Hemifacial Hyperplasia: Two Extremes of Age Presentation

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ABSTRACT
True Hemifacial hyperplasia is a scarce and obscure disorder, as much of cases have not been mentioned in literature. It’s a congenital disorder affecting half of face and is slow growing which stops after adolescence. Herein we report two cases of the same with two extreme of age presentation. One of the case having hyper pigmentation of the affected half. This added feature represents a new syndrome or it’s a syndrome in itself?
Keywords: Hemifacial hyperplasia; Skin pigmentation; Congenital disorder

INTRODUCTION
Congenital hemi hyperplasia is a rare congenital developmental disorder characterized by asymmetrical overgrowth of one or more body parts [1]. Described initially by Meckel in 1822 and later reported by Wagner, et al. [2]. In 1962 Rowe classified hemi hypertrophy into complex hemi hypertrophy which involved entire half of the body, simple hemi hypertrophy affecting one or both limbs, and hemifacial hypertrophy which involved half of face. He classified hemifacial hypertrophy further into true hemifacial hypertrophy which exhibits unilateral enlargement of all tissues, teeth, bones, and soft tissues, inferior border of the mandible inferiorly, midline medially, and ear including the pinna laterally and partial hemifacial hypertrophy. Were not all structures are enlarged to the same degree or limited to one structure [3]. Here we report two cases of true hemifacial hyperplasia with the aim to add the existence of this condition and knowledge on true hemifacial hyperplasia and its differentiating condition, along with enhanced skin pigmentation.

CASE 1
A 12 years old boy reported to our Bokaro general hospital maxillofacial OPD with the chief complaint of swelling on right side of face since 3 years (Figure 1). Patient was asymptomatic 3 years back when he complained of swelling on right side of his face. Patient gives a history of fall from stairs 5 years back while playing with his cousin. He and his mother both are unaware of side from which patient fell. According to his mother he had only small laceration on upper lip, with mild bleeding which stopped after sometime. There is no history of unconsciousness, vomiting or bleeding from any other region.

Figure 1: Showing facial profile.

Swelling increased from front of right ear to, right cheek region and then involvement of eye region. Increase in swelling was not associated with pain. Pain was only experienced while touching and eating hard food rest of the time he had no complains. No significant past history of illness. No significant dental history. On general examination patient was well oriented to person, place and time. He weight 24 kg, Height- 128.5 cm with left shoulder slightly drooped down. No abnormality detected on examination of CVS, Respiratory examination. His vital were...
within normal limits. Facial examination revealed entire enlarged right half of face. Submentovertex view showing deviated nasal tip to left side, enlarged right nostril and right upper half lip (Figure 2).

Temperature of right half of face was slightly raised. The facial swelling had a smooth surface, soft consistency, fluctuant. Also the swelling was not translucent. Patient also complains of recurrent redness of right eye, blurring of vision on right side for which he was referred to ophthalmologist. His right ear pinna was enlarged and tenderness at preauricular region. Intraorally there was swelling which extended from mid palatal suture to posterior palatal seal, anterior faucial pillar, maxillary tuberosity, buccal gingiva up to maxillary anteriors. Buccal sulcus not obliterated. There was spacing between the teeth of involved side, alveolar process thick especially in premolar and molar region, midline shift was present (Figure 3).

posterior open bite of right side but left side had class I occlusion. Cheek bite on right side. There was linear groove between mid palatal suture and right palatal gingival. Slight occlusal cant was present (Figure 4). Distinct tooth size discrepancy was observed between right and left side (Figure 5).

An OPG (Figure 6) true lateral cephalogram (Figure 7) and CT scan (Figure 8) was advised. On the basis of clinical, radiological and histopathological findings diagnosis of hemifacial hyperplasia of right side was confirmed.
CASE 2
A 58 year old male patient reported to our OPD with a chief complaint of painful mobile teeth after trauma. He had history of road traffic accident. The patient was well oriented with stable vital signs. On clinical examination it was seen that he had gross enlargement of right half of face (Figure 9). While taking history he revealed that he had asymptomatic swelling in his right cheek region since birth which had gradually increased to present size and ceased to grow when he was 20 years. There is no significant family history.

The swelling extended superiorly to upper canthus of right eye, inferiorly up to 5 cm below the lower border of mandible, anteriorly until nasolabial fold, and posteriorly till the tragus of the ear. Nose and chin were deviated to left side. Enlarged soft tissue mass was observed involving maxilla, mandible, and zygoma on the affected side. Right ear (Figure 10) and right half of lower lip were remarkably enlarged (Figure 11) and incompetent. On palpation swelling was nontender, hard in consistency, and noncompressible. Skin over the involved area was hyper pigmented. This hyper pigmentation started from pinna upto clavicle. Temporo mandibular joints movements were normal. Regional lymphnodes were not palpable. On intraoral examination, there was sufficient mouth opening. Enlarged right maxillary and mandibular alveolar arches, upper and lower labial mucosa, and buccal mucosa were observed.

Figure 9: Gross swelling of right side of face.

Figure 9: OPG reveals diffuse enlargement of skeletal and dental hard tissue enlargement on right side of the face.

Figure 7: Lateral cephalograms.

Figure 8: Axial CT scan showing lipomatous enlargement of the soft tissue on right side of face including buccal region, lips, tongue and soft palate. Also we can appreciate the enlarged right maxillary alveolus.

Figure 10: Right ear enlarged as compared to left.
Macroglossia of right half of tongue was seen with the tip deviated to left side. Dorsum surface of the tongue had polyloid excrescences representing enlargement of the fungiform papillae (Figure 12). Tooth size discrepancy could not be much appreciated as multiple teeth were missing. Orthopantomogram (OPG) figure revealed an obvious diffuse enlargement of right side coronoid, condylar processes, and lower border of mandible, inferior alveolar canal and jaws. Computerized tomography (CT) scan of the face revealed enlarged right petrous part of temporal bone, pituitary fossa, maxilla, mandible, condyle, zygoma, right orbit and cranial bones. Deviation of nasal bone and chin was observed towards left side due to an obvious enlargement of overlying soft tissues on right side of the face. No systemic abnormality was noted.

Routine blood investigations were also under normal limits. Based on the clinicopathological findings, a final diagnosis of Congenital Hemifacial Hyperplasia (CHH) was made. The patient refused to undergo extensive surgical procedures.

DISCUSSION

Geiser et al in 1970 suggested the term hemi hyperplasia to be more appropriate than hemi hypertrophy, as there is more increase in cell number than the increase in cell volume [4]. Hemifacial hyperplasia (HFH) was first described by Beck in 1836 [5].

True HFH is a congenital morphological anomaly resulting in facial asymmetry due to unilateral overgrowth of all tissues or part of the tissues on the affected side including soft tissues, bone, and teeth excluding sparing the eye [6,7]. The prevalence rate of HFH is 1:86,000 live births, with men affecting more than woman and right side more affected than left [8]. It has a special feature that it is present at birth, grows exponentially and then stop and stabilizes after adolescence[9]. Etiology of this condition is not well explained in literatures, but few possible factors have been mentioned such as; endocrine dysfunctions, chromosomal abnormalities, central nervous system disorders, vascular or lymphatic malformations, and somatic mutations, heredity, disturbances in the development of the first branchial arch overgrowth etc. [10]. HFH may be seen with other associated conditions such as acromegaly and pituitary gigantism or hypertrophy of other parts of the body [11].

Our present two cases demonstrates nearly all key imaging features of true HFH, including enlargement of all bones, soft tissues, teeth, ear and skull base foramina. Our second case also demonstrates unique feature of skin pigmentation of the affected area previously unreported.

HFH should be always differentiated from other conditions where we can see enlargement of half face. Few differential diagnoses are as follows [12]:

1. Benign fibroosseous lesions where overgrowth of bones, soft tissues, and skull base foramina are seen such as fibrous dysplasia, Pagets disease, Ollier disease, osteosarcoma and chondrosarcoma.

2. Conditions where there is unilateral enlargement of teeth and tongue: neurofibromatosis, Proteus syndrome, Beckwith-Wiedemann syndrome, epidermal nevus syndrome, hyperpituitarism, etc.

3. Hemifacial lipomatosis and congenital infiltrating lipomatosis where we can see asymmetry of mandible.

4. Conditions where facial muscles are predominantly hypertrophied.

CONCLUSION

True HFH is a scarce and obscure disorder, as much of cases have not been reported. Here we present two cases of true HFH with one case having hyper pigmentation of the affected half. So question arises that does this hyper pigmentation in conjunction with HFH represent a new syndrome or it’s a syndrome in itself. More such cases should be explored to answer this query.

REFERENCES


