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Original Article

Congenital Anomalies Associated with microtia – Anotia: Review of 30 Cases

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Abstract

Background: *Microtia-anotia has a global prevalence of 2.6 per 10,000 live births. Children with microtia-anotia will have an associated anomaly or an identifiable syndrome pattern in 20–60% of cases.[1] The most common anomalies are facial cleft, facial asymmetry, renal abnormalities, cardiac defects, microphthalmia, polydactyly, and vertebral anomalies.*

Methods: *This series consists of retrospective study of 30 patients who presented to the department of ENT, KEM Hospital between January 2010 to June 2013. Case Records of patients with congenital external ear deformity who presented to the E.N.T. Department during the time period from January 2010 to June 2013 were reviewed for the grade of microtia. The patients were also evaluated for associated congenital anomalies.*

Results: *Associated anomalies in patients included facial paralysis, unilateral renal agenesis, congenital heart diseases, Cleft palate, microphthalmia, microcornea, iris coloboma, hemivertebra. Also Goldenhar syndrome / Hemifacial microsomia, Treacher- Collin syndrome and Pierre- Robin syndrome were the related syndromes to congenital external ear deformity in our series.*

Conclusion: *Congenital external deformities may be associated with spectrum of other anomalies and the patient should receive best coordinated care from otolaryngologists, audiologists, paediatricians, heart specialist to improve the quality of life. Early hearing assessment should be followed by proper rehabilitation for adequate speech and language development.*

Keywords: *microtia, congenital anomalies, syndromes, coordinated care, Baha.*

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Introduction

The external ear consists of the auricle, the external acoustic meatus and the tympanic membrane. Microtia-anotia is a spectrum of congenital anomalies of the auricle ranging from mild structural abnormalities to complete absence of the ear (anotia) with or without deformity of the external auditory canal.

It mostly occurs as part of a spectrum of anomalies or a syndrome and may rarely occur as an isolated condition. It forms a part of first or second arch syndrome. Individuals with unilateral microtia-anotia may have normal hearing in the unaffected ear or 60 dB hearing loss due to congenital fixation of stapes. Therefore, speech and language

development may be typically normal, although children with microtia-anotia are at a greater risk of delayed language development and may have attention deficit disorders. Children with microtia-anotia will have an associated anomaly or an identifiable syndrome pattern in 20–60% of cases.[1] The most common anomalies are facial cleft, facial asymmetry, renal abnormalities, cardiac defects, microphthalmia, polydactyly, and vertebral anomalies.[1]

The child born with a malformed ear faces a lifelong hearing and communication impairment along with the social stigma of a facial deformity. Associated disturbances of the vestibular system may add to the developmental hurdle of a motor delay. Frequently, there are additional anomalies, such as mandibular hypoplasia, as well as other facial and skeletal deformities. There may be dysfunction of associated neural pathways, including cranial nerves and intracranial structures. Additionally, there are psychological factors to be considered, including parental guilt, peer ridicule, and the shame of "being different".[2-4]

Methods

Our series consists of retrospective study of 30 patients who presented to the department of ENT, KEM Hospital between January 2010 to June 2013. Case Records of patients with congenital external ear deformity who presented to the E.N.T. Department during the time period from January 2010 to June 2013 were reviewed for the grade of microtia. The patients were also evaluated for associated congenital anomalies. Comprehensive care was given to these children and they were evaluated by our audiologist and speech pathologist, paediatrician, paediatric cardiologist, paediatric nephrologist, plastic surgeon, medical and social worker and counsellor. Congenital external ear deformities were graded according to Marx classification into type I, II, III microtia or anotia. Associated congenital anomalies as in cardiac, renal or craniofacial malformations were noted from the information provided in the medical data. Hearing assessment was done using audiogram and BC BERA to rule out sensorineural component. HRCT Temporal was done to look for bone thickness behind temporo parietal suture line. Hearing rehabilitation was done using either Baha (Bone anchored hearing aid) band or Baha implant.

Results

In our study congenital microtia- anotia was found to be more common in males with a ratio of 4:1. The most common presentation was observed to be bilateral congenital microtia-anotia. In unilateral cases right sided microtia-anotia was found to be more common. Male predilection is found in cases of congenital microtia- anotia with grade III microtia being most common external ear deformity.

Amongst 30 patients of congenital external ear deformity, facial palsy was present in 21% of cases of which bilateral facial paralysis was seen in two patients. Unilateral non-functioning kidney was present in 3% of cases while unilateral ectopic kidney was present in 3% cases. Unilateral renal agenesis was found in 6.7% of cases. Undescended testis was seen in 6.7% of patients. Congenital heart diseases were present in 18% of cases which included atrial septal defect in two patients, only ventricular septal defect in one of the patients and ventricular septal defect associated with atrial septal defect, tricuspid regurgitation and pulmonary hypertension in one of the patients. Patent ductus arteriosus was also present in one of the cases. One of the patients had patent foramen ovale with left to right shunt. Associated mild mitral regurgitation was found in one case. Submucus Cleft Palate was found in 3% cases. Cleft palate was present in 3% of cases. Associated tongue tie was present in 6.7% patients. Goldenhar syndrome / Hemifacial microsomia was present in 13.3% of cases. Treacher- Collin syndrome was also found in 6.7% of subjects. Pierre- Robin syndrome being present in 3% of cases and microphthalmia, microcornea, iris coloboma, hemivertebra was found in 3% of patients. Unilateral absence of tonsil in 6.7% of cases.

Post auricular tag was seen in 3% of cases. Post auricular sinus in 3% of cases. Also pre auricular sinus was found to be associated in 3% of the cases.

Discussion

Microtia-anotia has a global prevalence of 2.6 per 10,000 live births.[5] They usually arise as a result of genetic and environmental factors. Exposure to teratogens such as thalidomide is an important environmental factor in its occurrence.[13] However, in our series, none of the patients neither had such a risk factor nor had any positive family history. But, mostly the etiology is uncertain whether it presents in isolation or as a part of an identifiable syndrome. The most frequently associated syndromes, such as Treacher Collins, Goldenhar and Pierre- Robin syndromes, usually occur as a result of specific genetic anomaly but the significance of those genes during pinna development has not yet been understood. According to the recent researches, neural crest cell disturbance and vascular disruption contribute to the pathogenesis of external ear deformity. Microtia-anotia in 20-60% of cases may have an associated anomaly or an identifiable syndrome pattern.[1]

Our protocol constituted of evaluating every patient with microtia-anotia for the presence of associated anomalies. Ultrasound of the abdomen was done to rule out renal anomalies. Echocardiography was performed to look for the congenital heart diseases. This helped us to identify high risk cases and early intervention in the presence of associated anomaly.

The results in our study were similar to those in the literature as has been depicted in the following table.

Congenital anomalies	Gosta etal[6]	Daniella et al[7]	Van Neumann et al[8]	Lie Jin et al[9]	Wang et al[10]	Our results
Treacher Collins	32%					6%
Hemifacial Microsomia	27%		27.5%			13%
Mobius						
Crouzon						
Diabetic embroyopathy						
Facial Assymetry		10.6%	8.3%	20%		21%
Congenital heart defect		18.5%	2.5%	5%		18%
Renal anomalies		3.3%		5%	29%	10.3%
Micro-ophthalmia and other eye defects		11.5%		45%		3%
EAC atresia			76%			80%
Pre auricular skin tags			30.5%			
Lip hemangioma				5%		
Cervical scoliosis				10%		
Cleft lip and cleft palate				10%		6%
Undescended testis						3%
Tongue tie						6%
Pierre-Robin syndrome						3%
Unilateral absence of tonsil						3%
Pre auricular sinus						3%
Post auricular sinus						3%
Post auricular tag						3%

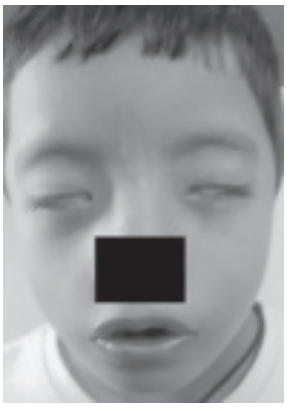

Fig.-1: *Child showing bilateral microtia with bilateral facial palsy*

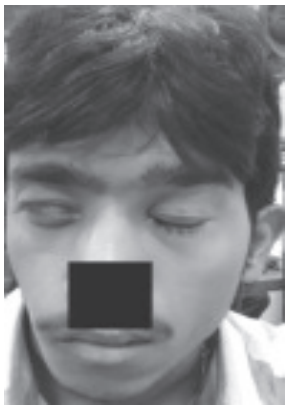

Fig.-2: *Patient with unilateral microtia with unilateral facial palsy*

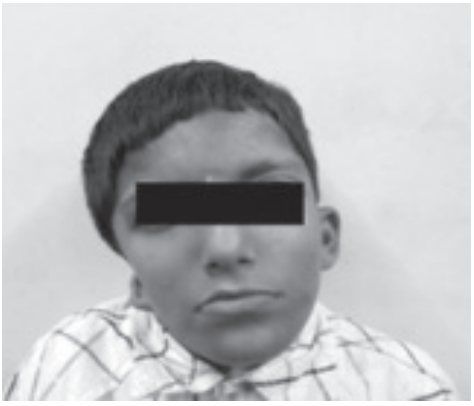

Fig.-3a: *Showing child with Goldenhar syndrome*

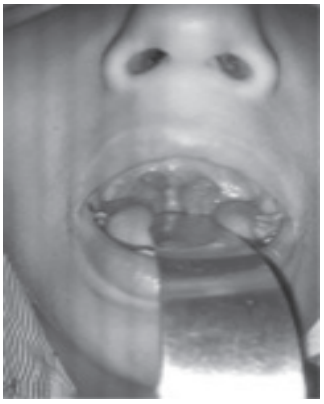

Fig.-3b: *Same patient with submucus cleft palate*

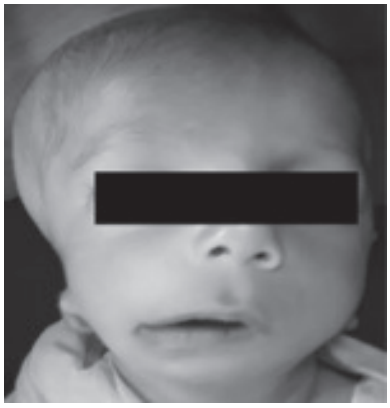

Fig.-4: *Child with Pierre- Robin syndrome*

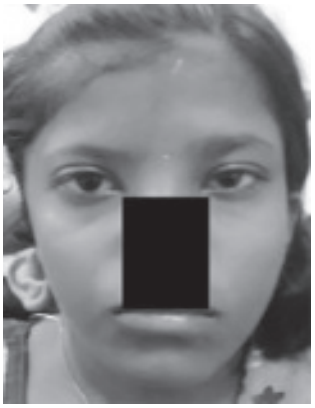

Fig.-5: *Patient with Treacher Collin syndrome*

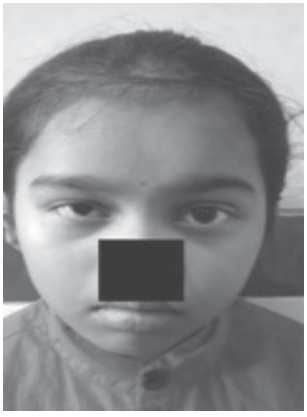

Fig.-6a: *Child with rt. sided microtia with right micro-ophthalmia and coloboma iris*

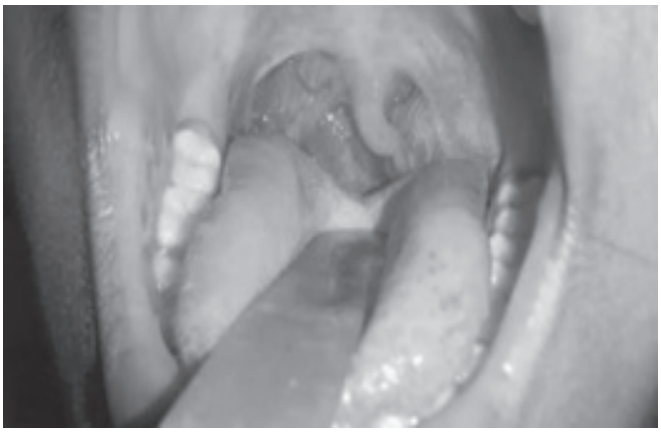

Fig.-6b: *Same patient with right sided tonsillar agenesis*

Also studies of Harris et al.[11] ;Mastroiacovo[12]; Canfield et al. [13]; Castilla and Orioli[14]; Forrester and Merz[15]; Shaw et al.[16]; Suutarla et al.[17] showed that microtia is associated with one or more additional congenital anomalies in 30–60% of the cases with a higher prevalence of congenital heart disease, cleft palate, esophageal atresia, vertebral anomalies, an/microphthalmia, and limb reduction defects.

In our series, all patients with atrial septal defect, ventricular septal defect and tricuspid regurgitation underwent surgery for correction of the same. Children with patent ductus arteriosus and mild mitral regurgitation were managed conservatively.

Four out of thirty patients underwent pinna reconstruction by the plastic surgeons. Others refused due to scar for rib graft and multistaged procedure. Hearing rehabilitation was done using Baha implant in nine patients and those who were young where bone thickness was not adequate were offered Baha soft band. Since ours is a tertiary care centre with patients belonging to lower socio-economic status, money for Baha implant and soft band was provided with the help of multiple donors and medical and social worker.

Since microtia associated syndromes may be associated with either single gene defects or chromosomal aberrations and hereditary forms of microtia with autosomal recessive or dominant pattern of inheritance with variable expression and incomplete penetrance are also known, genetic studies would be of great help for genetic counselling in families .[12] This was not included in our protocol due to high cost and poor socio economic condition of patients.

Conclusion

Thus, we conclude that congenital external deformities may be associated with spectrum of other anomalies and the patient should receive best coordinated care from otolaryngologists, audiologists, paediatricians, heart specialist to improve the quality of life. Early hearing assessment should be followed by proper rehabilitation for adequate speech and language development. We would like to emphasize that complete paediatric evaluation and additional investigations such as ultrasonography of the abdomen, 2-D Echo should be done to rule out commonly associated anomalies with microtia-anotia.

Conflict of interest: None

Ethical Approval: Yes

All procedures performed were in accordance with the ethical standards of the·institutional committee.

References

1. Simon C. Parisier MD, Jose N. Fayad MD, Charles P. Kimmelman MD. Microtia, Canal Atresia, and Middle Ear Anomalies. Ballenger's Otorhino-laryngology and Head and neck surgery (16th edition): 997-1008
2. Granstrom G, Bergstrom K, Tjellstrom A. The bone anchored hearing aid and bone-anchored epithesis for congenital ear malformations. Otolaryngol Head Neck Surg 1993; 109:46–53.
3. van der Pouw KT, Snik AF, Cremers CW. Audiometric results of bilateralbone-anchored hearing aid application in patients with bilateral congenital aural atresia. Laryngoscope 1998; 108:548–53.
4. Tjellstorm A, Hakannson B. The bone anchored hearing aid (BAHA) design, principles,indication and long term term results.Otolaryngol Clinic North Am 1995;115:1-20.
5. DV, Leoncini E, Mastroiacovo P. Microtia-anotia: a global review of

prevalence rates. Birth Defects Res A Clin Mol Teratol. 2011;91:813–822.

6. Anders Tjellstrom, Gösta Granstrom. The Bone Anchored Hearing Aid Otology Japan, Volume 20, Issue 1, Pages 23-30 2010 Journal Article, peer-reviewed.
7. Daniela V. Luquetti, Babette S. Saltzman, Jorge Lopez-Camelo, Maria da Graca Dutra4, and Eduardo E. Castilla. Risk Factors and Demographics for Microtia in South America: a Case-Control Analysis. Birth Defects Res A Clin Mol Teratol. 2013 November; 97(11): 736–743
8. van Nunen DP1, Kolodzynski MN2, van den Boogaard MJ3, Kon M2, Breugem CC. Microtia in the Netherlands: clinical characteristics and associated anomalies.. Int J Pediatr Otorhinolaryngol. 2014 Jun; 78(6):954-9
9. Mark Christopher Flynn . Cochlear Baha Attract System: Summary of clinical results and benefits. Technical report. Report number: E83122, Affiliation: Cochlear Bone Anchored Solutions AB.
10. Wang RY, Earl DL, Ruder RO, Graham JM., Jr Syndromic ear anomalies and renal ultrasounds. Paediatrics. 2001; 10: 8-32.
11. Harris J,Kallen B,Robert E.1996.The epidemiology of microtia anotia.J Med Genet 33:809-813.
12. Mastroiacovo P, Corchia C, Botto LD, Lanni R, Zampino G, Fusco D. . Epidemiology and genetics of microtia-anotia: a registry based study on over one million births. J Med Genet. 1995 Jun; 32(6):453-7.Source Department of Paediatrics, Catholic University, Rome, Italy.
13. Canfield, Langlois, Nguyen, and Scheuerle. Epidemiologic Features and Clinical Subgroups of Anotia Microtia in Texas. Birth defects research (part A) 85:905-913(2009).
14. Castilla EE, Orioli IM. Prevalence rates of microtia in South America. Int J Epidemiol. 1986 Sep; 15(3):364-8.
15. Forrester MB, Merz RD. 2005. Descriptive epidemiology of anotia and microtia, Hawaii, 1986-2002.Congenital Anom 45: 119-124.
16. Shaw GM, Carmichael SL, Kaidarova Z, Harris J.2004.Epidemiologic characteristics of anotia and microtia in California , 1987-1997.Birth Defects Res A Clin Mol Teratol 70:472-475.
17. Suutarla S, Rautio J, Ritvanen A, Ala-Mello S, Jero J, Klockars T. 2007. Microtia in Finland: Comparison of characteristics in different populations. Int J Pediatr Otorhinolaryngol 71:1211–1217.