INTERNATIONAL JOURNAL OF INNOVATIVE RESEARCH & DEVELOPMENT

ISSN: 2278 – 0211 (Online)

Bilateral Microtia : A Rare Case

Brijendra Singh
Additional Professor , Department of Anatomy, All India Institute of Medical Sciences, Jodhpur, India
Renu Gupta
Assistance Professor, Department of Anatomy, All India Institute of Medical Sciences, Jodhpur, India
Rajneesh Garg
Department of Orthopaedics, S.N. Medical College, Jodhpur, India

Abstract:

Anotia is a condition when ears are completely absent while microtia is a congenital anomaly of the face in which ear are malformed since birth that ranges in severity from mild structural abnormalities to complete absence of the ear and its parts. It occurs as an isolated birth defect or as a part of spectrum of anomalies or a syndrome. This may happen as a part of first branchial arch syndrome or in isolation too. The prevalence is considered to be higher in Hispanics, Asians, Native Americans, and Andeans. The etiology of microtia and the causes of this congenital anomaly is still not clearly defined or explained till now; there is wide variability in prevalence that too is poorly understood. Strong evidence supports the role of environmental and genetic causes for microtia. In this paper we tried to review current knowledge of the epidemiology and genetics of microtia, including potential candidate genes supported by evidence from human syndromes and animal models. Because our findings are entirely different about its hereditical and bilateral involvement makes this case a rare and special.

Key words: *Microtia, Anotia, Congenital Defect, Hereditary, Ear Deformity*

1.Introduction

The ear is composed of three parts: the internal, middle and external portions. Microtia is a congenital anomaly, characterized by a narrow, blocked or absent ear canal. Microtia can affect one ear (unilateral) or both ears (bilateral). It occurs in every 1 out 6000 to 12000 births. The right ear is more commonly affected than left and it is more common in male sex as compared to female sex. On basis of anatomical involvement microtia may be classified in grades; (Meurman 1957):

2.Classification

- Grade 1: A smaller version of a typical sized ear, still having same physical characteristic of a typical sized ear and containing a small but present external ear canal.
- Grade 2: A partially formed outer ear with very small or narrow ear canal. The ear canal may be very narrow or closed (canal stenosis) producing a conductive hearing loss.
- Grade 3: Absence of external ear with a small peanut shaped structure (some cartilage with mostly ear lobe) and an absence of the external ear canal and ear drum (known as aural atresia).
- Grade 4: Absence of the complete ear (anotia).
- In most instances the children with microtia disorders will have normal middle and inner ears, making a cosmetic condition rather than a typical cause of nerve deafness.
- In this paper we discussed a case of microtia on one side and anotia on otherside.

3.Case Report

A rare case of microtia was observed during OPD hours in Genetic Counseling Clinic at HAHC Hospital at Hamdard Institute of Medical Sciences and Research, Jamia Hamdard, New Delhi. A child was born with microtia (grade 3) on right side and a malformed external ear on the left side. On right side there was absence of external ear with small peanut shaped structure and absence of external ear canal (fig 1). While on opposite side a large lobulated external ear was there with stenosis in ear canal (grade 2) see (fig 2).

No one of either parents or their family members were having any congenital malformation or defects including face or ears specially. So a negative family history indicates role of some mutation or environmental factor had played some role at time of intrauterine development in form of radiation/ medication/ pollutants. A recessive genetic trait may be blamed for this anomaly because partial consanguinity in marriage of parent was there. Child was assessed and found free from other congenital malformation as well as diseases

4.Discussion

Cases of microtia may be difficult to define. Approximately 29% of the cases of diagnosed microtia do not have full atresia of the external auditory meatus (Castillo 1986). Anotia accounts for 13- 22% of the cases of microtia and anotia combined (Harris 1996, Mastroiacovo 1995). Anotia/microtia is an isolated condition in 65% of cases (Harris 1996, Mastrioiacovo 1995, Castillo1986), although several investigations reported isolated rates of less than 50% (Sanchez 1997, Castillo 1990). Over 80% of the cases of microtia or anotia are unilateral (Sanchez 1997, Mastroiacovo 1995). Of the unilateral cases of microtia or anotia, approximately 60% occur on the right side (Paulozzi 1999, Sanchez 1997, Harris 1996, Mastroiacovo 1995). Autosomal dominant inheritance of microtia or anotia has been reported in some families (Buyse 1990). By reviewing all studies mentioned above and other medical literatuire suggested that bilateral microtia/ anotia is a rare phenomena and if it is bilateral it may be heredity in nature but our findings are quite different because we found bilateral microtia/anotia without any hereditical association. That finding raise a question is microtia really has genetic correlation? Role of mutation can not be ruledout. Chromosomal abnormalities occur in 6-16% of cases of microtia or anotia (Sanchez 1997, Harris 1996). Chromosomal abnormalities associated with microtia or anotia include trisomy 21,trisomy 18, trisomy 13, and the deletion complexes 18q-, 18p-, and 5p- (Harris 1996, Carey 1993, Buyse 1990). Other birth defects associated with microtia/anotia include holoprosencephaly, facial clefts, cardiac defects, anophthalmia/microphthalmia, esophageal atresia, limb reduction deformities, renal anomalies, polydactyly, and vertebral anomalies (Wang 2001, Harris 1996, Mastroiacovo 1995).

5.Reference

1. Buyse ME, ed. Ear, Microtia-Atresia. In: Birth Defects Encyclopedia. Cambridge, Massachusetts:Blackwell Scientific Publications, 1990:591-592.
2. Carey JC. External ear. In: Stevenson RE, Hall JG, Goodman RM, eds. Human Malformations and Related Anomalies. New York: Oxford University Press 1993:193-219.
3. Castilla EE, Lopez-Camelo JS. The surveillance of birth defects in South America. In: Advances in Mutagenesis Research. New York: Springer-Verlag 1990:191-210.
4. Castilla EE, Orioli IM. Prevalence rates of microtia in South America. Int J Epidemiol 1986;15:364-368.
5. Harris J, Kallen B, Robert E. The epidemiology of anotia and microtia. J Med Genet 1996;33:809- 813.
6. 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
7. Genet 1995;32:453-457.
8. Meurman Y. Congenital microtia and meatal atresia. Arch Otolaryngol 1957;66:443.
9. Paulozzi LJ, Lary JM. Laterality patterns in infants with external birth defects. Teratology 1999;60:265-271.
10. Sanchez O, Mendez, Gomez E, Guerra D. Clinico-epidemiologic study of microtia]. Invest Clin 1997;38:203-217.
11. Wang RY, Early DL, Ruder RO, Graham JM. Syndromic ear anomalies and renal ultrasounds. Pediatrics 2001:108:2.