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Case Report

Congenital infiltrating lipomatosis of the face: Case report ☆,☆☆

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ABSTRACT

A rare disorder called congenital infiltrating lipomatosis of the face is characterized by a diffuse fatty infiltration of the soft tissues of the face. Muscle involvement and related bony hyperplasia may coexist. This particular form of lipomatous tumor is rare and typically appears in childhood. It is congenital in nature. Collections of mature, nonencapsulated lipocytes that infiltrate local tissues and frequently recur following surgery define congenital infiltrating lipomatosis, a unique clinicopathologic condition. The best modalities are, CT scan and MRI since they can determine the extent of the lesions and reveal their fat content. The treatment of this disease is surgical as soon as possible to restrict their infiltrative growth and improve the appearance of the face. We describe a case of massive facial invading lipoma that was investigated using MRI.

Introduction

An uncommon condition known as congenital infiltrating lipomatosis of the face is caused by groups of mature, nonencapsulated lipocytes that invade nearby tissues and cause craniofacial deformities [1].

All cases reported in the literature have been of patients in the first 3 decades of life, with the majority showing up as unilateral cheek or chin swelling in infancy as a result of hypertrophied tissues that are still working normally [2].

CT can show the mass's lipomatous character, its connection to other structures, and any osseous changes. There is visible diffuse infiltration of adipose tissue (−60 HU to −120 HU) and hyperplasia of the maxillofacial bones. Additionally, MRI is highly specific in identifying fat tissue and offers a more accurate indication of the degree of infiltration [3].

There are 2 available treatment options: excision and liposuction. This is being done for aesthetic purposes. Despite being benign, these tumors have an extremely high recurrence rate following surgical removal [1].

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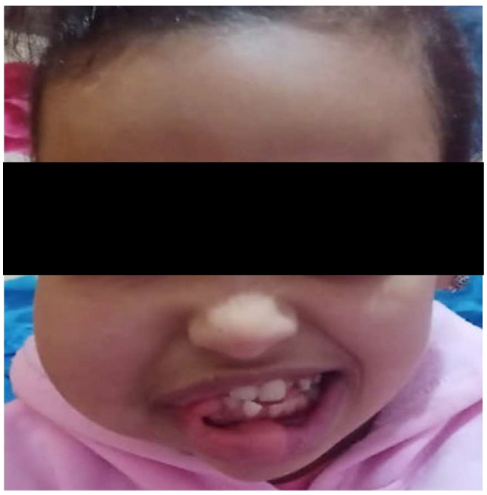

Fig. 1 – Facial asymmetry with enlargement of the entire right cheek and lip.

Case

A 9-year-old girl presented with swelling of the right side of the face since birth that had been progressively increasing in size (Fig. 1). There was no history of pain.

A painless, nontender soft mass with ill-defined borders was seen during a clinical examination. It covered the entire cheek, the lower half of the parotid and masseteric regions, and part of the submandibular region. The skin covering the mass was normal, showing no evidence of discoloration or abnormal vascularization. The lump was firm and fixed.

Cervical lymphadenopathy was absent.

The blood examination of this patient showed nothing special.

The lesion showed a nonenhancing, ill-defined, infiltrative, inhomogeneous signal intensity on T1-weighted images (Fig. 2A) in the subcutaneous, muscular, and intermuscular planes of the face and upper neck on the right side, with a moderate decrease in signal intensity on T2-weighted images (Fig. 2C). Additionally, there were irregular linear hypointensity regions. T1-weighted images with fat suppression showed partial suppression of the lesion, corresponding to fat (Fig. 2B). The lesion had fuzzy edges and no obvious cleavage plane between the mass and surrounding muscles, especially the hypertrophied and infiltrated temporalis and masseter muscles.

A radiological diagnosis of congenital infiltrating lipomatosis of the face was made.

The patient received surgical treatment: reduction of the fat mass (Fig. 3).

Discussion

Slavin et al. [2] initially identified infiltrating lipomatosis of the face as a unique clinical-pathologic entity. It is a congenital condition in which mature adipocytes or lipocytes penetrate neighboring tissue, both soft and hard, in the facial region. Rational treatment for congenital infiltrating lipomatosis requires knowledge of its natural history. It is important to note that recurrence and invasion may serve as distinguishing behavioral traits. Kauffman and Stout believed that mature lipocytes had the capacity with infiltrate local tissues; lipoblastomatosis, which is composed of embryonal lipoblasts, is not associated to local infiltration [4,5].

A somatic mutation in the PIK3CA gene, which is also found in malignancies and afflicted tissues from other nonheritable and overgrowth illnesses, is the current pathogenic theory. The protein PIK3, which is encoded by PIK3CA, is essential for controlling the motility, adhesion, survival, and proliferation of cells [6].

CILF and kindred benign overgrowth syndromes are known as PIK3CA-related overgrowth syndromes because postzygomatic activation mutations in the PIK3CA gene have been found in them [7]. The gene PIK3CA encodes p110α, a catalytic PI3K subunit involved in development and growth in the body, it often exhibits pathological hyperactivation in malignancies [8]. Genetic hyperactivation of PI3K/AKT signaling has been identified as a common “driver” mechanism in many solid malignancies, such as breast, endometrial, bladder, colorectal, and head and neck squamous cell carcinoma, thanks to the application of high-throughput gene sequencing [9–11].

On the other hand, postzygomatic activating mutations in PIK3CA with asymmetric overgrowth create benign overgrowth disorders like CILF, which represent a mosaic genetic activation of p110α. Unknown is the cellular substrate that underlies the abnormal tissue accumulation and the CILF’s infiltrative character. However, it was shown in an in vivo investigation on mice that, in the presence of systemic or local pro-adipogenic signals, a population of neural crest-derived fibro/adipogenic progenitor cells differentiates into mature adipose tissue and infiltrates craniofacial tissue in response to tissue injury [12].

External environmental conditions, including irradiation, trauma, and degenerative processes with fatty transformation, could trigger and accelerate the lipomatous change [6].

This is a rare entity and is classified as a subgroup of lipoma. Benign lipomas have 5 subgroups:

Simple encapsulated lipoma, lipoma variants like angiomyolipoma, hamartomatous lesions, infiltrating or diffuse lipomatosis, and benign tumors of brown fat [1].

In addition, there may be macrodontism, early eruption of permanent and deciduous teeth, hypertrophied bones, macroglossia, and parotid gland proliferation on the afflicted side [6].

The study of soft-tissue tumors using imaging has dramatically changed since the introduction of CT and MR imaging, which are highly helpful in making a definitive differential diagnosis [13].

Plain radiographs show hypertrophy of the facial bones and soft tissue swelling. USG may show adipose tissue but cannot delineate the real extent of the lesion [14].

The best modalities are CT scan and MRI since they can determine the extent of the lesions and reveal their fat content. Lesions with a high density can be seen in great detail on a CT scan. The intervening fibrous elements can give a feathery pattern or an inhomogeneous character. A typical CT of CDIL-F

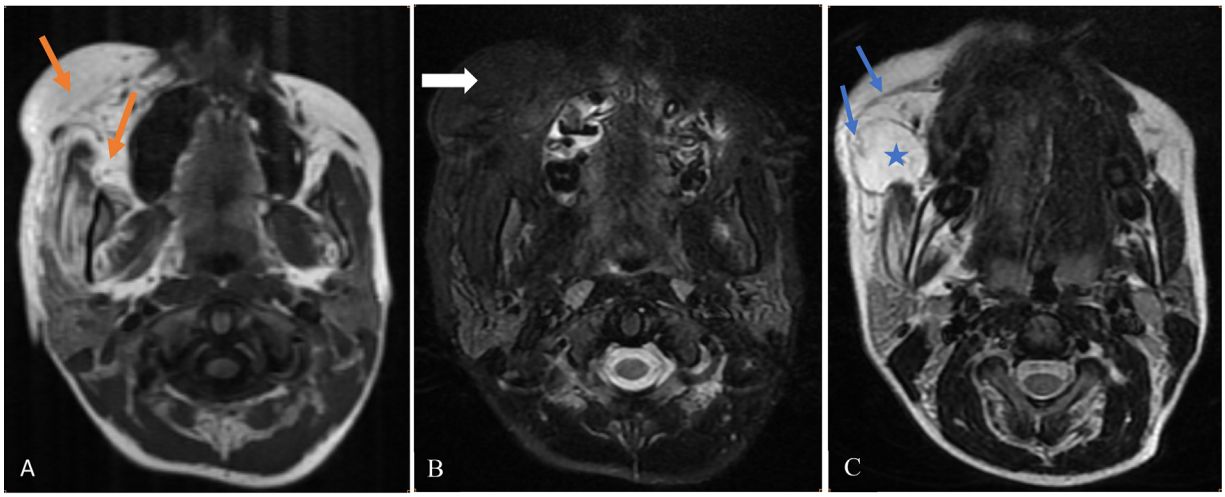

Fig. 2 – MRI images showed an infiltrative lesion in the subcutaneous, muscular and intermuscular planes of the face on the right side (orange arrow), with a moderate decrease in signal intensity on T2-weighted images (blue star), irregular linear regions of hypo intensity were also present (blue arrow). The lesion was hypointense on fat-suppressed images (white arrow).

Fig. 3 – Postoperative image showing a reduction in fat mass and a slight improvement in external appearance.

will show a diffuse, nonuniform fatty infiltration with strong evidence of nonuniform, unilateral hyperplasia of craniofacial bones. The observed nonhomogenous character in fatty infiltration is likely a result of the fibrous components that intervene in the intricate soft tissues of the face [15].

MRI is superior to CT because of its multiplanar capacity. It is able to show the exact extent of the lesion. On T1W images, the lesions show an uneven hyperintense appearance. An MRI provides a clearer picture of bone and muscle involvement. A biopsy may not be required if the typical findings are seen on an MRI [1,13].

Furthermore, an MRI study can rule out additional likely complications involving intracranial/cerebral abnormalities (ipsilateral hemimegalencephaly, asymmetric ventricular-sylvian fissure dilatation, arachnoid cyst, cerebellopontine angle lipoma), and encasement of the facial nerve [15].

The most probable disorders in the differential diagnosis are tumors (hemangiomas) and/or vascular malformations. The most effective diagnostic technique is magnetic resonance imaging. On both T1 and T2, weighted spin-echo sequences, infiltrating lipomatosis is shown as a bright signal with fatty expansion into the surrounding soft tissues [16].

The differential diagnoses included encephalocraniocutaneous lipomatosis and Proteus syndrome. The symptoms of encephalocraniocutaneous lipomatosis include epibulbar dermoids, localized alopecia, brain abnormalities, mental deficit, and lipomas of the scalp and central nervous system [16].

Vascular abnormalities, lipomas, hyperpigmentation, and various nevi are the hallmarks of Proteus syndrome. Proteus syndrome cutaneous lesions typically develop gradually, and the condition may not be diagnosed until late infancy, childhood, or even adulthood. In Proteus syndromes, intrathoracic, pelvic, and intraabdominal lipomatosis are the most common forms. Lipomatosis of the craniofacial region has not, as far as we are aware, been described to Proteus syndrome [16].

According to the clinicopathological manifestations in our case, both of these 2 syndromes were eliminated. Moreover, postoperative pathological identification quickly rendered other conditions like hemangiomas and lymphangiomas obsolete [6].

Liposarcoma is regarded as a differential diagnosis as well; infiltrating lipomatosis is distinct from liposarcomas in that it lacks pleomorphism and lipoblastic activity. Rarely can liposarcomas arise in infantile or congenital forms; they are typically found on the trunk and extremities [17].

The compromise between early resection, which prevents widespread infiltration, and the number of surgical surgeries required versus the increased risk of facial nerve injury in younger individuals complicates treatment [15].

The treatment modalities available were liposuction and excision [6].

An additional technique used to attain facial symmetry is liposuction.

Wide local excision is the main strategy recommended by several writers for the treatment of this ailment [15].

For cosmetic reasons, multiple procedures are required [6].

According to reports, as the number of surgeries increased, the capillary stain's intensity increased as well. This led to the hypothesis that angiogenesis might be involved in the recurrence of CDIL-F [15,18].

Doctors had to attempt to maintain the facial nerve when performing debulking procedures, which were often done through a rhytidectomy incision. If required, osseous reduction procedures of the mandibule, maxilla, and zygoma have occasionally been carried out [6].

A novel treatment approach including targeted chemotherapy and surgical resection was described by Tracy et al. in 2013. After examining the surgical material, medical professionals discovered that the platelet-derived growth factor receptor and c-kit (CD117) were expressed. Personalized targeted treatment with imatinib and celecoxib showed better face symmetry without signs of disease progression. Future therapy paradigms may involve a synergistic approach to treating this rare condition, which could involve combining surgical and medicinal treatments [6].

PI3K inhibitors, which are already being used in clinical trials for patients with cancer, may have a therapeutic role in treating patients with CILF because the PIK3CA gene mutation is the promising pathogenic theory [7]. Even if there was no treatment for the illness, PI3K inhibitor may stop CILF from progressing or recurring [6].

However, there weren't many reports of CILF because it's a very unusual disease. As a result, the surgeon's experiences were always followed when performing a correct surgical procedure. Moreover, the adjuvant treatment was still in research [6].

Both the infiltrating lipoma and the lipoma are composed of benign, mature adipose cells. But in contrast to the lipoma, since the infiltrating lipoma extensively affects the deep soft tissues and is not encapsulated, local excision often results in its recurrence [19].

There is evidence that the debulking or interceptive surgeries have a high rate of complications. This is explained by the lesion's infiltrative nature, the intricate structure of the face, and the variations in growth that occur in different facial regions at different ages. These elements influence the regrowth, together with the lesion's inherent characteristics.

The primary issue with CILF therapy is the high incidence of recurrence after surgical excision, even if the tumor is benign, with a recent study reporting a rate as high as 79% of recurrence [14].

The time to recurrence varies from months to years [20]. Incomplete excision, continued proliferation of fat cells, and malignant transformation have all been identified as contributing causes [5,21].

To enhance the patient's psychological health, short-term procedures including liposuction to correct asymmetry, removal of mucosal neuromas, and elevation of the ptotic upper lip may be carried out. Also, there is little risk involved in performing these operations, and they can be repeated as needed. Periodic debulking has been carried out in accordance with this procedure to enhance the patient's expectations and appearance [14,22].

Conclusion

Congenital infiltrating lipomatosis of the face is a rare benign condition occurring in childhood that leads to craniofacial deformity. Combination of clinical examination and imaging (CT scan and MRI) can establish the diagnosis. Surgery is done for cosmetic purpose.

The recurrence rate is very high after surgical excision, so adjuvant therapy and psychological intervention are important for patients [12].

Patient consent

Informed written consent was obtained from patient for publication of the case report and all imaging studies.

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