

# Rare Association of Bilateral Anotia with Congenital Facial Palsy

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## Abstract

Anotia is a birth defect and is associated with facial palsy and congenital heart defect as various presentation of cardiofacial syndrome. Herein we report a case of bilateral anotia and unilateral congenital facial palsy without congenital heart disease. It may occur due to single gene abnormality, teratogenic effect of maternal ingestion of thalidomide, retinoic acid, misoprostol or maternal diabetes, dietary lack of carbohydrates and folate.

**Key words:** Anotia, Congenital facial palsy

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## Introduction

Anotia or microtia, defined as absent or miniature external ears, is known to be a feature of cardiofacial syndrome and also seen to be associated with various other congenital anomalies like renal anomalies, short stature, small patella syndrome. Unilateral atresia is more common. Congenital facial palsy with bilateral anotia has not been seen so often. It is generally considered to be either developmental or acquired in origin. Developmental facial paralysis is associated with other anomalies including those of pinna and external auditory canal, ranging from mild defects to severe microtia and atresia<sup>1</sup>.

## The Case

A late preterm male with gestational age to 36 weeks and four days, a product of non-consanguineous marriage was delivered normally and cried soon after birth. His general physical examination was conspicuous by bilateral absence of external ear which was replaced by preauricular skin tag on the right side only (Figure 1). He also had unilateral congenital facial palsy, lower motor neuron type manifested by asymmetric crying facies, absence of right nasolabial fold and inability to close right eye (Figure 2). There was no other cranial nerve palsy and rest of the neurological examination was normal. His USG abdomen did not show any renal anomalies and ECHO was also within normal limits. MRI Brain was done to evaluate the cause for cong. facial palsy but, normal study warranted the need for HRCT Temporal bone which showed agenesis of petrous part of right sided temporal bone along with agenesis of facial and auditory nerve on right side only, non-visualization of bilateral external ears,

both bony and cartilaginous components with formation of bony bar in the expected location of tympanic membrane, non aeration of bilateral middle ear cavity and non pneumatization of bilateral mastoid air cells. The bilateral middle ear ossicles were dysmorphic forming a mass with non-delineation of normal malleus-incus anatomy. The right oval window was not visualized adequately. Bilateral internal auditory canals are symmetrical and normal in size. Mother denied any significant illness like diabetes or intake of teratogenic substances like retinoic acid thalidomide, misoprostol during antenatal period and there was no history suggestive of TORCHES infection during antenatal period. No folic acid supplementation was taken by her

during pregnancy. Hearing assesment done through BERA at the time of discharge revealed mixed type of hearing loss in right ear unlike conductive hearing loss alone in left ear.



**Fig 1:** Showing absence of ear on left side



**Fig 2:** Showing inability to close right eye and loss of nasolabial fold on right side suggestive of facial palsy

## Discussion

Microtia/anotia refers to abnormal development of external ear resulting in deformed auricle. It ranges from mild deformity to complete agenesis of ear. Incidence is 1:10000-20000 births, most often unilateral with right side predominance and male to female ratio is 2.5:1 associated with congenital aural atresia<sup>2</sup>. It is classified into four types i.e. Type 1 consist of mild deformity, major structures present to some degree and no tissue needed. In Type 2, major structures present but tissue needed to correct deficit. Type 3 has few recognizable landmarks, lobule present to some degree and Type 4 is anotia which is seen in our case<sup>3</sup>. Some of the syndromic associations<sup>4</sup> include congenital infections such as rubella, retinoic acid embryopathy, otomandibulofacial dysostosis, Meir-Gorlin syndrome, thalidomide embryopathy, fetal alcohol syndrome, diabetic embryopathy/holoprosencephaly, etc. According to retrospective review<sup>5</sup> of 118 cases of aural atresia, prevalence of ear abnormalities with facial palsy is seen in 13% of cases where as inner ear abnormalities are seen in 22% of cases.

Congenital facial nerve palsy is an infrequent condition with a reported incidence of 0.8- 2.1 per 1,000 live births<sup>6</sup>. In 78% of cases CFP is related to birth trauma. No such history was available in the index case. Other causes include, intrauterine posture, intrapartum compression, and familial and congenital aplasia of the nucleus; the last being most frequently reported for bilateral cases. There are a number of syndromes which encompass CFP as part of their symptoms, including the cardiofacial, Moebius, Poland's, and Goldenhaar's syndrome<sup>1,7</sup>. Some cases of CFP have been attributed to agenesis of the petrous portion of the temporal bone, with resulting agenesis of the facial and auditory nerves, the external ear and the mastoid region<sup>7</sup>. CFP may cause multiple problems in newborn like difficulty in nursing, eye closure, development of speech, expression of emotions, mastication. Cardiofacial syndrome is a well known clinical entity with asymmetric crying facies giving a clue to the underlying abnormalities like congenital heart disease, however, external ear anomalies are not described as a syndrome. Findings in our patient differs from the cardiofacial syndrome (asymmetric crying facies syndrome) reported by Patel<sup>8</sup> as well. His patients had congenital heart disease associated with unilateral weakness of only the lower portion of the face. The triad of anotia, facial paralysis, and congenital heart disease has been reported in association with the teratogenic effect of thalidomide and retinoic acid or as a possible variant of Goldenhar syndrome. However, these associations were not observed in the present case. As opposed to well formed ossicles and normal BERA found by Gathwala<sup>9</sup>, this case had dysmorphic ossicles.

In addressing CFP, some medical professionals advise initial surgery during preschool to avoid psychosocial problems associated with physical abnormality. However waiting till adolescence when facial growth is mature and child is able to understand the risks and benefits of surgery have merit<sup>10</sup>.

Muscle transplantation for facial palsy is effective but better results are observed in case of traumatic facial palsy<sup>11</sup>, in contrast facial palsy, as in the index case, carry a poor functional outcome. Reconstruction of auditory canal and pinna has to be done around 6 years of age when planning for muscle transplantation surgery with facial palsy<sup>12</sup>.

## Conclusion

Bilateral anotia is a rare congenital malformation. Moreover the well known and commonly reported

association of congenital heart disease with congenital facial palsy was not found in this case. The rarity of such phenomena prompted us to report the case. It was difficult to find out the cause here, but lack of folic acid supplementation during antenatal period can be further studied as one of the aetiology resulting in congenital malformations of ear etc.

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