

CONGENITAL DIFFUSE INFILTRATING LIPOMATOSIS OF FACE—A RARE ENTITY

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ABSTRACT

Congenital diffuse infiltrating lipomatosis of the face (CDIL-F) is a rare and definite clinical entity of unknown etiology. It comprises a subgroup of lipomatous tumors. It is congenital in origin and occurs in infancy or in early childhood. CDIL-F is characterized by diffuse infiltration of mature adipose tissues over normal muscle fibers, rapid growth associated with osseous hyperplasia with a high recurrence rate after surgical intervention. Complete surgical excision is difficult due to diffuse infiltration and involvement of important facial structures. Till now, 14 cases have been reported in the literature. We report a case of CDIL-F in a 4-year old male child presenting with diffuse painless swelling on right side of face. Ultrasonography, computed tomography and magnetic resonance imaging showed diffuse fat deposition in soft tissues in right malar region with thickening of right masseter muscle, fatty infiltration in right half of tongue and osseous changes in right maxilla and right zygomatic arch.

Keywords: Infiltrating lipomatosis, lipomatous tumor, congenital, face.

Introduction

Infiltrating lipomatosis of the face is a rare distinct clinico-pathological entity characterized by non-capsulated diffuse infiltration of mature adipose tissue (adipocytes) over normal muscle fibers, rapid growth rate and hyperplasia of underlying bones with resultant craniofacial deformities with a tendency to recur after surgery.¹ It causes diffuse overgrowth of subcutaneous fat, muscle and bone.² It was described by Slavin et al. in 1983.³

Till now, 14 cases of CDIL-F with bony involvement have been reported in the literature.⁴ It is a subgroup of lipomatous tumor like lesions characterized by a collection of non-capsulated, mature adipocytes which infiltrate local tissues. Lipomas are common benign tumors of mesenchymal origin. According to histologic features and growth patterns, benign lipomatous tumors are subclassified into classic lipomas, angiolipoma, fibrolipoma, intramuscular lipoma, pleomorphic

lipoma, infiltrating lipoma, hibernoma, lipoblastomatosis and diffuse lipoblastomatosis. Infiltrating lipomatosis of the face is extremely rare in head and neck region. It is an uncommon mesenchymal neoplasm in which mature adipocytes invade the adjacent tissues.³

Case Report

A 4-year-old male patient presented with diffuse painless swelling in the right malar region, overlying the right maxillary sinus since birth (Fig. 1). It was gradually increasing in size. Overlying skin was normal with no bluish or pinkish discoloration or nevus. Local ultrasonography (USG) was done with a linear probe (7-12 MHz) showed diffuse hypoechoic fat deposition subcutaneously in right malar region overlying right maxillary sinus (Fig. 2). No calcification or abnormal

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vascularity was noted. A diagnosis of diffuse lipomatosis was given.



Figure 1: Photograph of a 4-year-old male child diagnosed to have congenital diffuse infiltrating lipomatosis of face showing diffuse swelling on right side of face (White arrow).

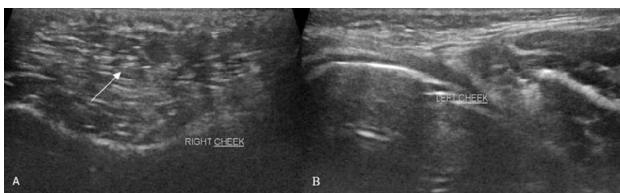


Figure 2: Transverse ultrasonography of right malar (A) and left malar (B) region with linear 7-12 MHz probe showing diffuse hypoechoic fat deposition subcutaneously in right malar region overlying right maxillary sinus (White arrow).

Computed tomography (CT) of maxillo-facial region (Fig. 3, 4) revealed diffuse fat density lesion (CT value -80 to -120 HU) in soft tissues in the right malar region anterior to the anterior wall of right maxillary sinus extending lateral to right zygomatic arch and along the anterolateral aspect of right maxilla and mandible. No calcification was noted. Right masseter muscle was thickened. Right half of tongue showed mild fatty infiltration. Right maxillary sinus was larger

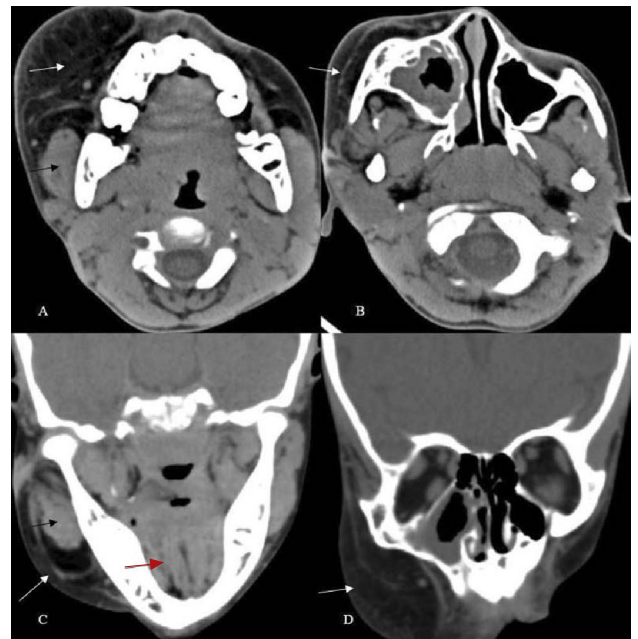


Figure 3: Plain axial (A,B) and coronal (C,D) sections showing diffuse fat density lesion in soft tissues in the right malar region anterior to the anterior wall of right maxillary sinus extending lateral to right zygomatic arch and along anterolateral aspect of right maxilla and mandible (White arrow). Right masseter muscle was thickened (Black arrow). Right half of tongue showed mild fatty infiltration (Red arrow).

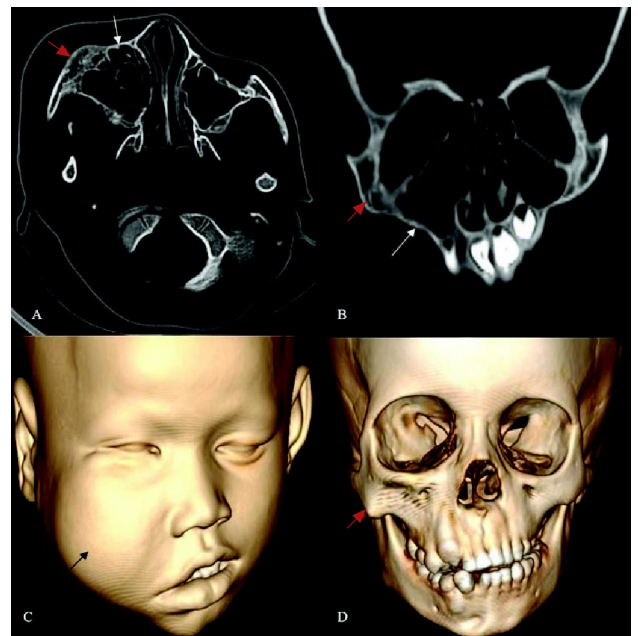


Figure 4: Axial (A) and coronal (B) bone windows showing relatively larger right maxillary sinus (White arrow) with thickening of anterior portion of the right zygomatic arch (Red arrow). Surface shaded volume rendered image (C) showing diffuse swelling in right malar region (Black arrow). Shaded surface volume rendering of bone tissue (D) showing thickened right zygomatic arch (Red arrow).

in size showing marked mucosal thickening due to sinusitis. The anterior portion of the right zygomatic arch was thickened.

Magnetic resonance imaging (MRI) showed diffuse fat intensity lesion in the subcutaneous plane in the right malar region anterior to the anterior wall of right maxillary sinus, appearing hyperintense on T1WI, intermediate signal intensity on T2WI and nullified on STIR sequence. A diagnosis of diffuse lipomatosis in the maxillo-facial region was made. Mild fatty infiltration was noted in right half of tongue (Fig. 5,6).

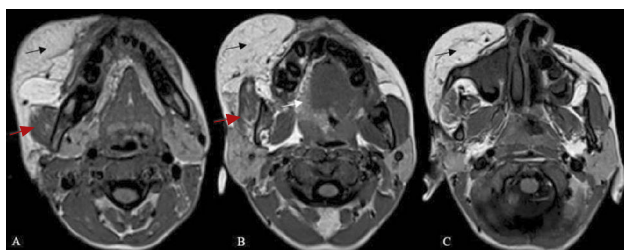


Figure 5: Axial T1 sequence showing diffuse fat deposition (diffuse hyperintense signal) subcutaneously in right malar region extending lateral to right zygomatic arch (Black arrow). Right masseter muscle shows mild thickening (Red arrow), with mild fatty infiltration in right half of tongue (White arrow).

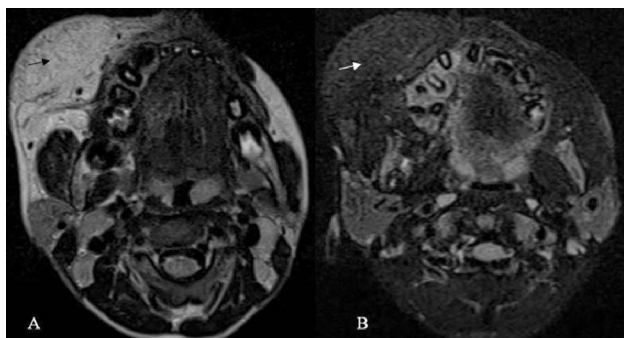


Figure 6: Axial T2WI (A) showing diffuse fat deposition (intermediate signal intensity) subcutaneously in right malar region extending lateral to right zygomatic arch (Black arrow). Axial STIR image (B) showing suppression of fat (White arrow).

Discussion

Males and females are usually affected equally by CDIL-F with no predilection between the right and left side of the face. It involves subcutaneous tissues, muscles and bone. It can involve cheek, mental area, buccal sulcus, floor of the mouth, lip, tongue and parotid gland.⁵

Though the cause is unknown, the proposed mechanisms for lipomatous change are trauma, chronic irradiation, congenital cytomegalovirus infection, alteration

in chromosome 12, degenerative processes with fatty transformation, multipotent cells of embryonic origin under the influence of hormones and muscular metaplasia.^{3,5}

Clinically, CDIL-F presents as diffuse painless large swelling on the side of the face. It is soft, non-tender and fluctuating. Unilateral hypertrophy of soft tissues of the face is seen especially involving the cheek with diffuse infiltration and skeletal overgrowth. There can be macrodontia and abnormal root formation on the affected side, an early eruption of deciduous and permanent teeth, protuberance on the tongue and buccal mucosa (due to underlying mucosal neuromas) and macroglossia.

USG shows diffuse fat deposition in the subcutaneous plane with no definite margins. It cannot delineate the extent of the lesion and involvement of underlying muscles and bones. CT and MRI can both detect lipomatosis, thickening of involved muscles and bony changes. On CT, diffuse lipomatosis is seen as homogeneous hypoattenuated masses showing CT value of -60 to -120 HU with no enhancement on contrast study associated with thin soft tissue density septa. Thinning of masseter and buccinators muscles may be seen with fatty infiltration. MRI due to superior soft tissue contrast resolution is useful in detecting the extent of the mass. Lipomatosis appears hyperintense on T1WI, intermediate signal intensity on T2WI and nullified on FAT SAT sequences.

Bone changes seen in CDIL-F are zygomatic hyperplasia, hemimandibular hyperplasia, accelerated dentoskeletal growth, hyperplasia and sclerosis of the skull and cervical vertebrae, remodeling of temporomandibular joint (showing hyperplasia of condylar process with eventual ankylosis). These bony changes occur due to regional mesenchymal malformation, periosteal irritation from overlying mass and increased vascularity.

Slavin described characteristic histomorphologic findings in CDIL-F - infiltration of fat into adjacent soft tissues and hypertrophy of the underlying skeleton, absence of lipoblasts, presence of fibrous elements, increased number of vessels with unifocally thickened muscular walls, increased number of nerve bundles of variable size with focal fibrosis and absence of malignant characteristics.³

Rosa et al. described following characteristics in CDIL-F: non-capsulated proliferation of mature adi-

pose tissues, diffuse infiltration of muscle and adjacent soft tissue, presence of fibrous tissue with nerve bundles and thickened wall of vessels, hypertrophy of subjacent bones, absence of lipoblasts and signs of malignancy in spite of rapid growth rate and high tendency to recur after surgical excision and congenital in origin. Lipomatous degeneration can occur, however, malignant transformation is not seen.⁶

Treatment is usually done for aesthetic reasons. Available modalities are surgical excision and liposuction. Due to infiltrating nature, changes of recurrence are very high after surgical excision.⁷

Differential diagnosis includes hemifacial hypertrophy, vascular malformation (hemangioma), Proteus syndrome, encephalo craniocutaneous lipomatosis, hemihyperplasia - multiple lipomatosis syndrome, Banayan-Riley-Rulvacaba syndrome.⁸

Conclusion

Congenital diffuse infiltrating lipomatosis of the face is a rare benign condition seen in childhood with no gender predilection, characterized by diffuse infiltration of fat in subcutaneous and muscle planes with underlying bony hypertrophy. CT and MRI are diagnostic which detect the extent of lipomatosis, changes in underlying muscles and bones. Surgery is usually done for cosmetic purpose; however, recurrence rate is high.

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