Clinics in Surgery

Short Communication
Published: 22 Jul, 2020

Craniofacial Microsomia: Clinical Features and Classification (Part I)

Bozkurt P* and Kisnisci R

Department of Oral and Maxillofacial Surgery, Ankara University, Turkey

Short Communication

Craniofacial Microsomia (CFM) is a facial congenital malformation, initially described in 1963 by Gorlin et al. It is one of the most widely encountered facial malformations following lip and palate clefts and indicates an extensive range of intricate soft and hard tissue malformations [1,2]. Assessing the exact prevalence of CFM is difficult, because of the lacking standard diagnostic criteria and data concerning ethnic and geographic variability, although it is likely to occur in every 1:3,500 live births to 1:20,000 live births. CFM more frequently affects the male gender, with an estimated female:male ratio of 2:3 [3-5]. The term CFM, is occasionally used interchangeably with the term Goldenhar syndrome (oculo-auriculo-vertebral dysplasia), which is a type of CFM described by Dr. Maurice Goldenhar in 1952 [6]. The term Goldenhar Syndrome is generally reserved for cases with internal organ and vertebrae involvement (malformations such as epibulbar dermoids, microphthalmia, coloboma, preauricular tragi, lower attachment of the auricle, hearing loss, scoliosis or hemivertebrae, mental retardation, congenital heart and renal defects) [6].

CFM is a Craniofacial Abnormality derived from the first and second branchial arches. It may present with unilateral or in 10% to 15% of cases a bilateral involvement of the orbits, jaws, ears, cranial nerves and associated soft tissues [1,4,7,8]. Cases of CFM are non-uniform, but mandibular hypoplasia is frequently the most noticeable malformation and occurs in 89% to 100% of affected patients [3].

The exact etiologic factors behind this malformation yet continue to be unexplored. Current evidence supports three etiological theories taking place during fetal development, which lead to compromised facial growth [1]; failure in the proliferation and migration of embryonic neural crest cells, [2] injury of the Meckel's cartilage and [3] hemorrhage of the stapedial artery causing a hematoma in the supplied region. These 3 theories can be considered to be interrelated, since none of the theories alone can completely meet the requirements of all the variable manifestations of CFM. Genetic factors, maternal intrinsic factors and external environmental factors, may contribute to CFM through ≥ 1% of these pathogenic processes [9].

Still, the most widely considered theory is the hemorrhage model [7,9]. The first and second branchial arches are supplied by the stapedial artery which is supplied by the ventral pharyngeal and hyoid arteries. This fetal artery system is substituted by the external carotid arterial system after birth. In the hemorrhage model, a hematoma is formed around the stapedial artery subsequent to a hemorrhage, leading to an insufficiency of blood supply, hypoxia of the tissue, and finally in impairment and malformation of the surrounding tissues [9].

Clinical Features

The first and second branchial arches, are formed by cranial neural crest cells deriving from the dorsal part of the neural tube, and contribute to most hard and soft craniofacial structures. The maxilla, zygoma, mandible, malleus, and incus, and the trigeminal nerve are derived from the cranial neural crest cells of the first branchial arch, while the stapes and facial nerve are derived from the cells of the second branchial arch. The cephalic myogenic mesodermal cells, in the branchial arches, are responsible for the musculature of the head. The masseter, temporal, and the pterygoid muscles are derived from the cephalic myogenic mesodermal cells of the first branchial arch and the facial expression (mimetic) muscles, the digastric venter posterior and stylohyoid muscles, the auricular and stapedius muscles are derived from the cells from second branchial arch [9]. All of these structures originating from the first and second branchial arches are the affected structures of the craniofacial region in patients affected with CFM. Facial clefts, macrostomia, retrognathism, craniosynostosis, external auditory canal atresia, preauricular tags, varying degrees of deafness,

OPEN ACCESS

Correspondence:
Bozkurt P, Department of Oral and Maxillofacial Surgery, Ankara University, Turkey,
E-mail: poyzanbozkurt@hotmail.com

Received Date: *01 Jun 2020*
Accepted Date: *02 Jul 2020*
Published Date: *22 Jul 2020*

Citation:
Bozkurt P, Kisnisci R. Craniofacial Microsomia: Clinical Features and Classification (Part I). Clin Surg. 2020; 5: 2870.

Copyright *© 2020 Bozkurt P. This is an open access article distributed under the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.*

Table 1: The OMENS-Plus classification system.

Affected Tissue	Degree	Presentation
Orbit	O0	Normal orbital size and position
	O1	Abnormal orbital size
	O2	Abnormal orbital position
	O3	Abnormal orbital size and position
Mandible	M0	Normal mandible
	M1	Normal mandible and glenoid fossa with a short ramus
	M2a	Short and abnormally shaped ramus: the glenoid fossa is in anatomically acceptable position with reference to opposite TMJ
	M2b	Short and abnormally shaped ramus: displaced TMJ with severely hypoplastic condyle
	M3	Complete absence of ramus, glenoid fossa and TMJ
Ear	E0	Normal ear
	E1	Mild hypoplasia and cupping ear with all structures present
	E2	Absence of external auditory canal with variable hypoplasia of the concha
	E3	Mal positioned lobule with absent auricle; lobular remnant usually inferiorly and anteriorly displaced
Facial Nerve	N70	No facial nerve involvement
	N71	Facial nerve involvement (upper temporal and zygomatic branches)
	N72	Facial nerve involvement (lower buccal, mandibular and cervical branches)
	N73	Facial nerve affected (all branches)
Soft Tissue	S0	No obvious deficiency of the soft tissue or muscle
	S1	Minimal deficiency of the subcutaneous/muscle tissue
	S2	Moderate deficiency of the soft tissue or muscle
	S3	Severe deficiency of the soft tissue or muscle
Plus		Systemic malformations

epibulbar dermoids, facial nerve palsies and extracraniofacial anomalies, including Fallot tetralogy, osteochondromatosis, horseshoe kidneys, lambdoidal synostosis, and scoliosis are some of the features observed in patients affected with CFM [1]. Though there is a large number of cranial deformities which can be observed in CFM, it has been reported that there existed little difference in endocranial morphologic measurements with increasing severity of CFM [10].

CFM is a clinical diagnosis and brings not only aesthetic complications but also functional problems and limitations. Some of these functional problems are hearing impairment related to ear involvement, obstructive sleep apnea, and feeding difficulty related to mandibular hypoplasia [11].

Feeding difficulty has been documented in some reports to be as high as 83% in patients suffering from CFM and consists of suckling and chewing problems, incoordination of deglutition, dysphagia, and related inability to thrive. Patients with bilateral CFM, a Pruzansky-Kaban III mandibular deformity (to be explained under the next title), obstructive sleep apnea, lip and/or palate clefts, and extra-craniofacial anomalies are significantly at higher risk for developing feeding difficulties. Patients with these risk factors more often reported to require supplementary feeding, for example *via* a nasogastric tube [11].

Bilateral hypoplasia of the mandible can be observed in numerous facial malformations including the Pierre Robin sequence, Treacher Collins syndrome and also CFM. This condition may be related to obstructive sleep-disordered breathing and obstructive sleep apnea. The description of obstructive sleep-impaired breathing may be explained by increased upper airway resistance and pharyngeal collapse, leading to upper respiratory tract dysfunction during sleep, presenting with a range of clinical entities from primary snoring and increased effort of inhalation to obstructive sleep apnea. Obstructive sleep apnea is also characterized by snoring, increased effort of inhalation throughout the sleeping period, and additional incidents of complete or partial airway obstruction. As obstructive sleep apnea can be associated with neurocognitive, metabolic, and cardiovascular concerns, defining groups with higher risk, like CFM, is important. The prevalence of obstructive sleep apnea is reported to be between 2.2% to 3.8% in lean children without facial deformities, though in children with mandibular hypoplasia, higher prevalence is reported. The prevalence of obstructive sleep apnea in patients with bilateral and unilateral CFM is widespread between 7% to 67% and patients with more severe involvement of the orbit and mandibula and/or patients with bilateral CFM are at higher risk of presenting with obstructive sleep apnea. Patients who have extracranial anomalies are also suspected to be or diagnosed with obstructive sleep apnea more often [8].

Social, linguistic and cognitive development, and dependant on these, literacy and academic success, are all highly associated with hearing status. For infants with hearing loss, early identification and intervention creates more optimal results of development [4].

External ear malformations, external auditory canal atresia, and Eustachian tube dysfunction are the common causes of hearing loss in patients with CFM, though microtia and external auditory canal atresia has been described as primary reasons for hearing loss. Patients with CFM have been reported to have abnormalities of the inner ear at variable levels, and patients with these abnormalities are at greater

risk of developing hearing loss [4]. The involvement of the mandible in the CFM patient is reported to be associated with an increase in the likelihood of hearing loss on the affected side. Nevertheless, other reports also reported a high rate of hearing loss in the side where facial anomalies are absent. The average prevalence of hearing loss in patients with CFM requires that all affected individuals undergo bilateral hearing evaluation as soon as possible regardless of the severity of disease [4].

Classification Systems

Classification of CFM deformities can provide clarity among medical professionals and in determining the ideal treatment plan and accurate prognosis of the patient [7]. Clinical phenotypes of CFM are usually easily distinguished from other craniofacial abnormalities, however because of variable degree of deformities, still there is no common opinion for clinical identification [1]. Nevertheless, while classifying the disease, it is a common idea that the main features should be abnormalities of the mandible and ear because of their high incidence rate and the positive association between mandibular hypoplasia and other involvements [1,12].

To assess the abnormalities of the orbit, mandible, ear, nerve and soft tissue, with variable clinical presentations, the OMENS classifications system has also been introduced by Vento et al. in 1991 [12]. The OMENS classification system, layers each anatomic manifestation of CFM on a scale from 0 to 3 (orbits, mandible, ears, facial nerve, soft tissue) according to severity. The classification of the orbit is graded according to its size and position, the mandibula is graded according to the Pruzansky classification (to be described), the external ear is graded according to size and the position of the auricle and external auditory canal, the involvement of the facial nerve is graded by the associated nerve section and the soft tissue is graded exclusively by subcutaneous and muscular deficiencies [12].

The OMENS system was later advanced to the OMENS-Plus classification (Table 1). Widening the OMENS system into OMENS-plus by adding indicators of extra-craniofacial anomalies (central nervous system, cardiac, pulmonary, renal, gastrointestinal, and vertebral deformities), has created a more comprehensive classification system and has majorly advanced the classification of CFM [1].

The Munro and Lauritzen classification system based on clinical findings, classifies CFM patients as IA: A horizontal occlusal plane with only mild hypoplasia of the craniofacial skeleton; IB: A cantered occlusal plane with only mild hypoplasia of the craniofacial skeleton; II: Absence of the condyle and part of the ramus; III: Absence of the condyle, part of the ramus, the zygomatic arch and glenoid fossa; IV: Hypoplasia of the zygoma and medial-posterior displacement of the lateral orbit wall; V: Inferior displacement of the orbit and decreased orbital volume.

Kaban's modification of the Pruzansky CFM classification is based on the presence or absence of the abnormality of the mandible and associated structures. Today, this classification system provides a useful guide for clinical diagnosis and treatment planning [7].

According to the Kaban's modification of the Pruzansky CFM classification; CFM type I deformity patient can be explained as a generalized mild hypoplasia of the glenoid fossa, mandibular condyle, ramus and muscles of mastication. The patient presents with mild mandibular retrognathia and facial asymmetry which leads to mild aesthetic problems. The temporomandibular joint can function normally with a normal rotation although the translation movement is restricted, and there is satisfactory temporomandibular joint/masticatory function. Surgical treatment is generally not considered for the CFM type I patient. The CFM type IIA deformity patient can be explained as a hypoplastic conical condylar head and hypoplastic glenoid fossa. The condylar head is placed anteriorly and medially in the glenoid fossa. Again, there is satisfactory temporomandibular joint/masticatory function and surgical treatment is generally not considered for the CFM type IIA patient. The CFM type IIB deformity patient can be explained as a moderate to severe hypoplasia of the glenoid fossa, mandibular condyle, and ramus. The CFM type IIB deformity patient is differentiated from the CFM type IIA deformity patient by the absence of articulation between the temporal bone and the condyle. Nevertheless, during manual maneuvering, a "stop" point is felt in the posterior, as condylar head touches the glenoid fossa. The choice of treatment is variable for CFM type IIB deformity patients. The CFM type III deformity patient can be explained as agenesis of the mandibular condyle and ramus, therefore manual manipulation cannot reveal a condylar seating or posterior "stop". Patients do not have satisfactory temporomandibular joint/masticatory function and aesthetic problems and generally require surgical treatment [7].

Today, 3-Dimensional Computed Tomography (3D-CT) scans are an all-important part of medical practice and have also been suggested in evaluating CFM. The use of 3D-CT not only can evaluate bony abnormalities but their relationship with the surrounding underdeveloped soft tissues including masticatory muscles and allows a more complete and objective classification for CFM patients, apart from classification system used [1].

Although currently tending to show inter-rater and intra-rater variability in evaluation, the Pruzansky-Kaban modification and OMENS classifications systems are up-to-date the basis of newly developed classification systems for characterizing facial features of CFM [3].

References

1. Ji Ung P, Hai T, Yong KG, Hyun CT. Statistical analysis using the OMENS classification in Oriental patients with hemifacial microsomia: A comparative analysis with Western centers. Ann Plast Surg. 2014;72(1):50-55.
2. Wang P, Wang Y, Zhang Z, Li X, Ye B, Li J, et al. Comprehensive consideration and design with the virtual surgical planning-assisted treatment for hemifacial microsomia in adult patients. J Craniomaxillofac Surg. 2018;46(8):1268-74.
3. Kaya O, Pluijmers BI, Staal F, Ruff C, Padwa PL, Koudstaal MJ, et al. Describing the mandible in patients with craniofacial microsomia based on principal component analysis and thin plate spline video analysis. Int J Oral Maxillofac Surg. 2019;48(3):302-8.
4. Mitchell RM, Saltzman BS, Norton SJ, Harrison RG, Heike CL, Luquetti DV, et al. Hearing loss in children with craniofacial microsomia. Cleft Palate Craniofac J. 2017;54(6):656-63.
5. Cassi D, Magnifico M, Gandolfinini M, Kasa I, Mauro G, Blasio AD, et al. Early Orthopaedic Treatment of Hemifacial Microsomia. Case Rep Dent. 2017.
6. Speranța S, Burcel M, Dăscălescu D, Popteanu LC. Goldenhar Syndrome-ophthalmologist's perspective. Rom J ophthalmolo. 2018;62(2):96-104.
7. Caron CJJM, Pluijmers BI, Maas BDPJ, Klazen YP, Katz ES, Abel F, et al. Obstructive sleep apnoea in craniofacial microsomia: Analysis of 755 patients. Int J Oral Maxillofac Surg. 2017;46(10):1330-7.

8. Chen Q, Zhao Y, Shen G, Dai J. Etiology and pathogenesis of hemifacial microsomia. J Den Res. 2018;97(12):1297-305.

9. Paliga JT, Tahiri Y, Wink J, Bartlett JP, Taylor JA. Cranial base deviation in hemifacial microsomia by craniometric analysis. J Craniofac Surg. 2015;26(1):e61-4.

10. Caron CJJM, Pluijmers BI, Joostena KFM, Dunaway D, Padwa LB, Wolvius EB, et al. Feeding difficulties in craniofacial microsomia: A multicenter retrospective analysis of 755 patients. J Craniomaxillofac Surg. 2018;46(10):1777-82.

11. Heike CL, Wallace E, Speltz ML, Siebold B, Werler MW, Hing AV, et al. Characterizing facial features in individuals with craniofacial microsomia: A systematic approach for clinical research. Birth Defects Res A Clin Mol Teratol. 2016;106(11):915-26.

12. Birgfeld CB, Heike CL, Saltzman BS, Leroux BG, Evans KN, Luquetti DV, et al. Reliable classification of facial phenotypic variation in craniofacial microsomia: A comparison of physical exam and photographs. Head Face Med. 2016;12(1):14.