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Congenital Isolated Hemifacial Hypertrophy: Clinical and 3-Dimensional Radio graphical Scanning Features

Ashish Agarwal[1]*, ND Gupta[2], Anshul Aggarwal[3], Rati Goyal[4]
1. Department of Periodontics, Institute of Dental Sciences, Bareilly, India
2. Department of Periodontics, Dr ZA Dental College, AMU, Aligrah
3. Deptt of oral pathology, oral medicine and radiology, Dr ZA Dental College, AMU, Aligrah
4. BDS (Private Practioner)

ABSTRACT

Hemifacial hypertrophy is a rare condition characterized by facial asymmetry caused by marked, unilateral, localized overgrowth of one or more tissues in the affected area, including the facial muscle, bones, and teeth. The cause of this condition is unknown. These asymmetries are often noted at birth and accentuated with increasing age, especially at the time of puberty. Hemifacial hypertrophy is more likely the mild form of congenital hemihypertrophy. Here we present the case of a 25 year old male patient affected by isolated congenital hemifacial hypertrophy with significant social stigma and major aesthetic morbidity. A retrospective review of the patients' case notes, clinical photos and three dimensional radiographic imaging was undertaken to assess condition, with particular reference to function and aesthetics. Congenital hemifacial hypertrophy can be assessed by clinical features, and tomographic imaging. Most of the cases, reported in previous literature, mentioned with traditional radiographic techniques of facial region. We characterized this condition with advanced diagnosis findings. The purpose of this case report of hemifacial hypertrophy is to supplement existing literature, and clinical knowledge with advanced diagnostic aid like three dimensional computed tomography imaging.

Keywords- congenital abnormality, congenital hypertrophy, hemifacial hypertrophy

*Corresponding Author Email: draggarwal.xray@gmail.com
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INTRODUCTION

Asymmetry is common and one of the most unusual and interesting error of human reproduction. Most of us have an insignificant degree of asymmetry of the two halves of the body. But in certain number of cases there may be a significant degree of hypertrophy of one side or a part of the body as compare to other. Congenital hypertrophy is a developmental disorder, characterized by marked unilateral asymmetry, in which the structural disproportions variation from one side of the body to the other counter part is far exceed than commonly seen in normal population [1,2] . Hemifacial hypertrophy (HFH) is a rare condition in which there is unilateral enlargement of the face and teeth. Various views for the origin of hemifacial hypertrophy have been implicated including hereditary, chromosomal abnormalities, atypical type of twinning, hormonal disbalance, altered intrauterine environment, anatomical and functional anomalies of lymphatics/vascular system and disturbance of central nervous system [3,4].

CASE REPORT

A 25 year old male patient reported to the Department of Periodontics, Dr Za Dental College AMU, Aligarh for seeking the treatment for his dirty and stained teeth. On the first sight the general appearance of the patient was that of quite a large young man who had a very peculiar stooping attitude of a general unkempt appearance. The contour of the face was that of an oblong, and strikes one at first glance as a most unusual type. The left side of the face was most noticeable than right half. On taking the history it was evident that the patient was second siblings born of a non-consanguineous marriage at full term normal spontaneous delivery with a birth weight of 3,600 g and there was no significant perinatal and post natal problem. However in the months following birth, the parents observed progressive enlargement of his left cheek and different eyelid width. All sensory and motor responses of the patient were with in normal limit. There was no history that any family member was affected. All vital sign of the patient was in between the normal limit. No sign and symptom go for any systemic involvement.

The detailed extraoral physical examination revealed an obvious enlargement of the left facial osseous component and soft tissue, compare to the right half of the facial region. The supra-orbital ridges were prominent; the left ridge was higher than the right ridge. The malar bone of the left side was quite prominent. The lips, particular the lower lip was twice large on the involve side than the normal size. The enlargement of the upper lip was also seen and this displaced the philtrum towards the uninvolved hemifacial side. The lip line was inclined towards the affected side. (Figure. 1)

On intraoral examination enlargement of the left maxillary and mandibular gingiva was revealed which abruptly stop at the midline. In dental anomalies distortion of the size and shape of the crown was noticed on the affected side. Generalized macrodontia, malocclusion, and diastema was seen on the ipsilateral side. (Figure 2, 3) Macroglossia, with polyploid excrescences representing enlarged fungiform papilla, were seen which was particularly involve the anterior two third portion, and this enlargement of tongue was precisely terminate at the midline. (Figure 4)

The computed tomography scanning of the facial region showed the hypertrophy and expansion of left half of maxillary and mandibular arch with macrodontia.(Figure 5,6) .including head and condyle with widening and expansion of left temporomandibular joint. Hypertrophy of left zygomatic arch with head of condyle was also revealed. (Figure7,8) .this unilateral osseous deformity of osseous component can easily be appreciated in 3d scan of the concerned region (Figure 9)

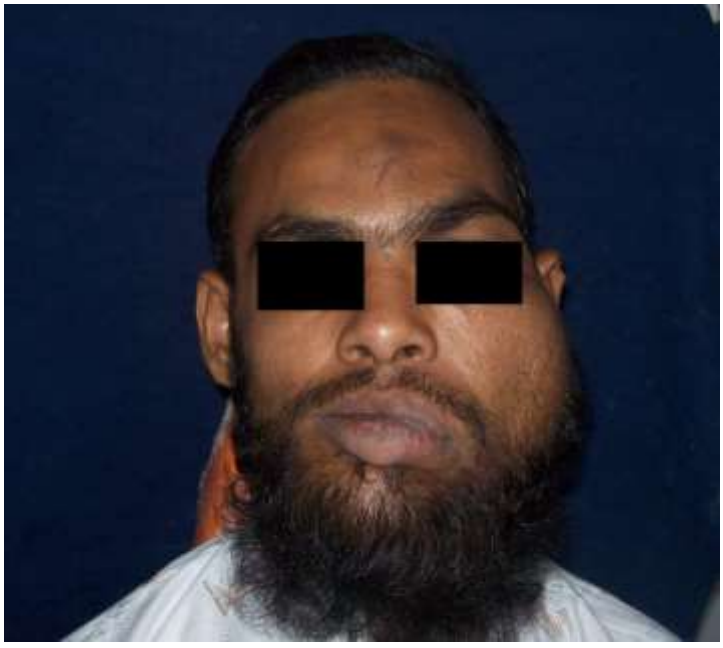

Figure 1:Extraoral view. Marked unilateral facial hypertrophy on the left side. Lips thickened, drooping of corner of mouth, high supraorbital prominence on involved side. Deviation of philtrum on opposite side.

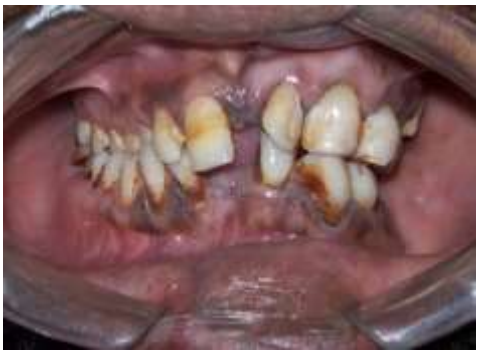

Figure 2: Intraoral view. Unilateral macrodontia of left side.

Figure 3:Intraoral view of maxillary dentition. Unilateral macrodontia & malocclusion.

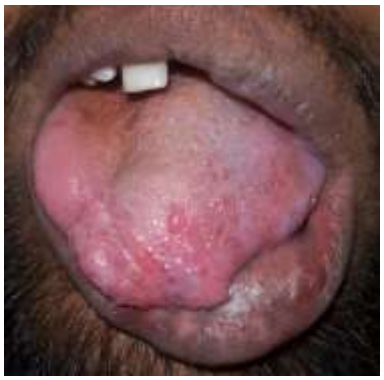

Figure 4: Macroglossia with enlarged fungiform papilla

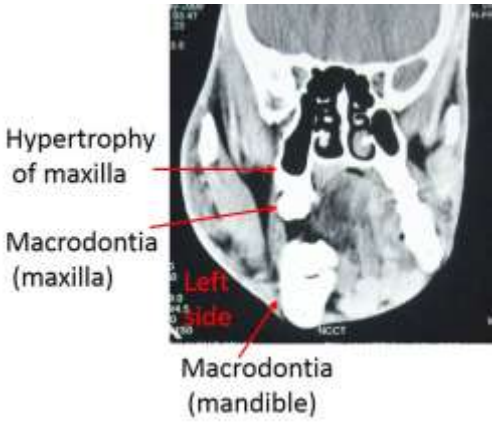

Figure 5: CT scan (Axial section) section showing hypertrophy of maxillary arch and macrodontia of left side.

Figure 6: CT scan (Transverse section) showing hypertrophy of left mandibular arch

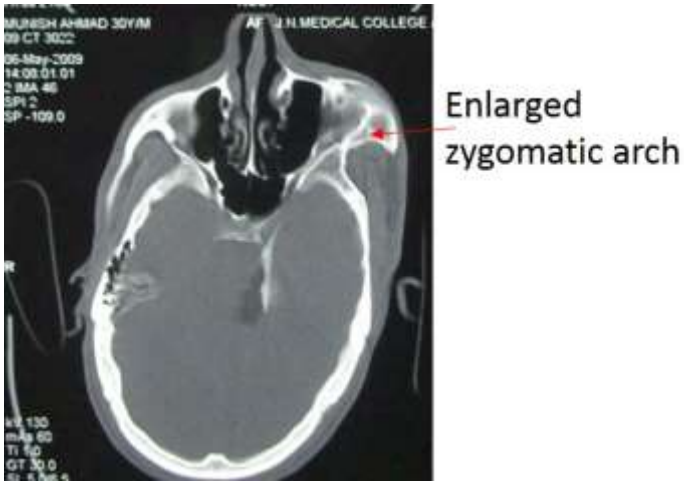

Figure 7: CT scan (transverse section) showed enlarged zygomatic arch

Figure 8: CT scan (transverse section) showing hypertrophy of head of condyle of left side.

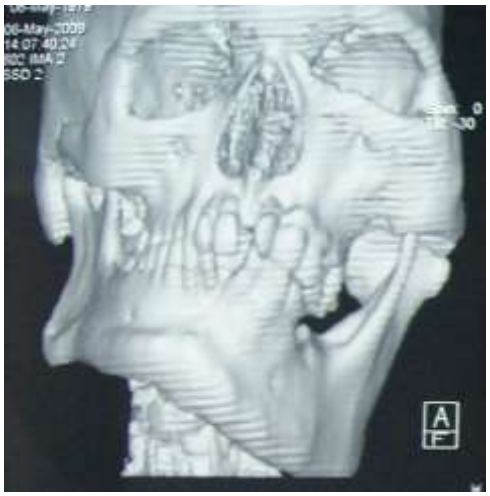

Figure 9: CT-3D scan showed unilateral deformity of osseous component

These extensive clinical and radiographic abnormalities were considered to justify a diagnosis of hemifacial hypertrophy. Condylar hyperplasia of the affected side and hemifacial hypoplasia of opposite side may be considered as differential diagnosis. The patient

underwent dental treatment, including restorations and prophylaxis. Periodic clinical and radiographic assessment was recommended to the patient

RESULT AND DISCUSSION

Subtle, asymmetrical variations of the contralateral structures of head and face occur commonly in the general population, and are an accepted feature of the morphogenesis. Occasionally a gross asymmetry easily perceptible to the eye may occur either due to physiological reason or due to growth abnormality [5]. One such condition is hemifacial hypertrophy, a rare maxillofacial dysmorphosis of debatable etiology, and is frequently associated with relatively serious appearance and function disorders depending of the degree of hypertrophy [6]. This entity has been term variously in literature as facial hemihypertrophy, partial unilateral gigantism and hemifacial hyperplasia [7]. In literature hemifacial hypertrophy was first documented by Meckel[8] in 1822. Rowe NH[2] in 1962 classified the congenital hemifacial hypertrophy as follows -

True hemifacial hypertrophy - Exhibiting unilateral enlargement of viscerocranium, bounded superiorly by frontal bone (not including the eye), inferiorly by inferior border of mandible, medially by the midline and laterally by the ear, including the pinna with enlargement of all tissue, teeth, muscle, bone and soft tissue in this area. Partial hemifacial hypertrophy - If not all structure are enlarged to the same degree or limited to the single structure.

Gesell[9] in 1927 described hemihypertrophy as "essentially a developmental anomaly antedating birth and arising in some way as partial deflection of the normal process of birth."

No single theory explains the etiology for this condition. Gesell [10] in 1921 proposed that pathogenesis of some form of congenital hypertrophy is due to chromosomal anomalies. In his second theory in 1927 Gesell [9] suggested that at the time of division, the zygote divided into two asymmetrical daughter cells and this unequal twinning lead to hemihypertrophy. Hemifacial hypertrophy is most likely a process that occurs early in the embryonic development during formation of the first branchial arch. Franheusar and Humphery [11] in 1959 dictated the disorder in the axolotl to diploid, triploid and tetraploid chromosal aberrations. Pollock et al [12] in 1985 proposed a new hypothesis for congenital hypertrophy according to which neural tube is the basic template for the early establishment of size and shape of the body. According to them one neural fold is larger than other, due to this neural crest cell are proportionally more in number on the involved side. This increase number of neural crest cell leads to unilateral overgrowth of crest cell derived bone, muscle and soft tissue.

Hemifacial hypertrophy is more common in men than in women and more commonly involves the right side of the face[2]. In keeping with this, the present case was a boy but

unusually left side of the face was affected.

Associated features of reported cases of facial hemihypertrophy have documentation of a wide range of maxillofacial and oral findings. Involvement of osseous component appears in the form of asymmetric growth of frontal bone, maxilla, palate and mandible. Dentition abnormalities with respect to crown size, root development, root size and shape, rate of development and number of teeth are seen[13]. Macrodontia is not uniform for all teeth; it is more in canines followed by premolars and first molar and least occurring in incisors, second molars and third molars. Deciduous dentition is generally not involved in asymmetric development. Early eruption of permanent dentition by up to 4-5 year can be reported [2,14].

Many features are also reported in the present case.

Various entities showing similar manifestations should be excluded. These include: hemiatrophy, showing unilateral underdevelopment with muscle weakness and neurological deficit; fibro-osseous lesions and other bony tumors, which do not involve the soft tissues and dentition; vascular and lymphatic malformations, cutaneous lesions, and neurofibromatosis, which are usually bilateral, involve different body parts and are diagnosed histologically. Other syndromes such as Beckwith-Wiedemann syndrome, Proteus syndrome, Schimmelpenning (epidermal nevus) syndrome, were excluded. There may be overlap of clinical manifestations between hemifacial hypertrophy and these conditions [15,16,17]. In Proteus syndrome, the patient has asymmetry of the limbs, overgrowth of the hands or feet or both, lipomas, connective tissue nevi, and vascular and lymphatic malformations. In Beckwith-Wiedemann syndrome, patients usually have increased birth weight, postnatal gigantism, macroglossia, omphalocele, and distinctive ear lobe grooves. In Schimmelpenning syndrome (epidermal nevus), patients have epidermal nevi; sebaceous nevi are said to be the hallmark of the syndrome. Other common features of Schimmelpenning syndrome are seizures, developmental delay, hemangiomas, kyphosis/scoliosis, and extension of nevus to the eyelid [17]. Segmental odonto maxillary dysplasia shows unilateral enlargement of the maxilla and fibrous hyperplasia of the overlying gingival tissues. All these conditions should be distinguished from hemifacial hypertrophy on the basis of specific clinical and radiographic findings [8,15,16].

Ringrose et al.[5] analyzed 129 cases of hemihypertrophy, finding the following as the most common features: mental deficiency; skin abnormalities; varicose veins; and compensatory scoliosis. Other clinical findings reported in the literature are thickened hair and skin, ipsilateral nevi and telengiectasis, macrodactly, polydactyly, syndactyly, and club foot [15]. Pfister et al. [18] showed that abdominal tumors are usually present in hemifacial hypertrophy of the body. However, in isolated hemifacial hypertrophy, not even a single case has been

disclosed with abdominal tumors [8]. As our patient was of poor socioeconomic status and neither she nor her parents ever complained of any abdominal problem, we did not perform ultrasonography, although we explained to her parents that she might require this examination for future management. Hemifacial myohyperplasia has been added as a new clinical finding in this disease, where hyperplasia is only limited to the facial musculature [19].

Congenital hemifacial hypertrophy is expressed as facial asymmetry resulting from unilateral overgrowth. Yoshimoto et al. compared the proliferative activity of primary cultured osteoblasts taken from the hypertrophic side with those taken from the normal side. They reported that both fetal calf serum and basic fibroblast growth factor stimulated osteoblast DNA synthesis and that cultured osteoblast proliferation was markedly increased on the hypertrophic side. They concluded that fibroblast growth factor and its receptor signal transduction axis may be selectively involved in affected osteoblasts, leading to hypertrophy [20].

Treatment of hemifacial hypertrophy remains limited at this time. Various craniofacial and maxillofacial surgical procedures with multidisciplinary team approach rather than restricting the patient to single specialist have been performed to improve function and appearance and are an option at the end of growth in patient with severe dysplastic changes[15]. Preoperative evaluation by routine and cephalometric radiographs and by computerized tomography allows surgical planning. These may include reconstructive procedures such as osteotomies or orthognathic surgical procedure and soft tissue debulking by excision of excess masticatory and subcutaneous tissues, with preservation of neuromuscular functions [8,15]. congenital hypertrophy is generally associated with good prognosis. An extensive search of the English language literature revealed no formal reports of malignant transformation. The asymmetry usually remains constant up to the end of adolescence and the condition stabilizes thereafter In adolescence and adulthood social stigma may occur, so psychological support is also required.

CONCLUSION

Hemifacial hypertrophy is a rare development anomaly characterized by unilateral enlargement of head and teeth. Hemifacial hypertrophy may occur a wide spectrum of defect or may involved muscle or bone. A variety of etiological factor have been proposed for this maxillofacial dysmorphosis. Clinical and radiographic information must always be kept in mind to achieve an accurate diagnosis.There is no specific treatment for this condition other than attempt at cosmetic repair.

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