**Otocephaly: A Rare Congenital Anomaly - A Case report**

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**ABSTRACT**

Otocephaly is rare lethal anomaly of the ventral portion of first branchial arch. The “Oto” Greek word meaning ear and cephaly meaning head; Otocephaly virtual meaning is type of head disorder. Otocephaly is a malformation characterized by Agnathia (agenesis of mandible), Melotia and Synotia (ventro-medial malposition and midline fusion of external ears) Microstomia (small mouth) and Aglossia (absence of tongue) or microglossia (small tongue). This condition is lethal because of poorly functioning airway leading to severe respiratory dysfunction. We hereby present female fetus of 28 weeks gestation, spontaneously aborted having Otocephaly alone with no other associated anomalies. Early diagnosis on routine prenatal radiological checkup, later followed by 3D ultrasound will contribute for more accurate diagnosis because Otocephaly-Agnathia syndrome complex.

**Key words:** Agnathia, Synotia, Microstomia, Aglossia, First branchial arch

**Introduction:**

Otocephaly is rare non-familial, neurocristopathy of First Pharyngeal arch.¹ This malformation is considered lethal due to severe respiratory dysfunction. The main feature is Agnathia, Melotia and Synotia of external ears in neck region, Microstomia and Aglossia. This anomaly is a consequence of failure of migration of Neural crest cells from Hindbrain which contributes to development of first branchial arch at 4th and 5th week of gestation. These birth defects can be detected prenatally by various investigations i.e. radiologically and USG. We discuss a rare case of Otocephaly complex patterning, associated with current perspectives and its review of literature.

Case report We present a case of 22 year old primigravida with no family history of any congenital anomalies or consanguinity, presented for routine antenatal examination at 28 weeks of pregnancy. There was no antenatal history of infection/radiation or toxin exposure/drug intake. There was no maternal history of Hypertension, Diabetes mellitus or Asthma. She was advised Ultrasonography (USG) to rule out any anomalies.

**USG report:**

1. It showed single live fetus of 28 weeks maturity with mild Polyhydramnios associated with various craniofacial anomalies. The Mandible was not made out, the Ear could not be made out, but an extra soft tissue in the region of neck and various craniofacial anomalies.

2. No other intracranial or other congenital abnormality was detected.

3. Umbilical cord and Placenta was normal. A diagnosis of Otocephaly was made. She was explained about the USG report and advised MTP. The consent was taken. Labor was induced, female baby was delivered. The baby showed multiple facial anomalies. The fetus expired immediately.

**On External Examination: (Fig.1)** The autopsy findings revealed a female fetus weighing 800gms, crown heel length 40cms and head circumference
25cms. The complete autopsy was performed which showed severe craniofacial anomalies - Agnathia (absence of mandible), Microstomia (small mouth), Choanal atresia, Ears were extremely low set in the anteromedial region of neck and fused (Synotia). External auditory meatus was not patent. Other facial features were down slanting of palpebral fissures and hypertelorism. Autopsy showed that brain, thorax and abdomen viscera were normal. Diagnosis was Otocephaly-Agnathia complex with no other associated malformations.

Discussion

Otocephaly is rare disorder is almost always lethal. It is characterized by Agnathia (absence of mandible), Melotia (ventro medial auricular malposition) Synotia (auricular fusion), Microstomia, and Aglossia with persistent Buccopharyngeal membrane. It was first described by Kerckring in 1717. Otocephaly is suspected on USG or on Radiological antenatal check-up when it is impossible to visualize the mandible and ears are in a low set and medial position. It has been considered as the 1st branchial arch structures, it may also result in dysmorphogenesis of other midline craniofacial structures involving Prosencephalon abnormalities like Holoprosencephaly most commonly associated and axial body abnormalities. Mainly affecting skeletal, genitourinary, gastrointestinal, cardiovascular systems. Sometimes resulting in Pituitary or Adrenal or Thyroid aplasia or hypoplasia and or entire Pituitary axis insufficiency. Prenatal diagnosis of Otocephaly is usually very difficult. Most of the cases were found incidentally when other malformations such as holoprosencephaly, situs inversus totalis or renal defects were identified. Most of cases are diagnosed during third trimester. Polyhydramnios is more often associated with Otocephaly. The anatomic sub classification of Otocephaly-Agnathia syndrome complex is divided into four types

1. Isolated-Agnathia.
2. Agnathia with Holoprosencephaly.
3. Agnathia with Situs inversus and other visceral anomalies.
4. Agnathia, Holoprosencephaly, Situs inversus and other visceral anomalies. The present case is under discussion of type 1 Otocephaly-Agnathia, Microstomia-Synotia malformation without Holoprosencephaly or Brain morphology or Situs inversus and visceral anomalies. It is an extremely rare anomaly and few cases have been reported in literature.

Embryonically the face, neck, nasal cavity, oral cavity, tongue, larynx and pharynx develop from Branchial arches. In Otocephaly-Agnathia syndrome complex is a consequence of failure of migration of neural crest cells from the Hindbrain to maxillary and mandibular prominences of 1st branchial arch. Paul et al. suggested an autosomal recessive theory. The 1st branchial arch patterning is also influenced by interplay between genes and gene products and of bone morphogenic protein 4 (Bmp4) Polyhydramnios is frequently associated with Otocephaly. Differential diagnosis includes Treacher Collins Syndrome, Goldenhar syndrome and Mobius syndrome. Agnathia-Microstomia-Synotia malformation without holoprosencephaly is an extremely rare malformation and few cases have been reported in literature. Recently it has been reported by Wagnar Jou Hisaba where he has found agnathia -otocephaly with no evidence of changes in brain morphology. Celik et al reported a female infant born of consanguineous parents with otocephaly who died shortly after birth from respiratory distress. On autopsy the baby had other associated anomalies like cardiac defects.

Aparna VP, Himabindu N et al reported a case with Otocephaly associated with Situs inversus complex with bilateral absence of mandibular nerves. The present case is under discussion of type 1 Otocephaly-Agnathia, Microstomia-Synotia malformation without Holoprosencephaly or Brain morphology or Situs inversus and other visceral anomalies. It is an extremely rare anomaly and few cases have been reported in literature. Otocephaly with spectrum of defects is rare and lethal, hence it should be properly evaluated in antenatal checkup for efficient management.

**Conclusion**

To conclude Otocephaly-Agnathia syndrome complex is rare and is usually in compatible with life. Early diagnosis on routine prenatal radiological checkup when the Mandible cannot be visualised and fetal ears are abnormally placed. Later followed by 3D ultrasound will contribute for more accurate diagnosis because Otocephaly-Agnathia syndrome complex has poor prognosis due to severe respiratory failure.

**References**

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