CASE REPORT

Congenital infiltrating lipomatosis of the face with exophytic temporomandibular joint ankylosis: a case report and review of the literature

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Congenital infiltrating lipomatosis of the face (CIL-F) is a rare lipomatous lesion with diffuse fatty infiltration of tissues and hyperplasia of underlying bone. We report clinical and CT findings in an unusual case of CIL-F presenting with progressive hemifacial asymmetry, manifesting as severely restricted mouth opening owing to exophytic temporomandibular joint ankylosis. The role of imaging in diagnosis is presented with a review of the literature. Differential diagnosis of CIL-F and its exclusion as a cause of hemifacial hyperplasia are also discussed.

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Introduction

Congenital infiltrating lipomatosis of the face (CIL-F) is a rare lipomatous lesion characterized by diffuse fatty infiltration of facial soft tissues and is usually seen in childhood. It was first described by Slavin et al in 1983.¹ The lipomatous tissue can infiltrate facial muscles, soft tissues and bones to cause ipsilateral hyperplasia of the underlying facial skeleton, asymmetry of the mandible and even dental abnormalities.^{1–3} There is no sex predilection; patients have normal psychomotor development and aesthetics are often the primary concern.^{4,5} This condition may progress rapidly with extensive hyperplasia in childhood (up to and including 1 year of age) or as an indolent form with gradual hyperplasia where patients present for treatment in adulthood.⁶

The aetiopathogenesis of CIL-F is poorly understood.^{1,4,5} Possible aetiologies for the lipomatous change and infiltration include somatic mutation. Multipotential cells

of embryogenic origin under the influence of hormones, trauma, chronic irradiation, muscle metaplasia or congenital cytomegalovirus infection could precipitate the somatic defect.^{7–11} This rare entity is classified as a subgroup of lipoma. Benign lipomas have five subgroups:⁸

1. simple encapsulated lipoma
2. lipoma variants such as angiomyolipoma
3. hamartomatous lesions
4. infiltrating or diffuse lipomatosis
5. benign tumour of brown fat—hibernoma.

Histopathological features of CIL-F include adipose infiltration of adjacent muscle and soft tissue without malignant characteristics, absence of lipoblasts, fibrous elements, increased numbers of vessels with unifocally thickened muscular walls and increased numbers of nerve bundles of variable size with focal fibrosis.¹

Bone changes in CIL-F include sclerosis and hyperplasia of the skull, cervical vertebrae, hemimandibular hyperplasia (including ramus, condylar process), accelerated dentoskeletal growth and zygomatic hyperplasia.²

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Table 1 Bone involvement in published cases of congenital infiltrating lipomatosis of the face

Author	Age at presentation	Sex	Clinical presentation	Bone involvement	Management
Bouletreau et al ²	16 years	F	Left facial asymmetry	Mandibular body, ramus, condylar process and left zygomatic bone	Inferior mandibular border ostectomy, alveolar graft ^a
Rajeswaran et al ⁴	11 years	F	Diffuse swelling right face	Right frontal, orbital, zygomatic and maxillary bones	Cosmetic (debulking) surgery
Heymans et al ⁵	43 months	F	Soft, painless swelling right cheek	None	Vibroliposuction
Kang et al ⁶	25 years	M	Right facial swelling	Maxilla	Soft tissue debulking, total parotidectomy and partial buccal fat, masseter resection ^b
Haloi et al ⁸	1 year	—		None	NA
Chen et al ⁹	10 months—4 16 months	F	Elastic non-tender left cheek mass	Ipsilateral regional macrodontia ^c Left zygoma and maxilla	Debulking surgery with preservation of facial nerve
Salvatore et al ¹⁵	37 years	F	Swelling right cheek, parotidomassetric and submandibular regions	No signs of bone erosion	Complete excision
Malik et al ¹⁶	22 years	M	Diffuse soft swelling right cheek	Right zygomatic arch, mandibular condyle and lateral pterygoid plate	Surgical excision
Malik et al ¹⁶	32 years	M	Soft swelling back of neck	C2, C7 and posterior elements of cervical vertebrae	NA
Kamal et al ¹⁷	16 years	F	Left facial mass	Zygomatic arch	LeFort I, zygoma osteotomy
Kamal et al ¹⁷	2 years	M	Left cheek mass	None	NA
Unal et al ¹⁸	2 years	F	Soft mass left cheek	None	Debulking with facial nerve preservation
Pires Fraga et al ¹⁹	2 years	M	Left hemifacial tumour	None	Partial and superficial (pre-parotid) removal ^d
Current study	53 years	F	Progressive right facial asymmetry and exophytic right preauricular swelling	Mandibular body, ramus, temporomandibular joint. Maxilla, zygomatic bone	Debulking, right hemimandibulectomy

F, female; M, male; NA, not available.

^a6 interventions done over 10 years starting at 16 years.^bPost-operative facial nerve palsy, ptosis of lower eyelid. Residual asymmetry corrected with liposuction.^cNoted in one out of four cases.^dClinical signs of lesion growth at 3 month follow-up. Patient death 6 months post surgery owing to cardiopulmonary complications.

These changes could be related to increased vascularity, periosteal irritation from overlying mass or regional mesenchymal malformation.^{12–14} Table 1 shows patient data, age at presentation and bone involvement in published reports of CIL-F. This report details clinical and CT features in a case of CIL-F presenting with secondary temporomandibular joint (TMJ) ankylosis, where progressive hemifacial hyperplasia and limitation of mouth opening were undiagnosed prior to CT.

Case report

A 54-year-old female patient reported for CT of the face owing to progressive reduction of mouth opening and right hemifacial hyperplasia. Right facial asymmetry was reportedly present since birth. The patient history revealed surgery for correction of facial asymmetry (debulking and reshaping of the mandible) more than 2 decades previously, for which no surgical records were available.

Clinically, there was marked right hemifacial enlargement and asymmetry involving the middle and

lower third of the face, from the right pre-auricular region to the mandibular midline and from the temporal to the submandibular region craniocaudally. The pre-auricular swelling was exophytic hard causing severely restricted mouth opening. No vascular blanching, cutaneous nevi or stigmata were noted (Figure 1a,b).

CT scan of the face using a SOMATOM Sensation 64 scanner (Siemens AG, Erlangen, Germany) at 55 mAs, 120 kV, revealed hyperplasia of subcutaneous fat on the right side of the face (frontal, temporal, maxillomandibular regions) and the right buccal pad of fat. Muscular fatty infiltration with increased bulk was seen in the right platysma, masseter, temporalis, medial-lateral pterygoid and anterior belly of the digastric muscles along with the intrinsic muscles of the right oral tongue (Figure 2a,b). Fatty infiltration was also seen in the right parotid and submandibular glands. There was thickening of the right greater wing of the sphenoid without definite widening of the skull base foramina.

There was hyperplasia of the right condylar process, including the bony components (condylar head, glenoid fossa, articular eminence) of the right TMJ. Exophytic new bone formation, osteophytosis and sclerotic

a

b

Figure 1 (a) Extraoral profile view of right facial enlargement. (b) Soft tissue rendered three-dimensional volumetric reconstruction with frontal view of right facial asymmetry and large preauricular swelling

remodeling was seen on the lateral aspect of the right TMJ. Joint articulation was noted where the hyperplastic bones of the TMJ apposed each other, forming a functional articular surface. Bony ankylosis was seen laterally to the right TMJ (extra-articular ankylosis) where the exophytic bone mass was contiguous from the temporal to the condylar components of the right TMJ. Thickening of the right mandibular ramus with diffuse trabecular sclerosis and loss of the marrow architecture was also seen (Figures 3a–c and 4) with fatty infiltration into the proximal mandibular canal.

No obvious abnormality was seen in the brain parenchyma, the rest of the muscles, the bones of head and neck and the contents of the visceral spaces of the suprahyoid neck.

Surgical treatment constituted a submandibular neck crease incision followed by mandibular exposure via subperiosteal dissection. Right hemimandibulectomy was performed with release of the extra-articular TMJ ankylosis (Figure 5). Histopathological examination showed non-encapsulated mature adipocytes infiltrating adjacent muscles and soft tissues.

Discussion

Diffuse fatty (–60 HU to –120 HU) infiltration and hyperplasia of multiple bones of the craniofacial skeleton noted in this case represent typical CT features of CIL-F.^{1,5,8,16} The inhomogenous character of fatty infiltration could be due to intervening fibrous elements.⁴ Fat infiltration into the right mandibular canal was seen on CT and also on histopathological examination of the resected mandible.

Articular surface irregularity and evidence of right TMJ remodelling was seen with CT (Figure 3a). However, the severely restricted mouth opening was seen owing to the extra-articular ankylosis and exophytic new bone formation on the lateral aspect of the joint (Figures 3b,c and 4). The findings appear consistent with Type-II TMJ ankylosis.²⁰ Restriction of movement at adjacent joints owing to bone overgrowth has been reported;¹⁶ however, severe restriction of mouth opening owing to TMJ involvement is an unusual manifestation of CIL-F. Bone involvement in CIL-F increases with age (Table 1); therefore, CT is a more relevant primary investigation for older patients as it permits composite radiological descriptions of soft tissue and bony changes.

MRI is useful to confirm the predominantly fatty nature of the condition, with the exclusion of lymphatic/vascular malformations.⁸ High signal intensity (isointense to fat) of the lesion on T_1 and T_2 weighted images (hypointense on fat suppressed images) in young patients is diagnostic in the presence of typical clinical features, thus eliminating need for biopsy.^{4,21} Facial nerve encasement and intracranial/cerebral abnormalities (ipsilateral hemimegalencephaly, asymmetric ventricular–sylvian fissure dilatation, arachnoid cyst, cerebellopontine angle lipoma) can also be seen on MRI.^{22–24} However, MRI features are not always reliable to differentiate CIL-F from well-differentiated liposarcomas.¹⁵ MRI was not considered in this case owing to absence of neurological symptoms, presence of typical CT features of CIL-F without intracranial abnormalities and presence of massive extra-articular TMJ ankylosis.

Differential diagnosis of CIL-F includes lymphatic/vascular malformations, lipomas (intermuscular/intramuscular, angiolipomas/fibrolipomas, liposarcoma and lipoblastomatosis) and congenital overgrowth syndromes causing progressive hemifacial hyperplasia (HH), as seen in CIL-F.^{9,17,19,21} Overgrowth of one or more mesodermal elements (vascular Klippel–Trenaunay–Weber syndrome, neural neurofibromatosis, osseous–fibrous

Figure 2 (a) Axial CT image (soft-tissue algorithm) at the level of the mid-maxillary alveolus shows hyperplastic subcutaneous fat, diffuse fatty infiltration in right buccal, masseteric, parotid and parapharyngeal spaces (arrows). (b) Coronal CT image (soft-tissue algorithm) shows hyperplasia and fatty infiltration of right parotid and submandibular glands (arrows), muscular fatty infiltration in right oral tongue (arrowhead)

dysplasia or varied tissues Proteus syndrome) causes HH, similar to the fatty infiltration in CIL-F.²⁵ Clinical and imaging evaluation will usually permit exclusion of differentials; however, differentiation from lipoblastomatosis and liposarcoma may require histopathology.⁸ Table 2 presents comparative features of CIL-F with some congenital overgrowth syndromes associated with HH.

HH associated with CIL-F has been described previously;^{2,6} striking similarities between the two conditions prompted Macmillan *et al*³³ to suggest that they are the same clinical entity. In their review of the literature on HH since 1881, Kang *et al*⁶ found three cases of HH associated with CIL-F prior to the report by Slavin *et al*¹ and remarked that this rare association

Figure 3 (a) Axial CT image (bone algorithm) at the level of the pterygoid plates shows hyperplastic, deformed right condylar head (short arrow) and new bone formation on lateral aspect of right temporomandibular joint (TMJ) (long arrows). (b) Coronal CT image (bone algorithm) shows hyperplastic condylar head and glenoid fossa (long arrows) with extra-articular bony ankylosis of right TMJ (short arrow). Articular surface irregularity and reduced right TMJ space are also seen (arrowheads). (c) Sagittal CT image (bone algorithm) through mid-right TMJ reveals degenerative changes (*i.e.* erosion and subarticular cystic change) in the right condylar head (small arrows). Note hypodensity of cancellous bone of condylar head and loss of marrow architecture owing to fatty infiltration (arrowheads)

Figure 4 Three-dimensional volumetric reconstruction image shows malaligned mandibular teeth and exophytic right preauricular bony mass

might have been under-reported previously. A possible example is a case of developmental hemifacial enlargement (hyperplasia) associated with a combination of lipoma and lipomatosis.³⁴

CIL-F has a reported¹⁸ post-resection recurrence rate of 62.5% with an average of 3 debulking procedures per patient (Kang *et al*⁶). Delayed surgical resection is recommended to prevent facial nerve damage and reduce the number of debulking procedures.³⁵

Figure 5 Intraoperative photograph shows diffuse fat infiltration

Conclusion

CIL-F may be considered an unlikely differential of facial hemihyperplasia. Typical CT/MRI findings are diagnostic. The osseous overgrowth itself or in combination with previous surgical exploration has a potential to cause exophytic bone formation and TMJ ankylosis. Pre-operative recognition is essential for surgical management to not only release the ankylosis but also debulk the infiltrating lipomatosis in view of high recurrence rates.

Table 2 Comparative features of congenital overgrowth syndromes causing hemifacial hypertrophy and congenital infiltrating lipomatosis of the face (CIL-F)

Condition	<i>Neurofibromatosis</i> ^{26,27}	<i>Klippel–Trenaunay–Weber syndrome</i> ²⁸	<i>Proteus syndrome</i> ^{29,30}	<i>Fibrous dysplasia (McCune–Albright syndrome)</i> ^{31,32}	<i>CIL-F</i> ^{1,3,4,17,21}
Origin	Neural	Vascular	Mixed	Osseous	Fat
Clinical features	Smooth café-au-lait spots	Soft tissue hypertrophy of extremity	Epidermal nevi	Irregular café-au-lait spots	Increased density of facial hair on affected side
	Neurofibromas	Venous malformations	Macrodactyly	Precocious puberty	Hemimacroglossia
	Lisch nodules	Capillary haemangiomas	Scoliosis	Pituitary adenoma	Ptosis of upper lip, enlarged lower lip
	Gracile bones/ribs	Limb overgrowth	Soft tissue/bony overgrowth.	Hyperthyroidism	Early eruption of deciduous permanent teeth
CT features	Hypodense soft tissue with homogeneous enhancement	Hypodense/mildly hyperdense soft tissue	Unilateral precocious dental development	Osseous expansion	Regional macrodontia
		Calcified phleboliths	Distortion of skeletal features with rapid progression	Cystic: fluid-like density	Fatty infiltration
MRI features		Nodular enhancement	Fatty infiltraton of muscles	Mature: ground-glass density	Poorly defined margins
		Marked T_2 hyperintensity	Vascular malformations	Poorly defined peripheral sclerosis	
		Phleboliths	Calvarial thickening	Intermediate to low signal on T_1 WI	Hypertrophy of underlying facial skeleton
			Focal fatty infiltration	Intermediate to high signal on T_2 WI	
	Low signal intensity on T_1 WI	Peripheral nodular enhancement	Asymmetric fatty masses in suprahyoid neck	Heterogeneous contrast enhancement	Diffuse fatty infiltration with T_1 , T_2 hyperintensity
	T_2 hyperintensity with central hypointensity (target sign)				Hypointensity on fat-suppressed images
	Homogeneous enhancement				

T_1 WI, T_1 weighted images; T_2 WI, T_2 weighted images.

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