

## **OTOCEPHALY - A CASE REPORT**

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### **ABSTRACT:**

Otocephaly is a condition characterized by aplasia or hypoplasia of mandible, a small oral fissure and low lying ears at the level of neck usually meeting in the midline. this case report is about a male child with otocephaly, who was unable to survive for long after birth.. The reporting of such cases is important to know the incidence of the anomaly, for further study on the developmental defects or determine the causitive agents/risk factors leading to such conditions and to make strategies to decrease the incidence.

Key words: Otocephaly, maldevelopment, hypoplasia, microstomia

**Introduction:**

Otocephaly is a rare often lethal, non familial syndrome characterized by severe mandibular hypoplasia, synotia, microstomia and severe hypoplasia of tongue<sup>1</sup>. Otocephaly is commonly associated with other midline defects like holoprosencephaly, cyclopia, proboscis & other cerebral malformations<sup>2</sup> patient may present with polyhydramnios. The defect is basically related to maldevelopment of first branchial arch. Such cases can be diagnosed prenatally by ultrasonography. Their poor prognosis makes them a suitable case for termination of pregnancy.

This case is being reported because of its rarity.

Prenatal diagnosis is sporadically done, mostly in third trimester of pregnancy. Diagnosing such cases at an early stage by the radiologists can decrease the maternal morbidity.

**Case report:**

A 29 years old female, housewife, from a low socio economic strata, gravida 2 para 1 presented in a private hospital (Kohli Hospital) at Gurdaspur, with breech presentation, labour pains, leaking per vaginam and non-progress of labour. On examination, patient was in distress, anaemic, had low grade fever and was not handled. Cervix was 3 cm dilated, thick with a foul smelling discharge. Fetal heart sound was present. Patient did not have any antenatal check-ups or any investigations done except for getting tetanus toxoid injections from the dispensary. There was no history of alcohol, smoking or any drug intake. No history of Diabetes, hypertension, Tuberculosis was there.

In the past history, Patient had had one full term normal vaginal delivery ( 2 years back) giving birth to a dead male baby (still birth) at home . The exact cause of death of that baby could not be ascertained as the parents neither took the baby to the hospital, nor the autopsy was done.

Investigations (Complete blood counts) revealed leucocytosis. Under aseptic conditions, lower segment caesarean section (LSCS) was done under spinal anaesthesia delivering a male baby of 36 weeks gestation. Baby was pink, had muscle tone but could not be revived as he could not breathe. The nasogastric / endotracheal tube could not be passed either through the nose or mouth because of some congenital malformation.

On examination, the baby had a very small oral aperture (microstomia), poorly developed lower jaw (mandibular hypoplasia), external ears displaced ventromedially in the neck close to the mid line (synotia) (figure-1 ). Autopsy was advised but refused by the attendants. The mother was given post-operative care & was discharged in a good condition. Risk factors leading to such congenital anomalies were explained to the mother and she was advised to go for further investigations like TORCH test to rule out any infection. Chromosomal karyotyping was advised to both the parents before the next conception to avoid recurrence of any malformation in subsequent pregnancy. Mother was advised to have regular antenatal check-ups/ ultrasonography in case of subsequent pregnancy to detect any such congenital malformation at an earlier stage. and to be managed accordingly.

**Discussion:**

The incidence of otocephaly which is a rare idiopathic malformation in humans is 1 per 70,000 new borns<sup>3</sup>. Every pharyngeal arch has a core of cartilage derived from neural crest cells, unsegmented mesoderm capable of forming skeletal muscle and bone, an artery from aortic arch and a nerve that enters it from the brain stem carrying motor fibres to supply the skeletal muscle derived from the mesoderm<sup>4</sup>.

Arrest in the development of the first branchial arch due to an insult to the neural crest cells has been suggested to be the cause

of this malformation, which may be induced by chromosomal mutation or teratogens<sup>4</sup>. Primary defect in the migration of neural crest cells to the distal end of mandibular arch leads indirectly to the failure of ascent of developing auricles.

Neural crest cells are vulnerable to a number of compounds like alcohol, retinoids etc, may be because of lack of catalase and superoxide dismutase enzymes that scavenge toxic free radicals<sup>5</sup>. Streptonigrin antibiotics, trypan blue<sup>6</sup> and theophylline<sup>7</sup> may also have teratogenic effects resulting in otocephaly. In this case, there was no history of alcohol intake, smoking or use of any drugs.

Mandible forms from the central portion of the first arch cartilage, by keeping pace with its growth, guides its early morphogenesis. The body of tongue, part of external ear and middle ear equally derive from the same arch explaining the association of otocephaly with microglossia and ear abnormalities. Downward displacement of ears with close proximity and microstomia can be explained due to lack of spatial separation because of hindrance in the growth of mandible<sup>1</sup>.

The infant in the present case had both the external ears located in the neck near the mid line, the mandible was not fully developed and mouth was also very small together indicating a case of otocephaly, failure to perform endotracheal intubation indicates the presence of other associated anomalies like mouth floor atresia. Nasogastric tube could not be passed, may be an indication of oesophageal atresia. Patients with otocephaly may have associated cardiac defects, oesophageal atresia, pulmonary agenesis<sup>8</sup>. In this case, Mother's medical, family history was non contributory and unremarkable, but she had an intrauterine death (IUD) in the past. Otocephaly has been studied extensively in guinea pigs, where it is thought to be inherited, same theory has been postulated for humans, but still not proved<sup>9</sup>.

### Conclusion:

Prenatal diagnosis of this condition is possible, as it should be considered as one of the differential diagnosis in case of polyhydramnios. 2 dimensional or 3 dimensional Ultrasonography can detect the abnormalities found in this condition<sup>10</sup>. Regular ultrasonography during antenatal visits, can detect this condition at an early stage. The poor prognosis of this condition should be discussed with the parents and termination of pregnancy should be offered as an option.

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Figure-1 Showing microstomia, hypoplastic lower jaw, external ears close to the midline.



Figure-2 Showing the external ears close to the midline.