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Case Report

Pathology Section

Otocephaly: Agnathia- Microstomia-Synotia Syndrome– A Rare Congenital Anomaly

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ABSTRACT

Otocephaly is a rare malformation characterized by the association of agnathia (agenesis of mandible) or mandibular hypoplasia, melotia (anteromedial malposition of ears), microstomia (small mouth), aglossia or microglossia (absent or rudimentary tongue). This rare anomaly of the ventral portion of first brachial arch is a consequence of failure of migration of neural crest cells from hind brain. It leads to the development of maxillary and mandibular prominences and starts to develop at the fourth and fifth week of gestation. We hereby present the autopsy findings of a fetus of 28 weeks gestation abortus having otocephaly without holoprosencephaly.

Keywords: Congenital craniofacial anomaly, Microglossia, Prenatal diagnosis

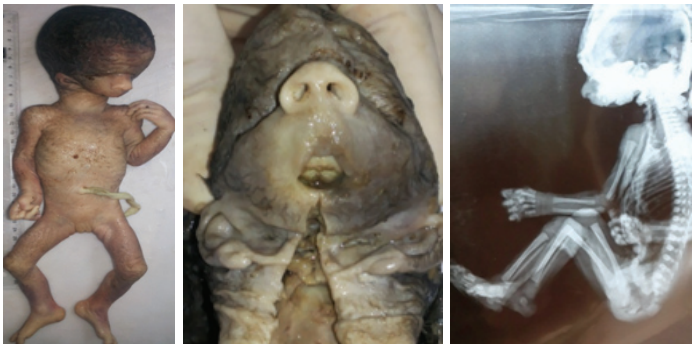
CASE REPORT

A 25-year-old female having consanguineous marriage came with history of 28 weeks of amenorrhea associated with pain in abdomen and per vaginal bleeding since 2 days. She had obstetric history of G1P1A0. There was no history of any contraceptive use. She had no history of any hormone intake, drug intake or radiation exposure. She had no significant systemic illness and familial history. There was no history of maternal hypertension, diabetes mellitus, recent infection or any exposure to teratogens.

USG examination showed 28 weeks (+ or – 1 week) gestation with oligohydramnios and absence of fetal cardiac activity suggestive of fresh intrauterine death. There was absence of mandible and various craniofacial anomalies. On gynecological examination, cervix was found to be dilated with spontaneous rupture of membrane and patient was in second stage of labour.

The autopsy findings revealed a female fetus weighing 720 gm, having crown to heel length 40 cm, with head circumference 24 cm. The complete autopsy was performed which revealed severe craniofacial anomalies – microstomia, hypertelorism, choanal atresia, high arched palate, low set anteromedial position of ears [Table/Fig-1], absence of mandible, microglossia [Table/Fig-2]. Cranial cavity showed hydrocephalus.

Radiographic examination of the autopsy specimen of fetus showed agnathia [Table/Fig-3]. There was no situs inversus. There was partial persistence of buccopharyngeal membrane with hypoplasia of both the lungs. Rest of the systemic examination was unremarkable on gross and histopathological study. Based on these above findings the final diagnosis was given as Otocephaly: Agnathia- Microstomia-Synotia Syndrome.



[Table/Fig-1]: Showing fetus with craniofacial anomalies. **[Table/Fig-2]:** Showing Cranio-facial anomalies: microstomia, low set anteromedial position of ears and microglossia **[Table/Fig-3]:** Radiograph of autopsy specimen of fetus showing agnathia.

DISCUSSION

Otocephaly is a rare, often lethal has familial anomaly characterized by microstomia, aglossia, agnathia and synotia [1]. The term 'oto" refers to the relationship of the ears to the face. Otocephaly, with associated anomalies are considered lethal due to severe respiratory dysfunction. Development of lower facial severe malformation can be isolated. It is also often associated with facial, cranial and extracranial malformation [2,3]. Otocephaly is usually suspected on radiological antenatal checkup when it is impossible to visualize the mandible and ears are in a very low and medial position. Otocephaly can be an isolated malformation or associated with other anomalies. The severe first and second arch defects [4,5] are responsible for this malformation. Its incidence is less than 1 in 70,000 births [2].

The syndrome complex of otocephaly is divided into four types: 1) Isolated agnathia; 2) Agnathia with holoprosencephaly; 3) Agnathia with situs inversus and visceral anomalies; 4) Agnathia, holoprosencephaly, situs inversus and other visceral anomalies [6].

In the present case it was an Agnathia-Microstomia- Synotia malformation without holoprosencephaly. It is an extremely rare malformation and few cases have been reported in literature recently as reported by Wagner Jou Hisaba [7] where he has found agnathia-otocephaly with no evidence of changes in brain morphology.

Holoprosencephaly is most common developing disorder of the developing forebrain in human. In Agnathia spectrum association of holoprosencephaly ranges from 0.8%-10% [8]. Other associated anomalies with otocephaly are neural tube defects, cephalocele, dysgenesis of corpus colossus, atresis of third ventricle, midline proboscis, renal ectopia vertebral and rib abnormalities, tracheoesophageal fistula, cardiac anomalies, vertebral anomalies and adrenal hypoplasia [9]. Otocephaly anomaly shows spectrum of various manifestations ranges in severity from severe micrognathia as a part of the robin sequence to cyclopia- holoprosencephaly complex invariably associated with fetal death. In our case it was severe form of anomaly without holoprosencephaly and situs inversus. A case reported by Wagner JH was noted that in 25 weeks of gestation with facial malformation in otocephly with proboscis, cardiac abnormalities were observed but without any changes in brain morphology [7].

Otocephaly with spectrum of malformations is rare and lethal, therefore it should be properly evaluated in prenatal checkup for proper management and diagnosis of cases.

CONCLUSION

Otocephaly is usually incompatible with life, so it is important to diagnose on routine antenatal radiological checkup when the mandible cannot be visualized and fetal ears are noted to be abnormally placed. First trimester screening with demonstration of fetal profile for facial anomalies and evidence of polyhydramnios with use of 3D ultrasound will contribute in future.

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