 cm and occupied the temporal and pre-auricular region; anteriorly pushed the right external canthus, sparing the eyelid, and downwards it was limited by the zygomatic arch, which was protruding. (**Fig.1)**

The mouth opening was limited at 3 cm, with vestibular filling at the level of the retromolar trigon, and its dental articulation was class II. There was no cervical lymphadenopathy. During the disease, the patient developed blindness of the right eye at the age of 39 years, associated with convergent strabismus and limited eye movement.

A CT-Scan and an MRI performed showed the presence of lipomatous infiltration in the deep spaces of the right hemiface, as well as in the right masticatory, pterygoid, and temporal muscles.it was associated with a bone remodeling and hypertrophy of the hemi-maxilla, hemi-mandible, and zygoma. Additionally, there was an impact on the maxillary and mandibular dentition on the right side. There was also hypertrophy of the right orbital space and a mucoid cyst with right parasphenoid development filling the orbital apex, which compressed the optic nerve and the lower and internal oculomotor muscles. At the cerebral level, there was no intracerebral expansive process. (**Fig. 3-4)**

The patient gives her consent for treatment after receiving all the necessary information and a surgical reduction of the jugal and temporal component was done associated with osteotomy to reposition the zygomatic arch. Histopathological examination shows the presence of mature adipose tissue infiltrating the muscle without any cellular atypia suggestive of malignancy. These results confirmed the diagnosis of lipomatosis infiltrating the face.

The outward appearance has demonstrably improved following surgical treatment. During a six-month follow-up period, no evidence of disease recurrence or metastasis was observed. Long-term follow-up will be conducted to obtain more information about this rare disease. **(Fig.2)**

III. Discussion

Congenital lipomatosis infiltrating the face (CILF) is a rare, congenital, non-hereditary facial proliferation first described in 1983 by Slavin et al. [5].

Currently, with the discovery of postzygotic mosaic activating mutations in the PIK3CA gene of CILF, it is part of PIK3CA-related proliferation syndrome [10].

In clinical settings, CILF presents as a unilateral, progressive, and painless enlargement of the soft tissue and bone structures of the face, occurring at birth or in early childhood (before one year of age). Notably, there is no sexual or secondary predilection.

Due to its extensive and infiltrating growth pattern, it can be associated with oral problems such as macrodontism, early eruption of baby and permanent teeth, and facial deformities, which have a significant social and psychological impact [11].

The diagnosis of CILF is essentially clinical. Imaging studies such as simple X-rays show enlargement of the facial bones and swelling of the soft tissues. [12]

Computed tomography (CT) and magnetic resonance imaging (MRI) are the imaging modalities of choice for the diagnosis of CILF because they can identify the fat content of lesions and delineate their extent. The CT scan shows fat density lesions in exquisite detail. The intermediate fibrous elements may result in a feathery pattern or an inhomogeneous character. [3, 14]

MRI, with its multiplanar capability, is superior to CT scans. It can describe the exact extent of the lesion. On T1W images, the lesions are hyperintense inhomogeneous. The best way to observe muscle and bone damage is through MRI.[19]

A biopsy or excision of the tissue can be performed to rule out other possible differential diagnoses in the facial area, such as lipoma, lipoblastoma, or liposarcoma. If the typical findings are seen on MRI, a biopsy may not be necessary. [12]

Histologically, CILF is characterized by an unencapsulated mass composed of mature adipose tissue without atypia, mitosis, or lipoblasts, with intermediate fibrous tissue septa that infiltrate adjacent muscles and soft tissues, with associated hypertrophy of the underlying bone, making complete excision extremely difficult [13].

Singh et al. observed that the disease appears to follow two distinct patterns of progression. One is a rapidly progressive form, which manifests in the first year of life, while the other is a more indolent form that progresses over decades. [2]

Consequently, the management of the disease depends on the speed of its progression. In early reports, Slavin et al. [5] and Kang et al. [15] advocated for a broad and early local excision, while Van Wingerden et al. [8] suggested a delayed resection to avoid facial nerve damage and unnecessary surgery. More recently, Kamal et al. [6] noted that delaying surgical procedures may provide the opportunity to observe a matching contralateral mature cheek during surgery.

Some articles also suggest liposuction and surgical excision as treatment [16, 1, 7]. Although considered benign, CILF has a high recurrence rate after excision (up to 62.5%) [3], requiring multiple surgeries for cosmetic reasons [18, 6]. In some cases, reduction of the maxilla, mandible, and zygomatic bone has been performed [16, 1,7]. Tracy et al. [13] proposed a combination of surgical resection and specific targeted chemotherapy. Imatinib and celecoxib were used as personalized targeted chemotherapy to manage CDIL. Evaluation of this treatment protocol demonstrated synergistic effects to improve facial symmetry and control disease progression.

Surveys about the evolution of CILF have reported no malignant features over 2 to 14 years of follow-up [13]. Furthermore, the involvement of the PIK3CA gene mutation in CILF renders PI3K inhibitors, such as alpelisib, which are currently being studied in clinical trials for many cancers, including lymphoma, a potential treatment option for CILF patients [10].

IV. Conclusion

Infiltrating congenital lipomatosis of the face is a rare benign disorder of childhood onset. It is characterized by diffuse infiltration of fat into the subcutaneous and muscle planes associated with bone hypertrophy. Clinical examination and imaging, including MRI, are used to make the diagnosis. PIK3CA mutations are a key pathogenic mechanism responsible for the development of this pathology. Surgery remains an important approach to correcting maxillofacial deformities, with the specific timing and procedure depending on the unique presentation of each individual case.

Restoration of complete facial symmetry is unlikely, and the patient should be informed of the high rate of recurrence.

The effectiveness of surgical treatment, adjuvant therapy, and psychological intervention is important to both patients and clinicians. Therefore, proper collection and analysis of reports on this disease is very important to help us learn more and do more for those patients.

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Fig.1 Facial and lateral view before surgery

Fig.2 Facial and lateral view after surgery

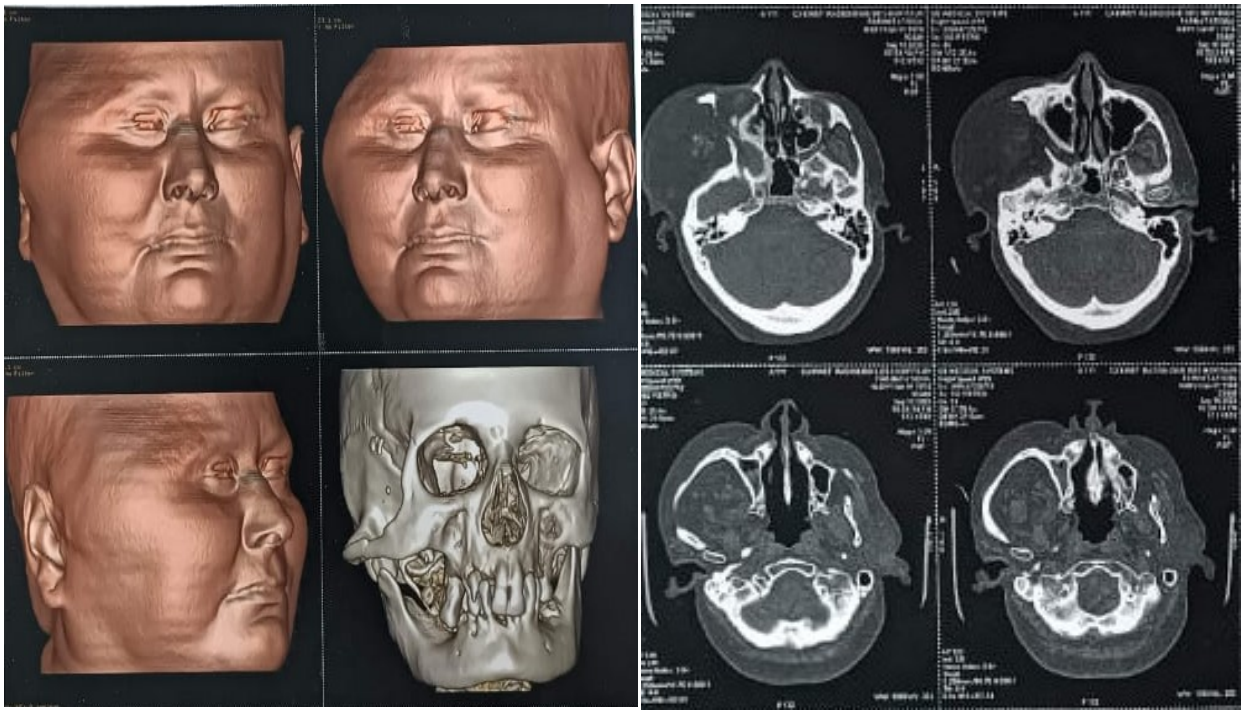

Fig 3. On the left: CT scan 3D reconstruction of soft tissue and bone
On the right: CT scan axial osseus view showing the enlargement of the infratemporal fossa and the deformity of the zygomatic arc

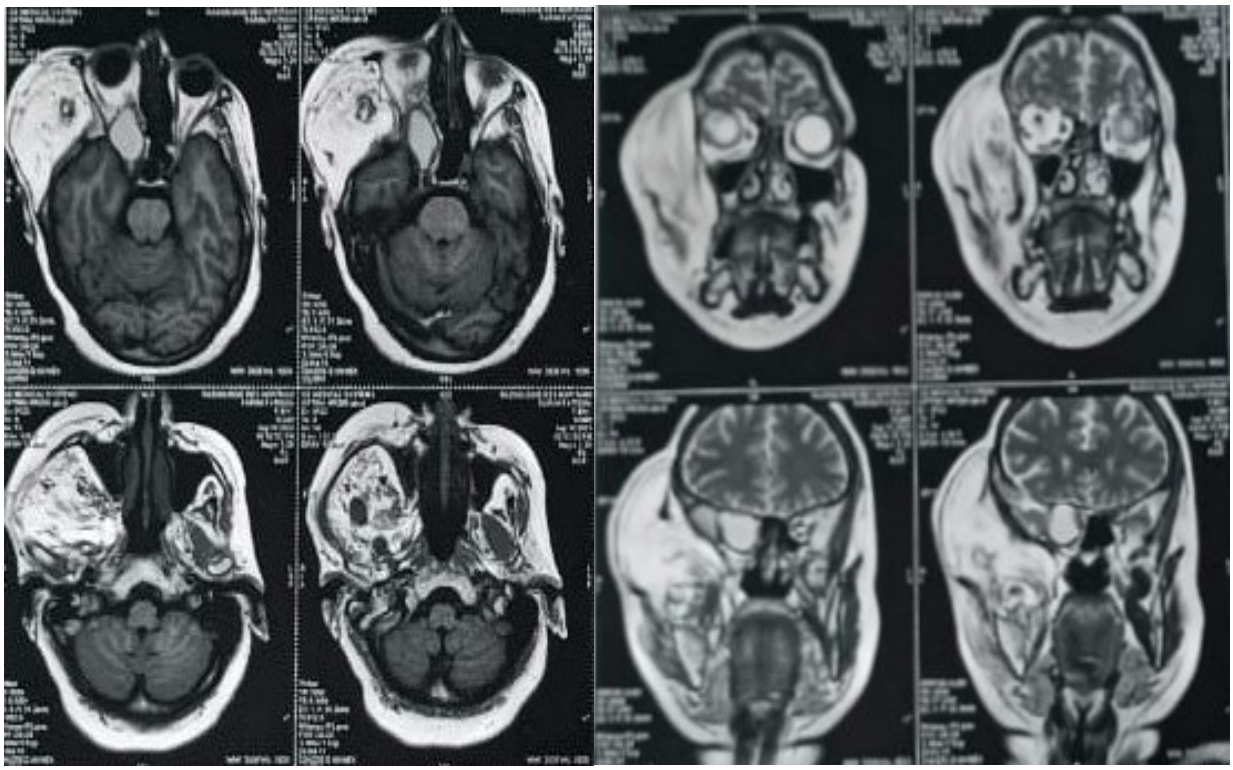

Fig.4 MRI axial and coronal views showing the mass in his infratemporal localization.