# UNDERSTANDING HEMIFACIAL MICROSOMIA: CORRELATION OF CT AND CLINICAL FINDINGS

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## ABSTRACT

**OBJECTIVE:** Using clinic-radiological approach, describing fundamental CT findings defining hemifacial microsomia and incorporating the format of OMENS/ OMENS modified classification system. **MATERIALS AND METHODS:** 12 patients selected from the database; 6 males and 6 females with the age ranging from 7 months to 22 years. **RESULTS:** Mandibular abnormality was observed in 83 % of patients, out of which 60% had mild, 20 % moderate and 20 % had severe deformity. Soft tissue deficiency observed in 75 % of patients of which 44% mild, 22 % of moderate and another 33 % with severe degree of deformity. Macrostomia was seen in 66 % of patients. The auricular deformity was seen in 58 % of cases. The orbital involvement was present in 33 % of cases ranging from mild orbital asymmetry to dytopia. **CONCLUSION:** The modified OMENS clinical classification system is so for the most accepted system for describing features of HM. Using the multi detector CT (MDCT) as imaging modality and incorporating the findings into the format of modified OMENS system a clinico-radiological approach was sought by which we were able to improve the perception of disease process and delineated imaging findings in much better manner with lesser chance of error.
**Key words:** Deformities,Asymmetry, Hemifacial microsomia, Goldenhar's syndrome, OMENS classification, CT Imaging.

**Key-points:**
1. Hemifacial Microsomias is genetic defect causing major cosmetic abnormality to patients
2. CT scan reformatted images serves main method of imaging.
3. Modified OMENS classification system overcomes difficulty in diagnosing and eventually treatment.

## Introduction

In the simplest terms, hemifacial microsomia manifests primarily as unilateral hypoplasia of the craniofacial skeleton and its overlying soft tissue. Although bilateral hypoplasia has been noted in 5 to 30 percent of cases,[1,2] when present, it is generally asymmetric.[3,4] Craniofacial microsomia or otomandibular dyostosis, other names of this disorder, is a spectrum of soft and hard tissue hypoplasia typically affecting the regions of Tessier's # 6, # 7, and # 8 facial clefts. It is a disorder of the first and second branchial arches with an incidence as high as 1 in 3000[5] live births. Mandibular pathology is the most evident and occurs to some degree in 89% to 100% of the patients with craniofacial microsomia. 66% to 99% of hemifacial microsomia patients have microtia. Other abnormalities of the ear include conductive hearing loss, middle ear deficiencies, and preauricular skin tags.

The classic presentation is unilateral mandibular hypoplasia, deficient soft tissues of the face and microtia. Gorlin's term, hemifacial microsomia, emphasizes that the disorder is usually a unilateral facial deformity (70-95% of the time) and should not be confused with Treacher Collins Syndrome (Also a Tessier's # 6, # 7, and # 8 facial clefts), which is bilateral and symmetric.

**EMBRYOLOGY OF THE FIRST AND SECOND BAs AND ASSOCIATED STRUCTURES**
Development of the craniofacial structures is a com-

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plex process that proceeds in an orderly fashion throughout embryonic and fetal stages of formation. Craniofacial growth occurs due to a relatively rapid and orderly composition of mesodermal and cranial neural crest cells via a complex signaling network. Syndromes of the first and second branchial arches (BA) manifest along a spectrum of hypoplasia and aplasia of the structures composing these arches. Some differences between abnormalities of the first and second BA derivatives may reflect differences in the embryologic age at the time of the insult with respect to neural crest cell migration. Other changes are related to deregulation of cell-signaling pathways triggered by a combination of genetic and environmental factors.

## VARIATION

Hemifacial microsomia is known for its wide spectrum and variation that ranges from the mild forms, with only preauricular skin tags, to moderate forms, which may have a hypoplastic mandible and diminished cheek fat but a near normal external ear, to the severe form of Goldenhar Syndrome (oculovertebral sequence). Goldenhar syndrome is a severe variant of craniofacial microsomia defined by colobomas of the eyelid and cervical vertebral anomalies, as well as the other head and neck sequelae of craniofacial microsomia, including mandibular hypoplasia, macrostomia, and microtia.

## ASSOCIATED ANOMALIES

The primary anomalies of hemifacial microsomia are that of mandibular hypoplasia, microtia, and deficient soft tissues of the face. These patients also have a high incidence of preauricular skin tags, conductive hearing loss, hypoplastic glenoid fossa, and cranial nerve VII palsy. Also associated are macrostomia and masicatory muscle hypoplasia. Some of these patients are born with upper airway obstruction related to micrognathia, with related posterior tongue-base collapse. Significant upper-airway obstruction may require tracheostomy or early mandibular intervention to improve the mandibular position. Many of the patients with craniofacial microsomia and upper airway obstruction also have symptomatic gastroesophageal reflux. There is a high incidence of bony midfacial deformities mostly seen as maxillary and zygomatic hypoplasia contributing to these patient's occlusal

cants. Temporal and frontal deficiencies may also be seen. Cleft lip and/or palate have been reported.

## CLASSIFICATION SYSTEMS

Several clinical classification systems are described in the literature focusing the subject of hemifacial microsomia. In 1960, Pruzansky published an article describing the mandibular anomalies of hemifacial microsomia into three grades (types I through III) of increasing hypoplasia based largely on the morphology of the ramus and condyle.[6] The Pruzansky classification was later modified by Kaban and colleagues[7] and is replicated with a minor modification in nomenclature, in the mandibular portion of the OMENS classification system. Another classification scheme focusing exclusively on the external ear and divides malformations into three grades of increasing severity was developed by Marx[2] and later modified by Meurman.[4] These three grades, similar to Pruzansky's mandibular grading, range from mild effacement of auricular architecture to nearly complete auricular aplasia. Other skeletal classification systems include those of Harvold and colleagues[8] and Lauritzen et al.[9] Both of these systems define five groups of skeletal deficiencies. Huisinga-Fischer and colleagues[10] developed a computed tomography-based system for describing the skeletal malformations of hemifacial microsomia. This system consists of a mandibular deformity scoring system that grades mandibular hypoplasia and a cranial deformity scoring system that grades the hypoplasia of other facial bone. One mixed feature skeletal, auricular, and soft-tissue classification system developed by David and colleagues[11] was used to independently analyze skeletal, auricular, and soft-tissue malformations in 47 patients. None of the aforementioned classification systems are widely accepted and adopted.

One of the recent classification systems, the OMENS system **(Appendix I & II)**, scores five clinical manifestations of hemifacial microsomia according to dysmorphic severity on a scale from 0 to 3: *o*rbital asymmetry, *m*andibular hypoplasia, *e*ar deformity, *n*erve dysfunction, and *s*oft-tissue deficiency. The OMENS classification is one of the most commonly used systems. Later it was modified with addition of acronym OMENS +, to describe the extra craniofacial anomalies and recently in 2006 Bartlett et al[15] suggested the pictographic representation of the

spectrum of disease in the OMENS classification system by adding macrostomia (teisser cleft #7) in the classification system.

### IMAGING TECHNIQUE

CT is the imaging technique for studying syndromes of the first and second BA. The multi-detector CT (MDCT) provides fast scanning capability and limits motion artifacts. The modern MDCT offers excellent multi planner reformats (MPR), curved multi planner reformats (cMPR), and 3D and volume rendered (VR) images which help clinicians & radiologists to understand the complex anatomy of craniofacial structures.

# Materials and Methods

12 patients from January 2010 to 2014, age ranging from 7 months to 22 years with 5 males and 6 females with clinical suspicion of Hemifacial microsomia, referred to Radiology department for CT scan were included in the study.

In this study CT scans were performed at 320 multi-detector CT scanner (Aquilion ONE™ 320 Slice CT) using a single rotation volumetric technique covering the entire face and maxillofacial area. No IV contrast was required. A 0.5-mm reconstruction on bone and soft tissue algorithm was done and data transferred to work station (Kodak caresteram) for imaging post-processing and analysis. Images were reconstructed and analyzed by a radiologist with experience of more than 10 years. The interpretation of datasets was done on workstation ( KODAK CARESTREAM), using axial sections as well as reconstruction algorithms, including maximum intensity projection (MIP), multi-plannar reconstruction (MPR) and volume rendering (VR). **CT parameters were as follows: (320 _ 0.5 mm detector collimation; 0.5 mm reconstruction thickness; 120 kVp tube voltage);**

In this article we describe the diverse CT features of hemifacial microsomia using the format of OMENS and modified OMENS clinical classification systems. This will help Radiologist to understand the complex clinico-radiological manifestations of hemifacial microsomia.

# Results

The mandibular abnormality, the main pathology in the hemifacial microsomias (HM) was observed in 83 % of patients, out of which 60% had mild, 20 % moderate and 20% had severe deformity. This was followed by soft tissue deficiency in 75% of patients in which 44% have mild, 22% of moderate and another 33% with severe degree of deformity. Macrostomia was seen in 66 % of patients, mostly (87%) having milder degree of abnormality. The auricular deformity was observed in 58% of cases with majority (71%) having moderate degree of malformation. The orbital involvement was present in 33% cases ranging from mild orbital asymmetry to dytopia. (Tab. 1)

| SR # | Orbit | Mand-ible | Ear Defor-mity | Nerve Dysfu-nction | Soft Tissue | Miscel-laneous | Macro-stomia | Omens | Omens Modi-fied |
|------|-------|-----------|----------------|--------------------|-------------|----------------|--------------|-------|-----------------|
| Case 1 | O-1 | M-1 | E-2 | | S-2 | | C-1 RIGHT MACR | | O1-M1-E2-S2, C1 |
| Case 2 | O-1 | M-3 | E-3 | | S-3 | | C-1 LEFT MACR | | O1-M3-E3-N0-S3-C1 |
| Case 3 | O-0 | M-0 | E-0 | | S-0 | | C-0 | | O0-M0-E0-N0-S0-C0 |
| Case 4 | O-0 | M-2A | E-2A | | S-1 | | C-1 RIGHT MACR | | O0-M2A-E2A-N0-S1-C1 |
| Case 5 | O-1 | M-1 | E-0 | | S-1 | | C-0 | | O1-M1-E0-N0-S1-C0 |
| Case 6 | O-0 | M-1 | E-0 | | S-1 | | C-0 | | O0-M1-E0-N0-S1-C0 |
| Case 7 | O-0 | M-1 | E-0 | | S-0 | | C-2 | | O0-M1-E0-N0-S0-C2 |
| Case 8 | O-2 | M-2B | RT E-2, LT E1 | | S-2 | | C-1 LEFT MACR | | O2-M2B-E2-N0-S2-C1 |
| Case 9 | O-0 | M-0 | E-2A | | S-0 | | C-0 | | O0-M0-E2A-N0-S0-C0 |
| Case 10 | O-0 | M-1 | E-0 | | S-3 | | C-1 RIGHT MACR | | O0-M1-E0-N0-S3-C1 |
| Case 11 | O-0 | M-1 | E-1 | | S-1 | | C-1 Right | | O0-M1-E1-N0-S1-C1 |
| Case 12 | O-0 | M-3 | E-2 | | S-3 | | C-1 LEFT | | O0-M3-E2-S3-C1 |

**Table 1:** Simplified tabulated results according to OMENS modified pattern.

# Discussion

In 1881, Hemifacial Microsomia (HM) was first described by Carl Ferdinand Von Arlt. HM involves first and second branchial arch derivatives with highly variable phenotypes. It is also known as first and second branchial arch syndrome, otomandibular-facial dysmorphogenesis and lateral facial dysplasia. HM is primarily a syndrome of the first branchial arch, which involves underdevelopment of the temporo-mandibular joint, mandibular ramus, muscles of mastication and the ear. Abnormal development of the auricular hillocks leads to microtia or atresia of the pinna and it is proportional in severity to the abnormal external auditory canal development.

The OMENS classification is the most comprehensive one and, therefore, it is one of the most commonly used systems. In 1991 the Vento and colleagues first[12] described this system in their elegant study of 154 hemifacial microsomia patients and created a spectrum of five different anatomical manifestations and described the system using different grades and further sub stratification. Later in 1995 there was an addition of the acronym OMENS + **(Appendix I & II),** in which extra craniofacial anomalies were recognized. Additional refinement was done in the orbital category which centered on the orbital dytopia by Cousley and Calvert.[13] There was suggestion by Cousley[14] which calls for addition of middle ear and preauricular defects in the classification. Recently in a study by Bartlett et al[15] in 2006, suggested the pictographic representation of the spectrum of disease in the OMENS classification system by adding macrostomia (Teisser cleft # 7) in the classification system.

Using this pictographic approach we studied and reviewed CT scans of our patients baring their clinical knowledge provided by our plastic surgery team. This provided us with an excellent approach towards the spectrum of disease itself identifying the key abnormalities and grading them according to modified OMENS (+) classification system.

Although the numbers of patients in our study were less, a full disease spectrum was observed ranging from mild facial asymmetry to the severest form and Goldenhar Syndrome, a variant of HM. Mandibular abnormality (Fig. 2A, B, C) remains the chief pathology and is recognized in 81 % of patients which is nearly in concordance with the international literature (89% to 100%).[13,14] We further sub-stratified the abnormality in to mild, moderate and severe category according to modified OMENS classification system. The auricular deformity (Fig. 3A, B) was noted in 54 % of patients which is slightly less than 66% to 99%,[13] known to literature. Most of our patients present with unilateral deformity however there was one patient

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**Orbit**
  O0 Normal orbital size and position
  O1 Abnormal orbital size
  O2 Abnormal orbital position (arrow up or down)
  O3 Abnormal orbital size and position
**Mandible**
  M0 Normal mandible
  M1 The mandible and glenoid fossa are small.
  M2A Short ramus, glenoid fossa is in anatomically acceptable position
  M2B Short ramus, TMJ is inferiorly, medially and anteriorly displaced with hypoplastic condyle
  M3 Complete absence of ramus, glenoid fossa and TMJ
**Ear**
  E0 Normal ear
  E1 Mild hypoplasia & cupping, all structures present

E2 Absence of external auditory canal with hypoplasia of concha
E3 Malpositioned lobule with absent auricle, lobular remnant inferiorly and anteriorly displaced
**Facial Nerve**
  N0 No facial nerve involvement
  N1 Upper facial nerve involvement (temporal zygomatic)
  N2 Lower facial nerve involvement (buccal, mandibular, cervical)
  N3 All branches of facial nerve affected
**Soft Tissue**
  S0 No obvious soft tissue or muscle deficiency
  S1 Minimal subcutaneous/muscle deficiency
  S2 Moderate–between the two extremes S1 and S3
  S3 Severe soft tissue deficiency due to subcutaneous and muscular hypoplasia

**Appendix 1:** The omens classification system.

# Modified O.M.E.N.S. (+) Classification of Hemifacial Microsomia[1]

## (check all that apply)

**Side** (Each side evaluated separately in cases of bifacial microsomia)
__ R __ L

## Orbit[2]



__ **O0** Normal orbital size and position __ **O1** Abnormal orbital size __ **O2↓** Inferior orbital displacement __ **O2↑** Superior orbital displacement __ **O3** Abnormal orbital size and position
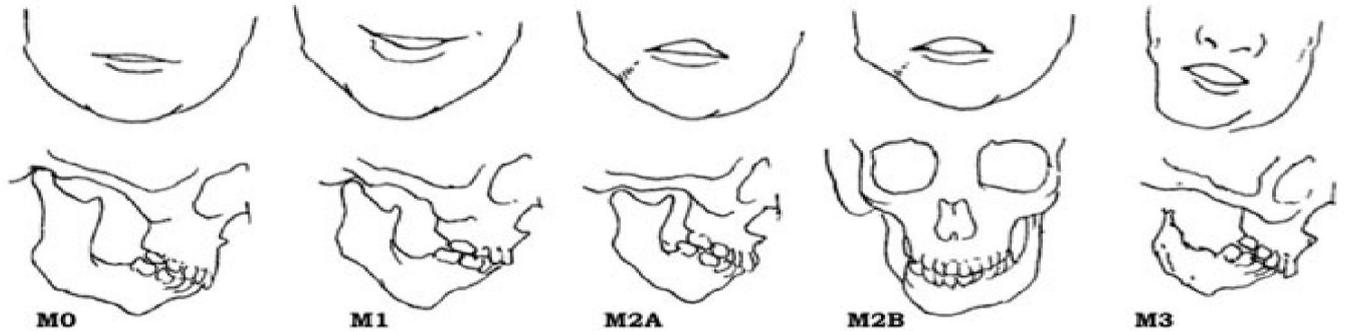
## Mandible



__ **M0** Normal mandible __ **M1** Small mandible and glenoid fossa with short ramus __ **M2A** Abnormally shaped and short ramus[3] (Glenoid fossa in acceptable position with reference to contralateral TMJ) __ **M2B** Abnormally shaped and short ramus (Glenoid fossa is inferiorly, medially and anteriorly displaced with a severely hypoplastic condyle) __ **M3** Absence of ramus and glenoid fossa (no TMJ)

## Ear



__ **E0** Normal auricle __ **E1** Mild hypoplasia and cupping with presence of all structures __ **E2** Absence of external canal with variable hypoplasia of concha __ **E3** Malpositioned lobule with absent auricle; lobular remnant typically inferiorly and anteriorly displaced

1. As modified from Vento AR, LaBrie RA. Mulliken JB. The O.M.E.N.S. classification of hemifacial microsomia Cleft Palate-Craniofacial Journal. 28(1):68-76.
2. Orbital position determined as follows; Midsagittal plane defined as vertical line between crista galli and anterior nasal spine. Horizontal line is then drawn perpendicular to midsagittal plane, tangent to supraorbital rims.
3. Evaluated with submental vertex radiographs or CT.

## Nerve
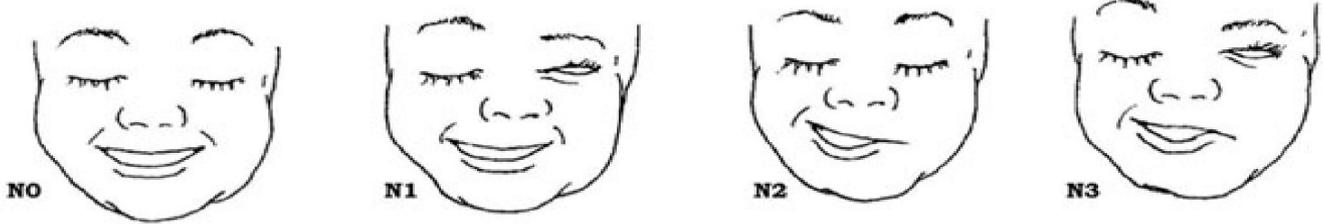


__ **N0** No facial nerve involvement __ **N1** Temporal and or Zygomatic branch involvement __ **N2** Buccal and/or Mandibular and/or Cervical branch involvement __ **N3** All branches affected

## Soft Tissue
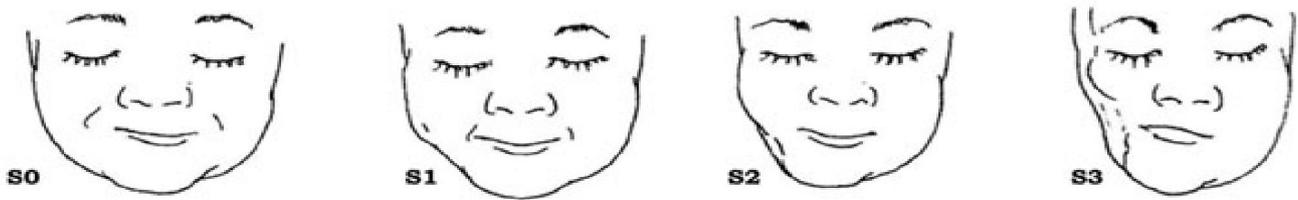


__ **S0** No soft tissue deficiency __ **S1** Minimal soft tissue deficiency __ **S2** Moderate soft tissue deficiency (between S1 and S3) __ **S3** Severe soft tissue deficiency

## Macrostomia (Tessier # 7 Cleft)
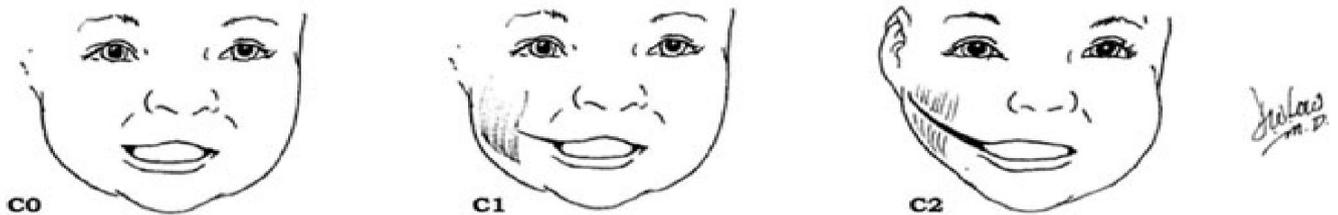


__ **C0** No cleft __ **C1** Cleft terminates medial to anterior border of masseter __ **C2** Cleft terminates lateral to anterior border of masseter

## Miscellaneous

O.M.E.N.S. (+)[4] { __ Yes __ No

__ **Goldenhars** (Hemifacial Microsomia with epibulbar lipodermoids and fused/hemivertebrae)

Notes:

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4. Horgan, J.E., Padwa , B.L., LaBrie, R.A., et al. OMENS-plus: analysis of craniofacial and extracraniofacial anomalies in hemifacial microsomia. *Cleft Palate Craniofac. J.* 32:405, 1995.

**Appendix II:** Modified O.M.E.N.S. (+) Classification of Hemifacial Microsomia
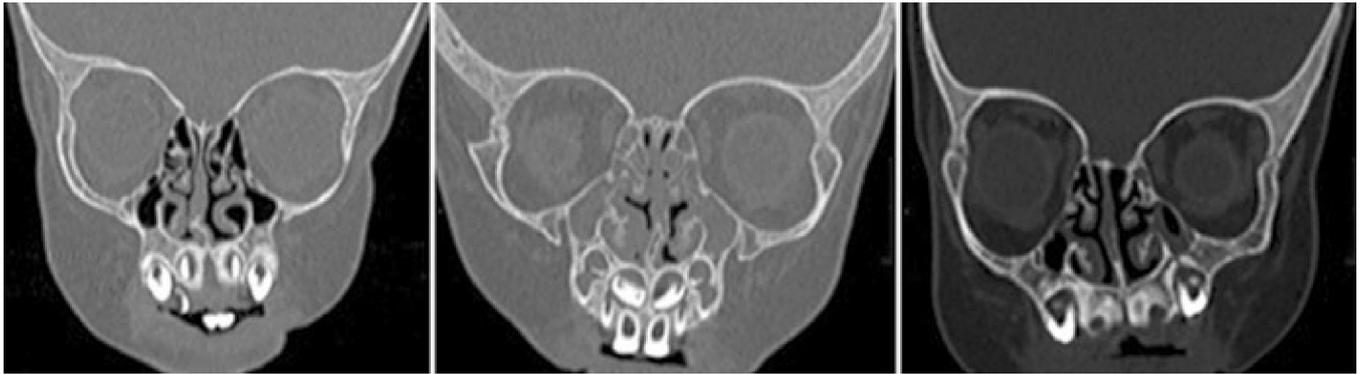
Figure 1: Various Orbital anomalies. (Left) Right O-2; (Centre) Right O-1; (Right) Left O-1.



**Figure 2A:** Various Mandibular Involvements: (Left) Right M1 - slightly small mandible and glenoid fossa, (Centre) 3D Recon image of same patient showing normal left mandible, (Right) 3D Recon image of same patient showing right M1 deformity.
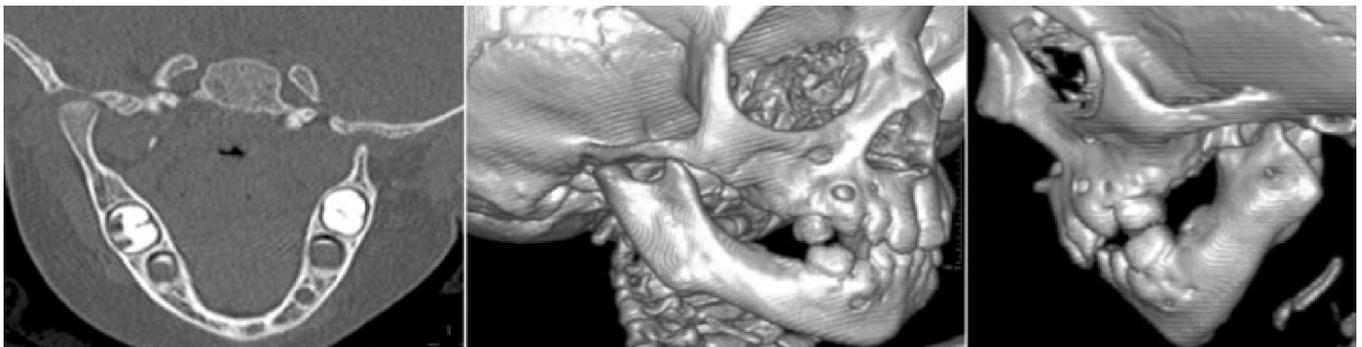


**Figure 2B:** (Left) Left M2-B --Short ramus, temporo-mandibular joint (TMJ) is inferiorly, medially and anteriorly displaced with hypoplastic condyle; (Centre) 3D Recon image of same patient showing normal right mandible; (Right) 3D Recon image of same patient showing left M2-B deformity.
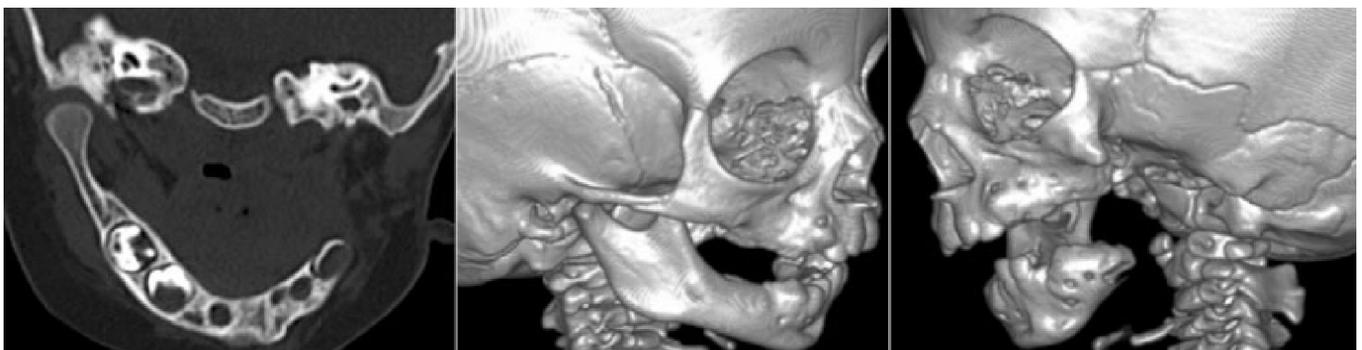


**Figure 2C:** (Left) Left M3--Complete absence of ramus, glenoid fossa and TMJ; (Centre) 3D Recon image of same patient showing normal right mandible; (Right) 3D Recon image of same patient showing left M3 deformity.

**Figure 3A:** Various Auricular Involvements: (Left) Absent left external acoustic canal (EAC);
(Centre) Same patient Left E3-- Malpositioned lobule with absent auricle, lobular remnant inferiorly and anteriorly displaced,
Left C1 macrostomia; (Right) Same patient normal right ear.
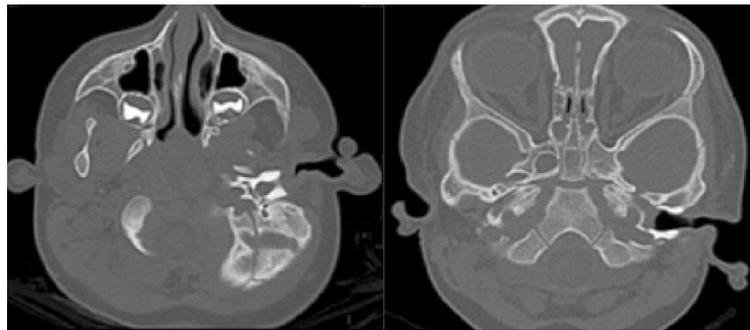


**Figure 3B:** (Left) Right E2--Absent right external acoustic canal (EAC), severe hypoplasia of concha  Left E1--mild hypoplasia and
cupping of left auricle and mild hypoplasia of EAC; (Right) Right E2—absent right EAC and conchal hypoplasia.

who had bilateral abnormality. The macrostomia (Fig. 5A, B), not described in earlier OMENS system, was present in 63 % of cases which is significantly higher compared to the 23-35%,[13,14] mentioned in earlier studies. The soft tissue deficiency (Fig. 4) was noted in a significant (72%) number of patients with involvement of muscles of mastication in all the cases. It

was observed that a slight-mild degree of soft tissue abnormalities were difficult to ascertain clinically, but with CT using soft tissue algorithm these were evident. Furthermore, with 3D volume rendered technique, a CT pictographic image was produced which helped us to visibly delineate & improved the understanding of clinical facial asymmetry. Similarly the subtle orbital



**Figure 4:** Various soft tissue abnormalities in our patients (CT scan soft tissue window).
(Left above) Moderate Soft tissue deficiency on left—S2:  (Right above) Minimal soft tissue deformity on left –S1
(Left below) Severe soft tissue deficiency on left—S3
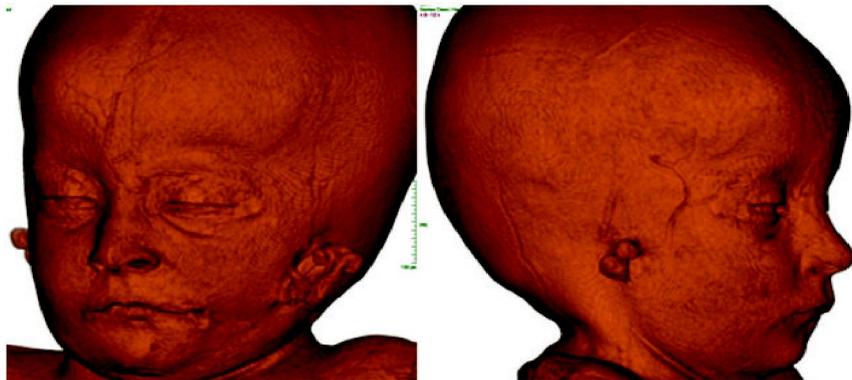
**Figure 5A:** Macrostomia presentation in our patient's 3D Pictographic representation: Left Macrostomia with bilateral preauricular skin tags and bilateral auricular deformities.
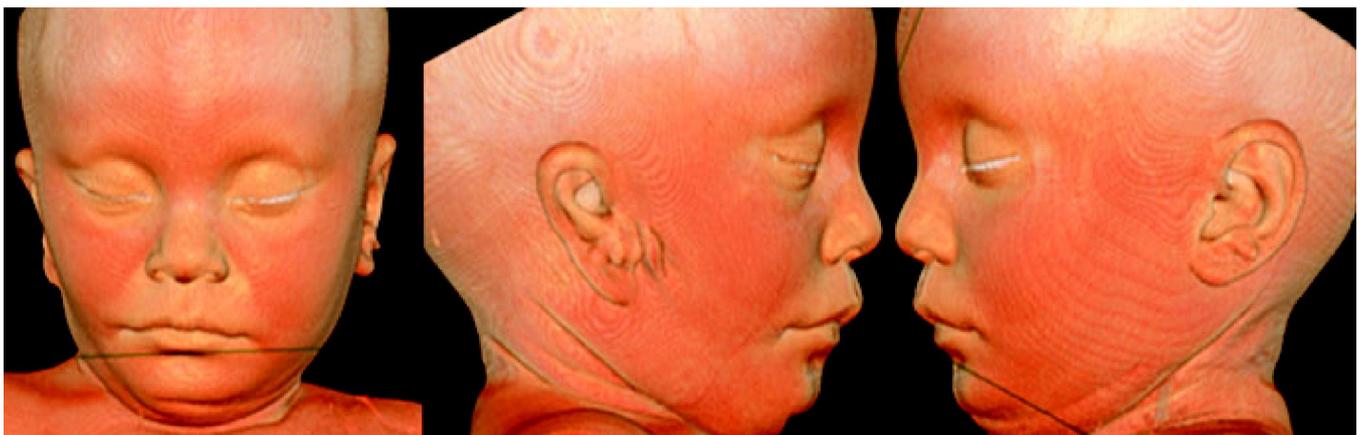


**Figure 5B:** (Left) Right C1 macrostomia; (Centre) Same patient right preauricular skin tag; (Right) Same patient normal left ear for comparison.

changes (Fig. 1) of shape and size, not clinically obvious, were also better visualized using bone window and multi planner reformats. In one patient, vertebral segmentation defects (Fig. 6) in cervical spine were noted, in addition to variety of OMENS deformities and macrostomia consistent with Goldenhar syndrome. A significant number of patients having mandibualr abnormality also show hypoplasia of other ipsilateral facial bones such as zygomatic arch, zygoma, pterygoids and maxilla. The preauricular skin tags were also present in many cases with aurciular abnormality. These findings have been described in the literature in various studies[13,14] in patients with HM but are not separately categorized in the OMENS/modified OMENS system. However the miscellaneous group in modified OMENS system can be used to designate these findings, though we in our study strict to using this title only for extra craniofacial anomalies, especially the vertebral.



**Figure 6:** Miscellaneous Findings: Vertebral defects C1, C2, C3 and decreased height of T4 with sclerosis.

## Conclusion

The hemifacial microsomia is a rare anomaly with fundamental features of unilateral hypoplasia of the craniofacial skeleton and its overlying soft tissue. The modified OMENS clinical classification system is so for the most accepted, versatile and comprehensive scheme for describing the distinct features of HM. Using the multi detector CT (MDCT) as imaging modality and incorporating the findings into the format of modified OMENS system, we were able to improve the clinical perception of the disease process and delineated the imaging findings in much better manner with lesser chance of error. We believe that this clinico-radiological rational i.e. integration of CT findings with clinical data, not only help to recognize the complex manifestations of disease itself as well as benefit the clinician in management of patients. We suggest similar and more comprehensive studies in this regard.

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