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HEMIFACIAL INFILTRATING LIPOMATOSIS WITH HEMIMEGALENCEPHALY : COINCIDENCE OR RARE ASSOCIATION

Radio-Diagnosis

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ABSTRACT

The co-existence of hemifacial infiltrating Lipomatosis (HIL) of the face with hemimegalencephaly (HME) that causes facial hemihypertrophy and ipsilateral hemimegalencephaly is very rare. Hereby we present a rare case of 17 years old male presenting with progressive left hemifacial and ipsilateral hemicranial enlargement since birth. Other stigmata concerning for neurocutaneous syndrome i.e. skin lesions, musculoskeletal or neurological abnormalities were not found. In isolation the occurrence of HIL or HME is rare, furthermore co-existence these two scarce conditions become extremely rare. We described the computed tomographic and magnetic resonance imaging findings of these very rare disfiguring conditions affecting unilateral brain and facial structures. In brief their genetic association has also been discussed.

KEYWORDS

Hemifacial infiltrating lipomatosis (HIL), Hemimegalencephaly (HME), Magnetic resonance imaging (MRI), Non contrast computed tomography (NCCT).

INTRODUCTION

HIL first described by Slavin in 1983 is a rare benign lipomatous pathology which describes a non-encapsulated collection of mature adipocytes infiltrating the local tissue and hyperplasia of underlying bone leading to a craniofacial deformity. It's etiopathogenesis is not well understood yet and always unilateral in nature. PIK3CA and FGFR3 gene Mutation could be the possible factor. [1]

HME is a major congenital malformation of the brain, characterized by hamartomatous overgrowth of unilateral cerebral hemisphere. The etiology of HME is unknown but it has been postulated by mechanism of congenital disorder of symmetry and cellular lineage at third week of gestation with positive correlation between epidermal growth factor and excessive proliferation of neuronal tissue.[2]

However, the co-existence of these two pathologies together is extremely rare and here we report a similar case.

Case Report

A 17-year-old male presenting with left hemifacial swelling since birth, his antenatal, developmental & family history were non-significant. He had no history of seizure or neurological deficit. Physical examination showed facial asymmetry with left side of the face was swollen without any overlying skin abnormality (Fig. 1). Oral examination demonstrates ipsilateral macroglossia with hypertrophy of the teeth. On palpation, the mass was diffuse soft, non-tender and non-fluctuating in nature. In view of the asymptomatic nature and slow progression of the lesion, a lipomatous tumor, namely lipoma, was suggested.

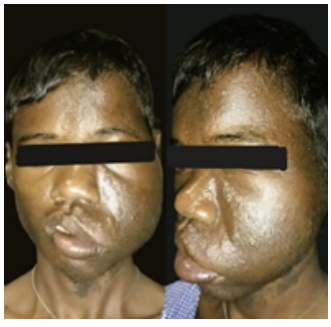

Fig 1 showing an adolescent male with ipsilateral (left) hemifacial swelling.

NCCT Head + Face along with MRI Brain + Face scan was performed. CT face images (soft tissue Window) shows diffuse hypodense fatty infiltration in left hemiface extending from superficial to left temporalis muscle, extending downwards to reach left malar, left jugulo-labial region and reaching upto left submandibular region. (Fig. 2 A, B, C, D).

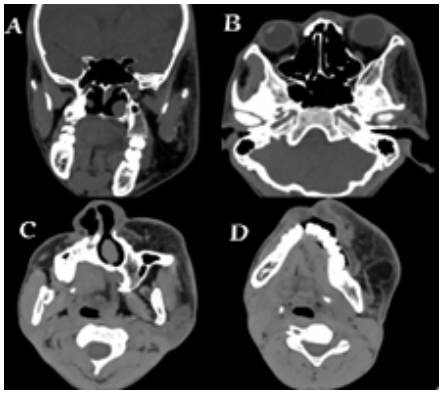

Fig 2 NCCT head+face soft tissue window coronal & axial view (A, B, C & D) showing hypodense lipomatous proliferation in subcutaneous plane of left hemiface.

In bone window and 3D reconstruction CT shows hyperplasia of underlying left frontal, zygomatic, maxillary, hemimandible and teeth. (Fig. 3 A, B, C, D).

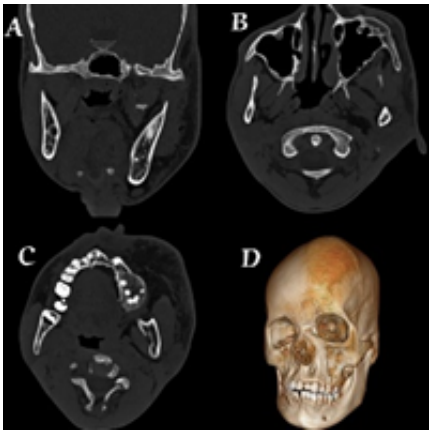

Fig. 3 NCCT head bone window axial & coronal view with 3D reconstruction (A, B, C & D) images showing bony hyperplasia of right hemiface.

Subsequent MRI Face revealed the presence of diffuse lipomatous tissue (T1/T2 hyperintensity with suppression in T1 fat saturation sequence) in the left cheek region, left pterygomandibular space, subcutaneous tissue superficial to antero-lateral wall of the maxillary sinus and adjacent to the left hemimandible. (Fig. 4 A, B, C, D).

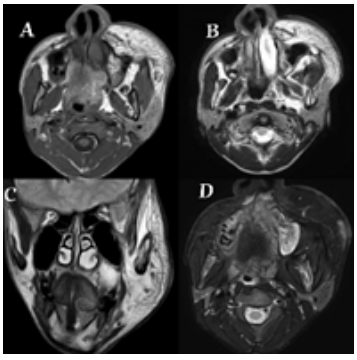

Fig 4 MRI FACE images, A. Axial T1, B. Axial T2, C. Coronal T2 Weighted images showing diffuse T1 & T2 hyperintense proliferated subcutaneous fat involving left hemiface which is showing suppression on T1 Fat Saturation (image D).

The MRI brain & NCCT head shows mild asymmetry and hypertrophy of the left hemisphere with few punctate deep white matter hyperintensity. However, no obvious midline shift or cortical dysplasia noted which corresponds to grade I HME(Fig. 5 A, B, C, D, E, F) [2]. In view of above clinical history, examination and radiological findings the diagnosis of HIL with HME was made.

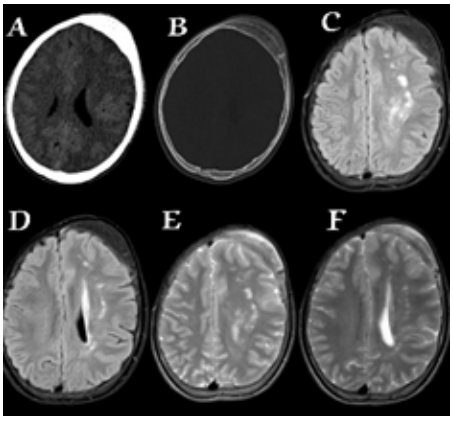

Fig 5. NCCT head brain window showing hypertrophy of left cerebral hemisphere with associated hypertrophy of frontal bone in bony window (Image A & B). MRI brain FLAIR & T2WI (image C, D & E, F) showing hemihypertrophy of left cerebral cortex with associated multiple punctate hyperintensities involving underlying deep white matter.

DISCUSSION

HIL is characterized by unilateral facial enlargement with adipocytic infiltration involving single or multiple tissues including the underlining bone on the same side. Chromosomal and neural abnormalities, unfavorable intrauterine environment, endocrinological abnormalities, anatomical and functional defects of the vascular and lymphatic systems are few of its etiological factors. [3,4]

Dyke-Davidoff- Masson syndrome, Neurofibromatosis, Beckwith Weideman syndrome, tuberous sclerosis, Parry Romberg syndrome & Proteus syndrome should be kept in mind before reaching to the diagnosis of HIL with HME [2,5]. The management in our case was conservative and follow-up since he is asymptomatic and cosmetic surgery will be advised with complete skeletal maturity attainment.

CONCLUSION

The co-existence of HIL with HME is extremely rare with very few cases has been reported yet [5]. Since the clear cause of association between HIL & HME has not been established yet and needs to be studied. The imaging study of choice for diagnosis of this condition is MRI Brain. In our case, both CT and MRI revealed that facial asymmetry was caused by enlargement of both subcutaneous tissues and the underlying skeleton with enlarged ipsilateral cerebral hemisphere. The imaging features with histo-pathological correlation becomes the high yield diagnostic tool to establish the diagnosis.

REFERENCES:

1. M. Siradji Harouna, H. B. (2021). Facial infiltrating lipomatosis, a rare cause of facial asymmetry to be known: Case report and literature review. Annals of Medicine and Surgery, 1-4 doi:10.1016/j.amsu/2021/103.118
2. Flores-Sarnat, L. (2002). Hemimegalencephaly: Part 1. Genetic, Clinical, and Imaging Aspects. Journal of Child Neurology, 373-383. doi:10.1177/08830738020170051 2
3. Shanmugasundaram, K. (2016). Congenital Hemifacial Hyperplasia: Clinical Presentation and Literature Review. Hindawi Publishing Corporation, 1-5. doi:10.1155/2016/5260645
4. Arora, P. C. (2012). Lipomatosis: a diverse form of hemifacial hyperplasia. Imaging Science in Dentistry, 191-195. doi:10.5624/isd.2012.42.3.191
5. Ajaj, S. S. (2022). Hemimegalencephaly with prominent ipsilateral facial hypertrophy. Marshall Journal of Medicine, 4(1), 1-5. doi:10.33470/2379-9536.115