Otocephaly: Case Report, Literature Review, and Ethical Considerations

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Introduction

Otocephaly complex is a rare congenital condition that presents with microstomia, mandibular hypoplasia or agnathia, and ventromedial malposition of the ears. These anomalies are due to failed mandibulofacial development by mesenchymal cells of the first pharyngeal arch, secondary to both genetic and teratogenic factors.

The incidence is less than 1 in 70,000 births and prenatal diagnosis remains a challenge. Due to the high prevalence of respiratory failure, only 7 cases have survived infancy. Tracheal intubation is usually impossible due to stomal choanal atresia, hypopharyngeal stenosis, or upper airway obstruction. Several prenatally diagnosed cases were managed via an ex-utero intrapartum treatment (EXIT). Of the long term survivors, all have required tracheostomy, as well as a feeding or gastrostomy tube, and the majority received reconstructive mandibular surgery.

We present a case of a neonate delivered with an unanticipated otocephaly complex who required emergent airway management. We also discuss the ethical quandaries associated with caring for a newborn presenting with both a confounding and probable lethal prognosis.

Case

A 20-year-old G4P1111 woman (71.4 kg, height 1.67 m) presented at 31-weeks gestation with preterm labor.

Medical History: hypothyroidism, major depression
Obstetric History: one preterm, one term, one ectopic pregnancy s/p laparoscopic salpingectomy. *E. coli* pyelonephritis during this pregnancy, with nitrofurantoin suppression therapy until 5 months gestation. Normal quad screen at 28 weeks gestation.

Initial exam:
Cervix: 6 cm dilated, cephalic presentation.
FHT: category 1, active fetal movement. FHR 130 bpm with both moderate variability and accelerations. No decelerations were noted.
Bedside fetal U/S: vertex, anterior placenta, grossly normal amniotic fluid level, EFW 1650 g.

Admitted to L&D with preterm labor: pretreated with betamethasone, magnesium sulfate, and penicillin G.

7 hours later, cervix 9 cm dilated. Brought to OR for imminent delivery.

Neonate delivered vaginally. No cry or respiratory effort. Congenital malformations of the head noted.
APGAR: 2/2/1 @ 1/5/10 mins.

BMV was immediately initiated by a single provider.
Anesthesiology team arrives, the neonate was cyanotic and SpO_2 ~70%.

A 2-person technique was subsequently utilized for BMV.
Air was found to be emanating from both of the patient's ears, which were on her neck. Successful mask ventilation was achieved by covering ear canals b/l with earlobes and neck extension → SpO_2 ~90-100%.

Tracheostomy by trauma team while 2-person BMV continued → cardiopulmonary arrest → withdrawal of care.

Autopsy report: marked mandibular hypoplasia, microstomia, oropharyngeal hypoplasia, a probe-patent communication between the oropharynx and the trachea, and synotia. These were consistent with **otocephaly complex**. The oropharynx communicated with a narrowed upper trachea. Below the tracheostomy site, the trachea was of normal caliber, with no appreciable obstructions. The ears were low-set and came together under her malformed mandible. No other abnormalities noted (Female, 1435 g, 35 cm).

Medical Knowledge

Prenatal diagnosis remains difficult in spite of recent advances in prenatal imaging:
• 3-D and 4-D U/S, fetal MRI
• Many found incidentally through careful exam due to other existing pathology

U/S:
• 10 weeks gestation + labor
• Both failed to reveal craniofacial abnormalities
• Impossible to prepare for EXIT procedure

Fetal exposure to teratogens has been implicated:
• Smoking, alcohol, radiation, streptonigrin antibiotics, trypan blue, theophylline, beclomethasone, salicylates, amidopyrine, mycophenolate, and phenytoin

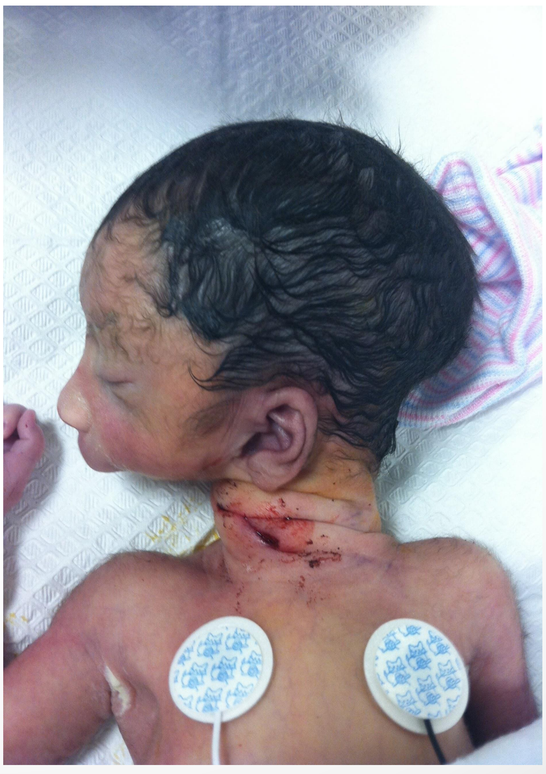
• Nitrofurantoin at months 1-5 gestation, Levothyroxine throughout

Ethical Views

Antenatal discussion
• Bioethical principles of beneficence and non-maleficence
• Viability of neonate
• Futility of care
• Benefits and burdens of treatment

Acute and Emergent Situation
• Unexpected circumstances

Responsibilities of the neonatologist and anesthesiologist
• Primary resuscitation
• Family discussion
• Stopping care after starting
• Bereavement and support



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