Original Paper

Congenital infiltrating lipomatosis of the face

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Abstract Congenital lipomatosis of the face is a very rare syndrome defined as a collection of non-encapsulated mature lipocytes infiltrating local tissues: resulting in cranio-facial deformities. Due to the normal psychomotor development of the children, aesthetic appearance often remains the main concern. After a precise description of a case, a complete overview of the literature data was made. New clinicopathologic data concerning bone involvement will be presented and analyzed in the light of embryonic growth.

Keywords Congenital - Lipomatosis - Vibroliposuction

Introduction

Congenital lipomatosis of the face is a very rare syndrome, first described by Slavin et al. [14] followed by other reports, the total number of cases being 28. As with many other birth defects, the etiopathogeny is poorly understood. However, due to the normal psychomotor development of the children, aesthetic appearance often remains the main concern.

After a precise description of a case, a complete overview of the literature is presented. New clinicopathologic data concerning bone involvement will be discussed and analyzed in the light of embryonic growth.

Case report

A 43-month-old girl was referred to the plastic surgery department with a right diffuse hemi facial deformity. This had been noticed at birth, but was still increasing in size. Clinical
examination demonstrated an infiltration of the subcutaneous tissue extending from the cheek and the mandible to the right fronto-temporal region (Fig. 1). The lesion was soft and painless. We noticed a hyperaemia on the right cheek but no sign of facial nerve compression or bony overgrowth. Although dental occlusion was normal three masses were located on the right side of the lower lip (Fig. 1).

![Clinical pictures. Left preop view. Right postop view](image)

The CT imaging and MRI did not show central nervous deformity nor ocular, bony, arterial or venous anomaly. A non-encapsulated fatty tissue was infiltrating the cheek, the masseteric and pterygoid region. Fatty tissue was also noticed in the right fronto-temporal subcutaneous layer and diploe (Fig. 2).
Fig. 2 MRI of the fronto-temporal region. Left preop view, Right postop view

The parents were adamantly in wishing for a surgical approach because of the child’s appearance. We chose the vibroliposuction technique used for subcutaneous fat resection. A total amount of 90 cc was aspirated after tumescent infiltration. Pathologic examination of the excised fat did not reveal any particular appearance. Although painless, the lip tumors resected were neuromas.

In the short term, the child’s appearance was improved. Fourteen months later, the frontal mass recurred slightly as well as the mucosal neuroma leading to another vibrolipoaspiration session and lip resection. An MRI of the face was taken 12 months after surgery, demonstrating improvements as shown in the illustrations (Figs. 1, 2).

Discussion

Slavin et al. [14] first reported three cases of what they termed as congenital infiltrating lipomatosis of the face (CIL-F). It was therefore defined as a distinct clinicopathological entity: a collection of non-encapsulated mature lipocytes infiltrating local tissues. The pathogenesis of this condition is not well known. Two cases of CIL-F associated with congenital Cytomegalovirus infection have been reported but no strong relationships have been demonstrated [6, 13]. Somatic mutation at a mosaic state could give rise to an adipose stem cell producing infiltrated lipomatosis. These mutated cells could result in altered production of tissue growth factor or modify their receptor response to these factors, resulting in mucosal neuroma, bony overgrowth, and precocious tooth development [7].

The lesions are composed of increased quantity of normal fat infiltrating adjacent tissue and there is no report of malignant fat tumors over a 2–14-year follow-up [14]. The absence of
lipoblastic proliferation, pleomorphism, and mitosis distinguishes congenital infiltrating lipomatosis of the face from liposarcoma. Morphologic criteria have been described [14] for CIL-F combining an adipose infiltration of adjacent muscle and soft tissue, without malignant characteristics, an absence of lipoblasts, fibrous elements, an increased numbers of vessels with unifocally thickened muscular walls and increased numbers of nerve bundles characterized by variable size and focal fibrosis.

The facial asymmetry is noticed at birth, located in the face (cheek being involved in 12 cases out of 13) and remains unchanged with growth [12]. In their review of 13 cases, Padwa and Mulliken [12] extended the description of the congenital infiltrating lipomatosis of the face including clinical presentation and radiological findings, thereby helping to establish the diagnosis. Clinical findings include a vascular blush, an increased density of facial hair on the affected side, an ipsilateral hemimacroglossia, a ptosis of the upper lip, an enlarged lower lip, an early eruption of deciduous and permanent teeth, a poor formation of roots in one or more permanent teeth, some missing permanent teeth, larger permanent teeth, dentigerous cyst around permanent teeth, ipsilateral hypertrophy of the underlying facial skeleton with asymmetry of the mandible (this observation has not been reported for the youngest patients but bony asymmetry seems to increase with growth in older patients). Mucosal neuromas were found inside the labial commissure or the tongue of six patients; four were positive for RET mutation predisposing for MEN-2B.

Although there was no bony, dental or lingual deformity in our case, it seems convenient to remember the embryonic origin of the craniofacial structures [9]. Craniofacial membranous bones derive from ectomesenchymatous cells migrated from the neural crest. Facial muscles are derived from mesoderm, located in paraxial cranial somitomeres and occipital somites through the branchial arches. Odontogenesis starts with the dental germ, which is an association of the epidermal crest cells and mesenchyme derived from the neural crest. The anterior two-thirds of the mucosal part of the tongue derive from the first branchial arch and the posterior third from the third and fourth arches. The melanocytes migrate initially from the neural crest. Myoblasts of each of the first three branchial arches derive from an adjacent somitomeres. The appearance of these myoblasts is not determined when in the somitomeres, the migration and differentiation of these cells is controlled by migrated cells of the neural crest. Experimental findings on chickens have shown that the appearance of the neural crest cells is determined before they start to migrate into the branchial arches; if the supposed first arch neural crest cells of a chicken are transplanted into the supposed second arch neural crest of another chicken, they migrate in the second branchial arch but differentiate into ectopic structures of the first arch. All these observations could suggest that the origin of all the deformities observed in CIL-F (muscular and calvarian fatty infiltration, mucosal neuroma, etc.) would be neural crest cell anomalies.

Ha et al. [8] first reported MR imaging of CIL-F. The MRI provides excellent delimitation of the extent of the lesion and its margin including marked bony deformation. Pure fatty tumors in childhood (high signal intensity similar to subcutaneous fat in both T1 and T2 weighted images) are almost always benign and a morphologic characterization by MRI may be sufficient to make the critical therapeutic judgment [8]. Therefore, young patients do not need to undergo biopsy. Thus, the diagnosis can be made on the basis of clinical examination and magnetic resonance imaging [12]. Although ultrasound examination can be helpful in assessing the growth of adipose tissue, it remains inferior to CT and MRI in calculating the extent and the exact size of the masses relative to other functionally important structures [4].

In their review, Padwa and Mulliken [12] discuss the differential diagnosis of infiltrating lipomatosis of the face including Proteus syndrome, encephalocraniocutaneous lipomatosis,
hemihyperplasia-multiple lipomatosis syndrome, and Bannayan–Riley–Rulvaca syndrome. The most likely conditions in the differential diagnosis are vascular malformation and/or tumor (hemangioma). Proteus syndrome is characterized by variable overgrowth of tissues from all three germ layers, presenting in a wide spectrum of findings: hemihyperptrophy, macrodactyly, exostoses, osseous hypertrophy, linear sebaceous nevi, deep hyperkeratotic rugae on the soles of the feet, and multiple "hamartomatous" tumors. Although some of these tumors are called lipomas, they are not well encapsulated and are composed of infiltrating mature adipocytes. The lipomatosis in Proteus syndromes is most commonly intrathoracic, pelvic, and intraabdominal. Encephalocraniocutaneous lipomatosis is characterized by lipomas of the scalp and central nervous system, brain malformations, mental deficiency, seizures, epibulbar dermoids, and focal alopecia. There is a wide spectrum and variability in severity of manifestations characterized by focal overgrowth of cellular elements in skin, bone, and other connective tissues. Similar to both facial infiltrating lipomatosis and Proteus syndrome, asymmetric enlargement is the principal and constant feature of this disorder. Other entities that may belong in this spectrum include hemihyperplasia-multiple lipomatosis syndrome and Bannayan–Riley–Rulvaca syndrome. Patients with hemihyperplasia-multiple lipomatosis syndrome have moderate soft-tissue asymmetry with infiltrating lipomatosis and cutaneous capillary malformations. These abnormalities are noticed at birth and grow commensurately with the child. Bannayan–Riley–Rulvaca syndrome, an autosomal dominant condition with variable expressivity and male preponderance, is characterized by multiple encapsulated or infiltrating lipomas, multiple "angiomas," and macrocephaly. The angiomas are a minor component of this disorder and are usually small capillary or venous malformations.

Some authors [14, 16] advise against radical surgical treatment, the lesion being benign and localized in a dangerous region (proximity of the facial nerve). Nevertheless surgical treatment can improve the aesthetic appearance of the child despite evidence of tumor persistence. Although these tumors are benign, Slavin et al. [14] recommended an early aggressive surgical approach to control the infiltrative nature of their growth and to improve facial appearance. Delayed surgery with succession of aesthetic procedures gives poor results [2]. Van Wingerden [16] advocates a delayed approach underscoring less risks of damaging the facial nerve, fewer procedures being necessary, more mature controlateral cheek for the comparison at the time of surgery, benign character of the tumor. He was the first to report a case of face liposuction in a child; in his experience he removed about 50 cc of fat tissue and it turned out not to be enough. Early aggressive surgical resection associated with liposuction was not satisfactory [2]. Out of Padwa and Mulliken’s experience [12], the mean number of operations per patient was 2.5, asymmetry returning to the preoperative state within months. Typically there is an important risk of recurrence after surgery estimated at 62.5% by Dionne and Seemayer [5] and confirmed by other authors [2, 7, 12, 14, 15, 16].

However, excised mucosal neuroma did not recur. Therefore they advise delaying extensive resection as long as possible and advocate temporizing measures to improve asymmetry (liposuction, excision of mucosal neuroma and elevation of the upper lip); this type of treatment can be performed in young patients with minimal risk and can be repeated if necessary. We think that vibroliposuction is a valuable technique in this context due to its particular ability to penetrate firm tissue as developed after recurrences or in secondary cases.

References

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