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Otocephaly: Agnathia Microstomia Synotia Syndrome - A Case Report

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1. Introduction

Otocephaly is a rare fetal malformation characterized by the association of agnathia (mandibular agenesis)/ mandibular hypoplasia, melotia (ventromedia / auricular malposition) with or without auricular fusion (synotis) and microstomia[1]. This condition is rare, seen in less than 1 in 70000 births[2,3]and is considered lethal because of ventilatory difficulties which typically lead to death in affected babies shortly after birth. To the best of these authors' knowledge, no case of Otocephaly has been reported in Nigeria and West Africa. We report here a case of Otocephaly complex seen in a Nigerian preterm in a private hospital in Port-Harcourt Nigeria.

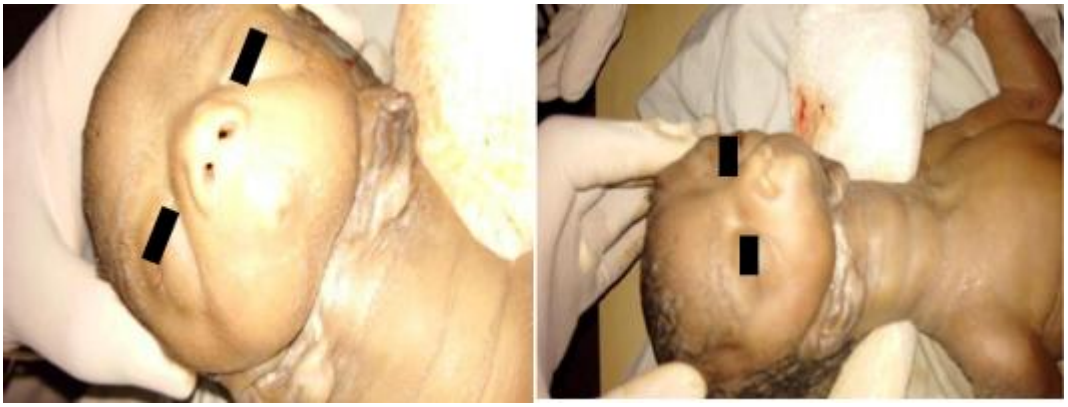
2. Case Report

Baby S.I. was a few minutes old female whose mother, a 24year old booked Primigravida was admitted into our facility on account of preterm rupture of membrane at 32weeks gestation and was placed on dexamethasone and prophylactic antibiotics.

Two days later, the baby was born through spontaneous vertex weighing 2300g with Apgar scores of 2^1, 2^5 and 3^{10} minutes respectively. Conception was preceded by 9months of primary infertility. Obstetric scan done at 9weeks was normal but a repeat at 29weeks showed polyhydramnios. There was no history of consanguinity in parents, febrile illness nor history of exposure to radiation or use of unprescribed medication in early pregnancy.

On examination, gross neonatal abnormalities were observed including: small oral aperture, absence of mandible, external ears were displaced ventromedially in the neck close to the midline, bulging anterior chest wall and scaphiod abdomen. The baby died few minutes after birth.

Parents were counseled initially on ultrasound finding of polyhydramnios and subsequently on the need for autopsy following patient's demise but they declined on religious grounds.



3. Discussion

Otocephaly also known as agnathia-Otocephaly is considered a rare lethal congenital abnormality[1,2] . It was first described by Kerckring in 1717[1].Its incidence is estimated at less than 1 in 70000 births[2, 3] . It could present as an isolated agnathia (absence or hypoplasia of the mandible) [1,4] or in combination with other anomalies such as microstomia (a small or totally absent mouth), melotia (a ventromedial auricular malposition), synotia (auricular fusion at the midline), cardiopathy, situs inversus[1], other visceral anomalies and holoprosencephaly[1,2].

The physical abnormalities of small oral aperture, absence of mandible, ventromedially displaced external ears at the neck close to the midline as seen in this index case, were in keeping with those described in Otocephaly[1,2]. Other abnormalities noted were: bulging anterior chest wall and scaphiod abdomen. The presence of bulging anterior chest wall is suggestive of marked cardiomegaly/ congenital cardiac disease, and a scaphiod abdomen is suggestive of diaphragmatic hernia. The baby died shortly after birth due to respiratory difficulties, a major challenge in Otocephalic patients. It is possible that autopsy may have revealed other internal organ malformations.

This rare abnormality is thought to be a consequence of first pharyngeal arch malformation at 4th and 5th weeks gestational age due to failure of migration of neural crest cells from the hindbrain[2,5,6]. Different authors have postulated different theories regarding the etiology of Otocephaly. *Schiffer et al*[7] and *Sergi et al*[8], identified a heterozygous loss-of-function mutation in the PRRX1 gene while *Donnelly et al*[9] reported a heterozygous frame shift mutation in possible gene PRRX1. The mode of inheritance is not clear as *Erlich et al* [10] in a study found dysgnathia in mother and daughter which suggested an autosomal dominant inheritance. They also suggested that the defect in the OTX2 gene could be the basis of the disorder[10].

Exposure to teratogens such as streptonigrin antibiotics, trypan blue, theophylline[11] and prenatal alcohol[12] have also been implicated in mal-development of the mandible.

Diagnosis of Otocephaly depends mostly on a combination of prenatal 2D and 3D ultrasound and magnetic resonance imaging (MRI) [2,10]. Most cases are diagnosed incidentally following abnormal imaging findings such as polyhydraminous, holoprosencephaly, situs inversus, totalis encephalocele or renal defects[2,6,10,13].

Prenatal diagnosis of Otocephaly and other rare congenital malformations relies on both expertise in imaging studies and the availability of 3D ultrasonography machines and MRI all of which are not readily available in resource-poor setting such as ours. Even where these are obtainable, affordability is a challenge to a majority of patients who has to pay out of pocket. Until universal health insurance is available to all, cost will continue to be a major hindrance to prenatal diagnosis and treatment in these settings.

4. Conclusion

Accurate prenatal diagnosis of Otocephaly is possible, hence the 22weeks anomaly scan should be done routinely by experienced radiologists especially in the presence of polyhydramnios. Prenatal diagnosis of lethal congenital malformations will facilitate appropriate parental counseling and enhance decision regarding intra-partum and immediate post-partum intervention. Otocephaly should be considered as a differential in all cases of polyhydramnios especially whenever there is failure of visualization of the mandible and the ears are located in a lower and medial position.

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