



Radiographic Findings of Congenital Facial Infiltrating Lipomatosis: A Case Report

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ABSTRACT

Congenital diffuse infiltrating lipomatosis of the face (CDIL-F) is a rare clinicopathological entity with an unknown etiology, in which mature adipose tissue infiltrates the soft tissue structures in one side, and causes considerable asymmetry. Herein, we report a case of CDIL-F who underwent many surgical procedures without definite diagnosis during 12 years. CDIL-F presents symptoms with various severity levels due to infiltration of adipose tissue that makes removal difficult. Thus, it is recommended to inform patients about the high rate of recurrence.

Keywords: Congenital Abnormalities; Facial Asymmetry; Lipomatosis; Prognathism

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INTRODUCTION

Congenital diffuse infiltrating lipomatosis of the face (CDIL-F) is a non-neoplastic mature adipose tissue proliferation into the facial soft tissue, which can cause severe esthetic problems [1,2]. It may also compress the vital structures of the face. Pathological infiltration of fat has six different presentations: diffuse lipomatosis, pelvic lipomatosis, symmetric lipomatosis (Madelung's disease), adiposis dolorosa (Dercum's disease), steroid lipomatosis, and nevus lipomatosis [1,3]. Diffuse lipomatosis has been described as a rare mature adipose tissue infiltration in the trunk or extremities [1,3,4]. It is not common in facial areas [1,3,4]. CDIL-F was first described by Slaviv et al, [5] in 1983. Facial asymmetry, parotid involvement, osseous

hypertrophy, macroglossia, macrodontia, early eruption of dentition, mucosal neuroma, cutaneous capillary blush, and facial hair overgrowth have all been reported in the affected side in different cases [5-11]. CDIL-F has been scarcely reported in Iran. It should be notably pointed out that this reviewed case took 12 years to accurately diagnose; hence, in the current study, the discussion about the diagnosis and treatment of the lesion has focused on early detection and better management of such cases.

CASE REPORT

A 27-year-old female was referred to the Oral and Maxillofacial Radiology Department of Faculty of Dentistry, Tehran University of

Medical Sciences for cone-beam computed tomography (CBCT) due to facial asymmetry with 12 years of duration. She reported several biopsies and cosmetic surgical procedures started at age 17 for the first time. The details of the surgical procedures were not unavailable but several surgical scars were seen on her right side of the face (Fig. 1).



Fig. 1: Clinical examination: scarring, hypertrophy, asymmetry, enlargement of the maxillary alveolar bone, the buccal fat pad, and mandible are seen in the right side

She was also under orthodontic treatment for malocclusion. On clinical examination, hypertrophy of the soft tissue of the cheek and underlying bone, macrodontia, macroglossia and malocclusion were all seen in the right side of the face.

The patient declared early eruption of permanent teeth, and facial hair overgrowth in the affected side. CBCT images showed diffuse bone enlargement in the right maxillary and mandibular alveoli, as well as the right zygomatic bone. Changes in trabecular patterns were seen in both jaws. Root resorption was seen in teeth #13, 14, 15, 16 and 46. Periodontal ligament space widening was obvious in teeth #44 and 45 (Fig. 2). The latter two findings might be features of the disease or a consequence of uncontrolled orthodontic forces. Magnetic resonance images revealed fat infiltration into the right masseteric space up to the skull. Gadolinium was poorly absorbed by the lesion (Fig. 3). Computed tomography (CT) scan images showed massive deformity in facial bones at the right side (Fig. 3). No familial disease or remarkable medical history was reported. First incisional biopsy of the lip was performed at the age of 17. Microscopic evaluation showed numerous haphazardly-distributed nerve bundles from subepithelial stroma deep into minor salivary glands and between striated muscle fibers accompanied by many irregular thick- and thin-wall blood vessels. Disorganized adipose and fibrous tissues were also reported. The pathologist put hamartomatous lesion as the diagnostic impression and recommended further investigations for multiple endocrine neoplasia (MEN type II syndrome).

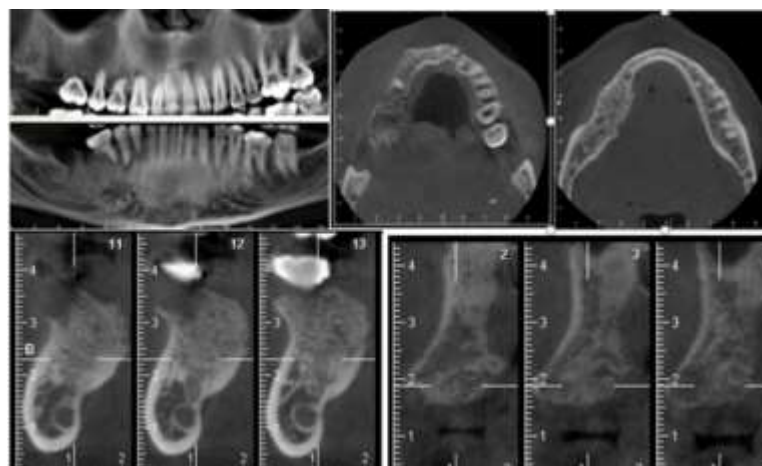


Fig. 2: Bone enlargement and changes in trabecular pattern of both jaws in the right side can be seen on CBCT images. On the panoramic view, root resorption can be seen in teeth #13, 14, 15, 16 and 46

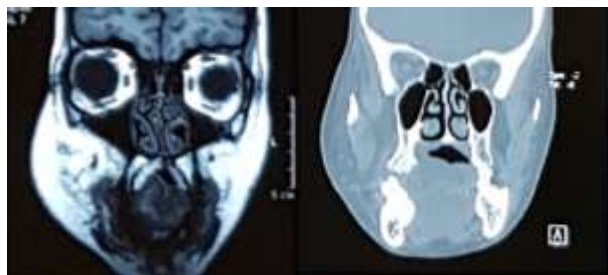


Fig. 3: T1 MRI shows high signal fat in the right buccal area, and CT shows fat density in the right cheek area

Two subsequent biopsy samples were taken when she was 18 and 19 years, and also at the age of 25 after cosmetic surgical procedures, and intramuscular lipoma and mucosal neuroma were respectively reported by the pathologists.

Considering the clinical and microscopic features, and also MRI, CT and CBCT images, the final diagnosis of CDIL-F was made, which had remained misdiagnosed for 12 years. Written informed consent was obtained from the patient to publish this report.

DISCUSSION

Slavin et al. [5] described CDIL-F in 1983. The corresponding histomorphological findings include: (I) unencapsulated diffuse infiltration of mature adipose tissue into the adjacent muscles and soft tissue, (II) presence of fibrous component and numerous nerve bundles in different sizes, and vessels with thickened wall, (III) rapid growth which causes hypertrophy of the underlying bone without any malignancies, (IV) high tendency for postsurgical recurrence, and (V) presence of fibrous elements and absence of lipoblasts [1,2,4,5,11-13].

According to the pathological findings of CDIL-F, radiography plays a contributing role in identification of this lesion [9,10]. MR Images demonstrated fatty nature of the disease and osseous abnormality, after infiltration was observed on CBCT images. As mentioned earlier, despite previous pathological reports, CT, CBCT and MR images led to accurate diagnosis of CDIL-F in our patient after 12 years.

Singh et al. [4] reviewed 35 cases in the

literature and reported phenotypic features of CDIL-F to be unilateral hypertrophy of the facial soft tissue, unilateral skeletal hypertrophy, capillary blush in the skin, macrodontia in the affected side, abnormal roots, early eruption of permanent as well as deciduous teeth in the affected side, ipsilateral macroglossia, and protuberances on the tongue and buccal mucosa as a presentation of mucosal neuromas [4].

The pathogenesis of CDIL-F is unclear. Some authors believe that CDIL-F is the consequence of somatic mutations in PTEN and RET genes [4,11]. On the other hand, cytomegalovirus inclusions were reported in secretory cells of the parotid glands in some cases with parotid involvement. However, a causal relationship between cytomegalovirus and lipomatosis has not yet been found [4,14]. A study reported deletion of chromosome 1q24.3q31.1 in a girl with CDIL-F [15]. Recent literature proved a somatic mutation in the PIK3CA gene in CDIL-F which is also observed in cancers and overgrowth disorders. PIK3 plays a potential role in proliferation, survival and adhesion of cells [16]. Irradiation, trauma, muscular metaplasia, and fatty transformations as environmental conditions can enhance the occurrence of lipomatosis. Hormonal effect on multipotent cells of embryogenic origin and alterations in chromosome 12 can change the lipomatosis behavior [5,9,13,17]. Differential diagnosis for unilateral facial enlargements includes congenital hemifacial hyperplasia, vascular malformations, benign and malignant tumors of the hard and soft tissues, hamartomas, congenital lymphedema, and segmental odontomaxillary dysplasia [9,11]. Vascular malformation and lymphangioma pathologically differ from facial lipomatosis [9,11]. Encephalocraniocutaneous lipomatosis and Proteus syndrome could also be considered as differential diagnosis. Embryonic mutations from somatic mosaicism could lead to Proteus syndrome. According to the time of mutation and diffusion of mature cells, variable phenotypes such as lipoma, hyperpigmentation, and vascular disorders may be seen.

The Proteus syndrome may be diagnosed from infancy up to adulthood [9]. Encephalocraniocutaneous lipomatosis includes lipoma of the scalp and central nervous system. Encephalocraniocutaneous lipoma and the Proteus syndrome were ruled out according to the clinicopathological features [9,16]. Segmental odontomaxillary dysplasia is an uncommon, nonhereditary developmental malformation characterized by unilateral enlargement of posterior segment of the maxilla, and enlargement of gingiva, and dentition of the same arch. Ill-defined bone sclerosis with thickened and coarse vertically oriented trabeculae of woven bone with a granular appearance are usually observed on radiographs. Congenital hemifacial hyperplasia is a rare developmental anomaly exhibited by unilateral enlargement of hard and soft tissues of the face. Predominant enlargement of cranial bones, zygoma, maxilla, and mandible with midline shift and deviated occlusal plane are reported. Variations in size, shape, and number of teeth are also seen. Radiographically, this lesion presents with hard tissue and soft tissue enlargement at the affected side [9,11,16]. In T1 MR Imaging of CIL-F, fat infiltration is reported in the buccal region of affected side which is different from other conditions.

According to Singh et al, [4] the progression of the condition seems to follow two patterns: A rapidly progressive form with early onset during the first year of birth and a more indolent form which progresses over decades. Therefore, management depends on the rapidity of the disorder development. In early reports, Slavin et al, [5] and Kang et al. [7] recommended wide and early local excision while Van Wingerden et al. [18] suggested delayed resection to prevent facial nerve damage and unnecessary surgical procedures. More recently, Kamal et al. [17] mentioned that delay in surgical procedures can give the chance of observing contralateral mature cheek to match during surgery.

Some articles also suggest liposuction and surgical excision as a treatment [9,19]. Several surgical procedures are needed to achieve cosmetic goals. In some cases, reduction of

maxilla, mandible, and zygomatic bone was performed [9,19].

Tracy et al. [20] proposed a combination of surgical resection and specific targeted chemotherapy. Imatinib and celecoxib were used as personalized targeted chemotherapy in order to manage CDIL. Assessment of this treatment protocol demonstrated synergistic effects to improve facial symmetry and control the progression of disease. Although CDIL-F is a rare disorder of lipomatous tissue, surveys declared no malignant features over 2-14 years of follow-up [20].

Recurrence after surgery is an important feature of CDIL-F. According to Balaji [1], about 48% of cases showed recurrence after surgery. Padwa and Mulliken [11] believed growth hormones to have a role in recurrence and stated that any mass reduction before the end of adolescence may fail. But in some patients with a rapidly-progressive CDIL-F, dealing with severe facial deformities until that time might be psychosocially difficult. Despite recurrence and progression, surgical procedures in early childhood improve esthetics [7].

The pathogenesis and etiology of CDIL-F are not fully understood. According to the literature, the disease is a congenital condition with various severity levels. Patients with CDIL-F often undergo many surgical procedures to remove excessive fatty tissue and achieve esthetic results; but the prognosis is not always satisfactory because of deep infiltration of fatty tissue between other facial tissues. The patients should be informed about the high rate of recurrence and inadequate results.

CONFLICT OF INTEREST STATEMENT

None declared.

REFERENCES

1. Balaji S. Congenital diffuse infiltrating facial lipomatosis. *Ann Maxillofac Surg*. 2012 Jul;2(2):190-6.
2. Sahai S, Rajan S, Singh N, Arora H. Congenital infiltrating lipomatosis of the face with exophytic temporomandibular joint ankylosis: a case report and review of the literature.

- Dentomaxillofac Radiol.2013;42(3):16128745.
3. Weiss SW, Goldblum JR, editors. 4th ed. London;Enzinger and Weiss's Soft Tissue Tumors; 2002, 574.
 4. Singh K, Sen P, Musgrove BT, Thakker N. Facial infiltrating lipomatosis: a case report and review of literature. *Int J Surg Case Rep.* 2011;2(7):201-5.
 5. Slavin SA, Baker DC, McCarthy JG, Mufarrij A. Congenital infiltrating lipomatosis of the face: clinicopathologic evaluation and treatment.*Plast Reconstr. surg.* 1983 Aug;72(2):158-64.
 6. Bouletreau P, Breton P, Freidel M. Congenital infiltrating lipomatosis of the face: case report. *J Oral Maxillofac Surg.* 2000 Jul;58(7):807-10.
 7. Kang N, Ross D, Harrison D. Unilateral hypertrophy of the face associated with infiltrating lipomatosis. *J Oral Maxillofac Surg.* 1998 Jul;56(7):885-7.
 8. Kim J-E, Gottschall JA, Bachman RP, Nemzer L, Puligandla B, Schauer G. Facial infiltrating lipomatosis: physical, radiological, and histopathological findings. *Arch Otolaryngol Head Neck Surg.* 2010 Mar;136(3):301-3.
 9. Li Y, Chang G, Si L, Zhang H, Chang X, Chen Z, et al. Congenital Infiltrating Lipomatosis of the Face: Case Report and Literature Review. *Ann Plast Surg.* 2018 Jan;80(1):83.
 10. Nair N, Ramachandran A. Congenital infiltrating lipomatosis of the face. *Medical Journal of DY Patil Vidyapeeth.* 2018 Sep;11(5):439.
 11. Padwa BL, Mulliken JB. Facial infiltrating lipomatosis. *Plast Reconstr Surg.* 2001 Nov;108(6):1544-54.
 12. D'souza D, Babu GS, Shetty SR, Rasquinha V. Congenital infiltrating lipomatosis of the face: A case report with review of literature. *Indian Dermatol Online J.* 2014 Jul;5(3):303-5.
 13. Mahadevappa A, Raghavan VH, Ravishankar S, Manjunath GV. Congenital infiltrating lipomatosis of the face: a case report. *Case Rep Pediatr.* 2012(2); 134646.
 14. Heymans O, Ronsmans C. Congenital infiltrating lipomatosis of the face. *Eur J Plast Surg.* 2005;28(3):186-9.
 15. Capra V, Severino M, Rossi A, Nozza P, Doneda C, Perri K, et al. Pituitary deficiency and congenital infiltrating lipomatosis of the face in a girl with deletion of chromosome 1q24. 3q31. 1. *Am J Med Genet A.* 2014 Feb;164(2):495-9.
 16. Maclellan RA, Luks VL, Vivero MP, Mulliken JB, Zurakowski D, Padwa BL, et al. PIK3CA activating mutations in facial infiltrating lipomatosis. *Plast Reconstr Surg.* 2014 Jan;133(1):12e-9e.
 17. Kamal D, Breton P, Bouletreau P. Congenital infiltrating lipomatosis of the face: report of three cases and review of the literature. *J. Cranio-Maxillofac. Surg.* 2010 Dec;38(8):610-4.
 18. Van Wingerden JJ, Erlank J, Becker JH. Liposuction for congenital infiltrating lipomatosis of the face. *Plast Reconstr Surg.* 1988 Jun;81(6):989.
 19. Urs AB, Jeyaseelan Augustine PK, Arora S, Aggarwal N, Sultana N. Infiltrating lipomatosis of the face: A case series. *J Nat Sc Biol Med.* 2013 Jan;4(1):252-7.
 20. Tracy JC, Klement GL, Scott AR. Interdisciplinary management of congenital infiltrating lipomatosis. *Int J Pediatr Otorhinolaryngol.* 2013 Dec;77(12):2071-4.