A case of otocephaly-agnathia

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Objective

Otocephaly is characterized by an absent mandible (agnathia), a small mouth (microstomia) and a midfacial location of the ears in the form of auricular malposition called melotia/synotia extremely rare lethal anomaly. Otocephaly occurrs following 4 types: 1. Otocephaly-agnatia 2. Otocephaly+holoprosencephaly 3. Otocephaly+situs inversus 4. Otocephaly-agnatia+ holoprosencephaly+situs inversus. This was the first antenatal diagnosed case in Mongolia.

Methods

Otocephaly can be found isolated or associated with anomalies such as holoprosencephaly, neural tube defects, cephalocele, midline proboscis, tracheoesophageal fistula, cardiac anomalies, adrenal hypoplasia, genitourinary and renal malformations, and skeletal anomalies. Most cases are sporadic, but some drugs such as theophylline, beclomethasone, salicylates and maternal alcohol consumption are reported as associated substances. In this case, we are not able to confirm and to deny in association with otocephaly and drug exposure using unknown contraceptive pills.

Results

A 37 years old, gravid 5, para 4, woman with no family history congenital anomalies. She had 4 normal delivered. She has been monthly used Chinese contraceptive pill since her last para (2013), her menstrual period was irregular. She revealed became pregnant about 20 days later taken the pill. The patient's medical, obstetrical, and family histories were unremarkable and noncontributory. Sonographic evaluation demonstrated a single live fetus biometric data consistent with 29-30 weeks. Detailed examination of the fetal anatomy revealed multiple anomalies of the fetal face, characterized by agnathia, sloped forehead, fetus has big nose (like a pig nose), low set big ears, hypotelorism. The remaining fetal anatomy was normal, with increased amniotic fluid volume (AFI=40. 4 cm). Ultrasound examination performed by Philips, EPIQ5 and GE, Voluson E8. Three times amnioreduction was done to reduce mother's discomfort due to polyhydroamnios, total amount 2600 ml of amniotic fluid. Medical team together with the parents decided to the deliver in face of the maternal risks of the polyhydramnios and the poor fetal prognosis. Delivery occurred at 33 weeks by labor induction after cervix preparation by acupuncture. A male newborn with 1700g with large amount of 5l amniotic fluid. Death occurred spontaneously after a minutes of the birth. In postmortem X ray revealed absent mandible. The autopsy findings confirmed the prenatal ultrasonographic diagnosis of isolated otocephaly-agnathia. No other malformations were uncovered on postmortem analysis.

Conclusion

Thus otocephaly -agnathia, astomia, and synotia diagnosed by 3D/4D ultrasound and TUI (tomographic ultrasound image) programme in National Center for Maternal and Child Health.