CASE REPORT

Carniofacial Microsomia

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Abstract

Cranoifacial microsomia is an unique clinical presentation of '1st and 2nd arch syndrome' with asymmetrical craniofacial development alongwith conductive hearing loss. A series of 11 patients (4 males, 7 females) is presented which include two patients of 'Goldenhar's variant' with epibulbar dermoids. Two patients had no response on pure tone audiometry (blank audiograms) and underwent CT scan of temporal bone which revealed 'Michel's aplasia' (complete labyrinthine agenesis), rarely reported in the literature.

Keywords

Cranio-facial, Microsomia

Introduction

Craniofacial microsomia was first described by Arlt in 1881. Gorlin *et al.*(1) used the term hemifacial microsomia to these patient with unilateral microtia, macrostomia and malformation of mandibular ramus and condyle whereas, the Goldenhar's syndrome was described as a varient, with vertebral anomalies and epibulbar dermoids. The name, craniofacial microsomia, was proposed by Converse *et al.*(2) when cranial deformities were included. Other synonyms include first arch syndrome, first and second bronchial arch syndrome, otomandibular dysostosis, oculo-auriculovertebral dysplasia and lateral facial dysplasia (3).

This disorder may exhibit wide phenotypic heterogenicity and several classification systems, namely SAT (skeletal, auricular and soft tissue categories) (4) and OMENS classification (orbit, mandible, ear, nerve and soft tissue categories) (5) have been devised to aid diagnosis and treatment planning.

Case Series

We present a series of 11 patients (4 males and 7 females) of craniofacial microsomia between 3 months to 25 years of age, the clinical profile of whom is given in Table 1. Four patients had bilateral asymmetrical presentation, 3 had left side involvement and rest 4 patients had right-sided craniofacial microsomia (Fig. 1). All patients underwent hearing assessment, 7 of whom co-operated for pure tone audiometry (PTA) which revealed ipsilateral conductive hearing loss in 5 patients ranging from moderate to severe degree (> 40dB to 90dB). Two of these seven patients had profound sensorineural hearing loss with no response at any frequency with the maximum possible stimulus (blank audiogram). High resolution CT scan of temporal bone of these two patients revealed 'Michel's aplasia' (Fig. 2), complete agenesis of labyrinth, a finding rarely reported in the literature.

In rest of four patients, free field testing (FFT) and subsequently brainstem evoked response audiometry (BERA) was performed. Two patients had severe hearing loss on BERA, one patient had severe deafness on FFT with inconsistent wave formation on BERA, and fourth patient had inconsistent FFT but mild to moderate deafness on ipsilateral side on BERA.

In total, CT scan of temporal bone of 4 patients with bilateral asymmetrical craniofacial microsomia was undertaken. Two patients with bilateral conductive hearing loss revealed bilateral external auditory canal atresia with hypoplastic middle ear cavities and normal inner ear structures. Rest of two patients were of Michel's aplasia as discussed above.

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Table 1. Clinical profile of 11 patients of craniofacial microsomia.

S. No.	Sex / Age	Clinical profile	Hearing loss	CT scan temporal region
1.	F / 10 yr.	B/L asymmetrical	B/L Profound (> 90dB) SNHL	Bilateral Michel's aplasia
2.	F / 8 yr.	B/L asymmetrical	B/L Profound (> 90dB) SNHL	Bilateral Michel's aplasia
3.	F / 1 yr.	*Left craniofacial involvement	Lt. Severe (BERA)	-
4.	F / 9 yr.	Right craniofacial involvement	Rt. 70dB, severe	-
5.	M / 6 mth.	Right side	Rt. Severe (BERA)	-
6.	F / 18 yr.	*Right side	Rt. 65dB (Mod-severe)	-
7.	M / 8 yr.	B/L asymmetrical	Rt. 50dB, Lt. 35dB	B/L Ext. auditory canal atresia
8.	F / 3 mth.	Right side	Severe on FFT (BERA inconsistent)	-
9.	F / 20 yr.	B/L asymmetrical	B/L 60dB (Mod-severe)	B/L Ext. Bony canal atresia
10.	M / 25 yr.	Left side	Lt. 65dB (Mod-severe)	-
11.	M / 8 yr.	Left side	FFT inconsistent, BERA, mild-moderate Lt.	-

*Goldenhar's variant. BERA - Brainstem Evoked Response Audiometry. FFT - Free Field Testing. SNHL - Sensorineural Hearing Loss.

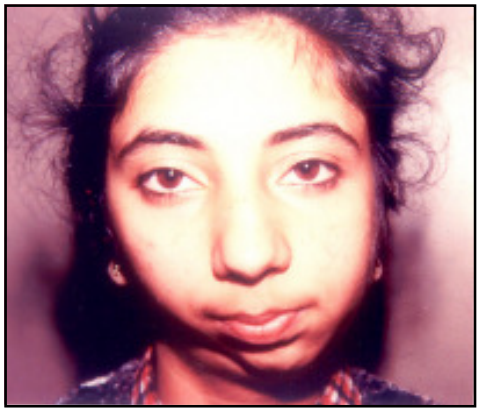

Fig. 1. Patient of craniofacial microsomia (right sided) exhibiting facial asymmetry, ipsilateral hypoplasia of mandibular ramus, macrostomia and microtia.

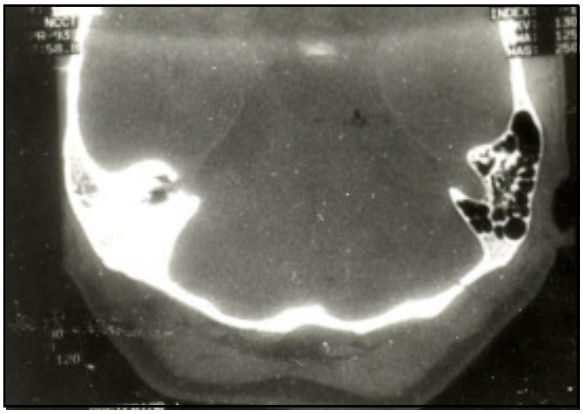

Fig. 2. High resolution CT scan of craniofacial microsomia with profound sensorineural hearing loss showing Michel's aplasia. Mastoid air cells are well pneumatized on left side and ossicles are visualized on right side, with total labyrinthine aplasia on both sides.

Discussion

Incidence of craniofacial microsomia is between 1:5000 and 1:5600 live birth with 1:1 sex ratio (6). It is usually unilateral (70%) and always asymmetrical if exhibits bilaterally. Embryologically, it is supposed to be due to destruction of differentiating tissues in the region of the developing ear and jaws, leading to branchial arch dysplasia, which in turn is multifactorial in origin (7).

Clinically, the facial appearance is characteristic, which is always asymmetrical, with hypoplastic maxilla, zygoma and temporal bone and deformed pinna on the involved side. Frontal bossing with ipsilateral low set eyes and parotid hypoplasia is usually seen along with minor degree of macrostomia. Approximately 30% of the patients have bilateral asymmetrical involvement. Micrognathia, if significant, can lead to sleep apnoea. Hypoplasia and / or paresis of palatal muscles and pharynx, alongwith tongue musculature is also reported in these patients. Cleft lip and palate is seen in approximately 10% of cases but velopharyngeal insufficiency is seen in 35% of cases. Encephalocoele mainly in occipital region along with mental retardation is seen in approximately 10% cases (3).

As observed by James and Baggs(3), the external ear malformations vary from complete aplasia to a crumpled, distorted pinna alongwith ear tags. 40% of cases have conductive hearing loss due to external and / or middle ear malformations. The other important finding includes epibulbar dermoid, located at the limbs or corneal margin in the lower and outer quadrant, and lipodermoid seen in upper and outer quadrant. Unilateral coloboma is usually seen in upper lid alongwith epibulbar dermoid. Microphthalmia and anophthemine are usually seen alongwith mental retardation (3, 8).

Approximately, 50% of patients have congenital heart disease (8). Also reported are imperforate anus, absent or ectopic kidney, vertebral anomalies and pulmonary hypoplasia (3,8).

Radiologically, the meatal atresia and middle ear anomalies are almost constant findings. Occasionally, hyperplasia of the external ear structures, especially the tympanic bone is seen, but the mastoid is always hypoplastic and unpneumatized. Middle ear cavity is small and ossicles may be absent, hypoplastic and/or malformed (9).

The differential diagnosis of the condition include Pierre Rubin Syndrome, Moebius syndrome, Tracheal-Collin's syndrome and frontonasal dysplasia. As regards the management of these conditions is concerned, there exists no general agreement for the best and appropriate time to carry out the reconstructive surgery. In general, the facial asymmetries are more difficult to correct than horizontal or vertical disproportions (3).

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