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PICTORIAL ESSAY

Prenatal Diagnosis of Otocephaly: A Rare Facial Anomaly

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Abstract

Introduction Otocephaly is a rare malformation characterized by agnathia (absence of the mandible), melotia (medially displaced ear pinna), aglossia (absence of the tongue) and microstomia (small oral aperture). This results due to failure of migration of the neural crest cells and is a defect of the first branchial arch. It is incompatible with life and early prenatal diagnosis is useful.

Case Report Our patient a primigravida with 19 weeks 6 days gestation was referred for micrognathia and polyhydramnios. On ultrasound examination, she had unilateral mild ventriculomegaly and posterior fossa cyst in the fetal brain. The fetus had agnathia and anophthalmia. There was an echogenic intracardiac focus and echogenic bowel. The stomach was not seen clearly. This could be due to agnathia and microstomia leading to swallowing difficulties. The patient was explained about the guarded prognosis. The pregnancy was terminated. A diagnosis of otocephaly was made.

Discussion Otocephaly is a rare disorder of development of the first branchial arch. The reported incidence is 1 in 70,000. It is mostly lethal due to respiratory difficulties and may be associated with cranial and extracranial malformations. Most case reports have found that it is sporadic and could be due to mutations in the PRRX1 gene. Other anomalies that may be associated with otocephaly are neural tube defects, cephalocele, dysgenesis of corpus callosum, atresia of the third ventricle, midline probocis, hypotelorism, renal ectopia, cyclopia, vertebral and rib abnormalities, tracheo esophageal fistula, cardiac anomalies and adrenal hypoplasia. Most of the cases reported so far were diagnosed in the second or the third trimester. Facial anomaly screening has undergone a huge evolution in the recent years. In addition to the usual facial screening, we recommend mandibular arch screening in the first and early second trimester. If there is a doubt the patient may be called back at 15 to 16 weeks of gestation considering the fact that these anomalies are usually lethal and medical termination is safer earlier in pregnancy than later. MRI may be a handy tool to confirm antenatal diagnosis as it can detect the abnormal ears. Agnathia and polyhydramnios occur together in the third trimester but in the first or second trimester polyhydramnios may not be observed.

Conclusion Otocephaly, though rare, poses a clinical challenge for both patient and the reporting doctor. Considering the time limitation for termination of pregnancy in our country, early prenatal diagnosis is important. A detailed face evaluation in the first trimester can help detect this defect as early as 11–14 weeks. Early diagnosis of lethal anomalies helps in completing the fetal work up and offering a safer termination. Correct diagnosis and work up of fetal anomalies allows for documentation and awareness of the presence of these conditions in our population.

Keywords Otocephaly · Anophthalmia · Agnathia · Microstomia · Synotia · Melotia

A 25-year-old primigravida 19 weeks of gestation was referred in view of micrognathia and polyhydramnios. Ultrasound at our centre revealed multiple facial anomalies including anophthalmia and agnathia. She opted for termination of pregnancy and the pictures of abortus taken post termination helped us to confirm our diagnosis of Otocephaly, a rare anomaly with reported incidence of 1 in 70,000 (Figs. 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13, 14).

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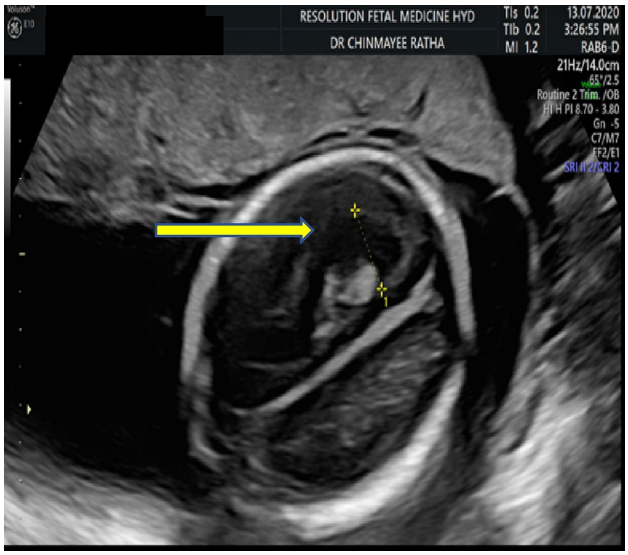

Fig. 1 Normal skull with mild ventriculomegaly. Skull is normal. Midline falx seen. Ventricle > 10 mm.

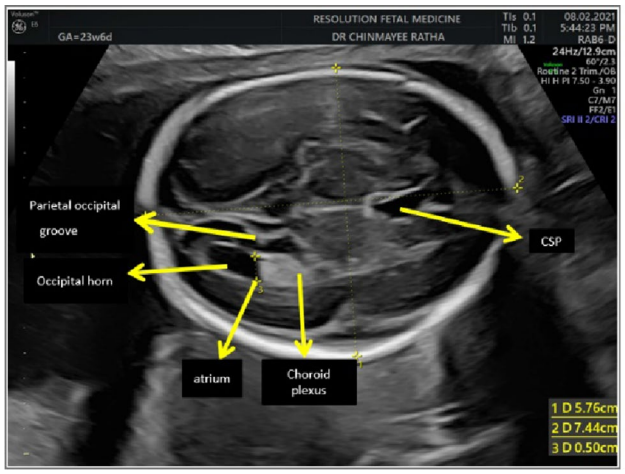

Fig. 2 Picture of ultrasonographic appearance of cerebral ventricle of a normal fetus is presented here for comparison.

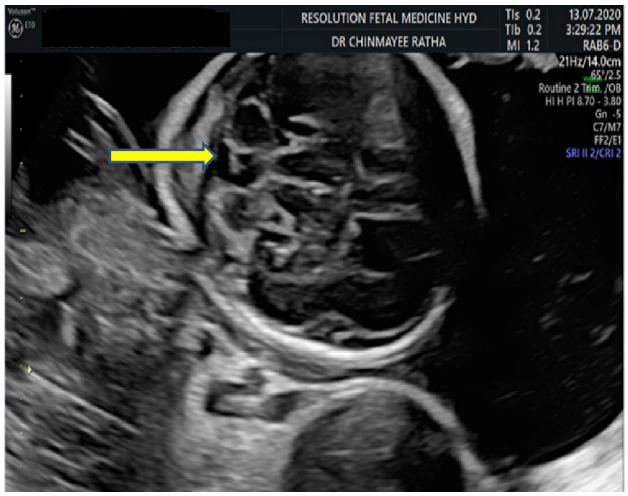

Fig. 3 Posterior fossa cyst (arrow) seen in the transcerebellar view.

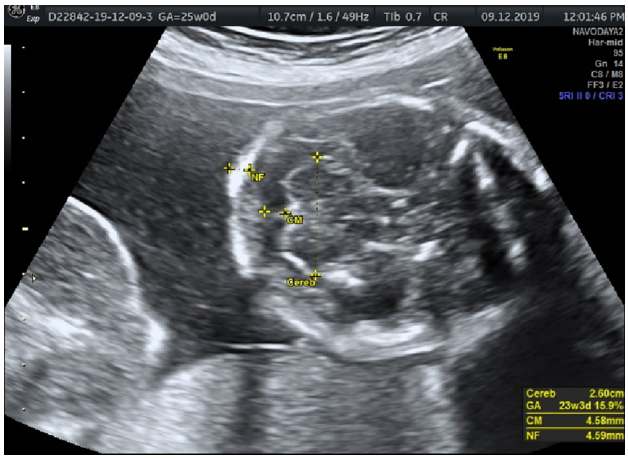

Fig. 4 Picture of ultrasonographic appearance of cerebellum and vermis of a normal fetus is presented here for comparison.

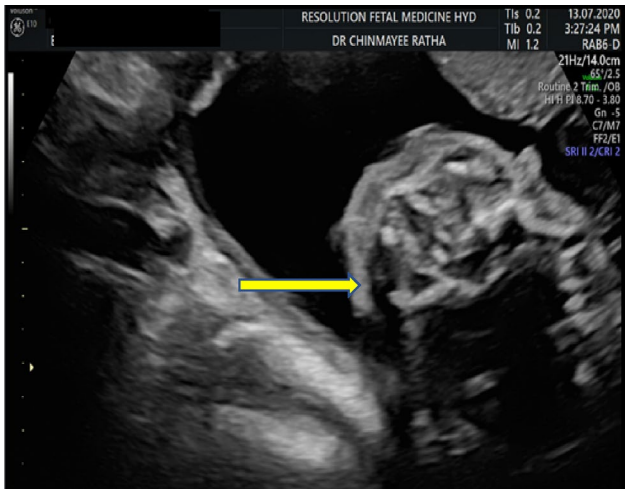

Fig. 5 Anophthalmia. The fetal orbits were not visible bilaterally, the nose was not visualised properly.

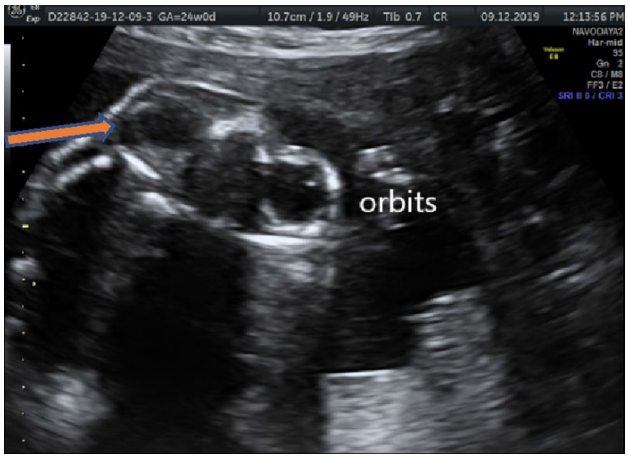

Fig. 6 Picture of ultrasonographic appearance of orbits with nasal protruberence in between them of a normal fetus is presented here for comparison.

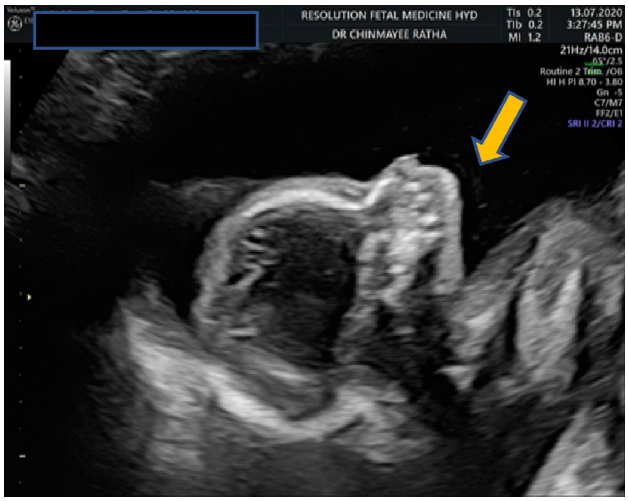

Fig. 7 Mandible is absent—agnathia. Yellow arrow shows the missing part as compared to the normal mandible seen in the smaller picture as shown by the red arrow in Fig. 8.

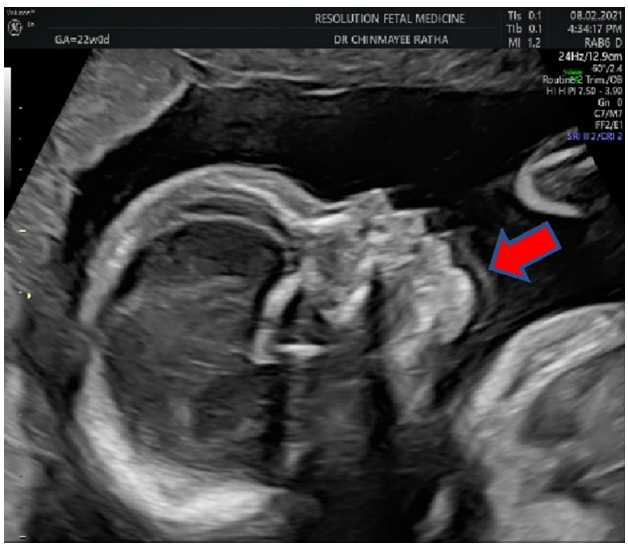

Fig. 8 Picture of ultrasonographic appearance of fetal facial profile with red arrow indicating mandible of a normal fetus is presented here for comparison.

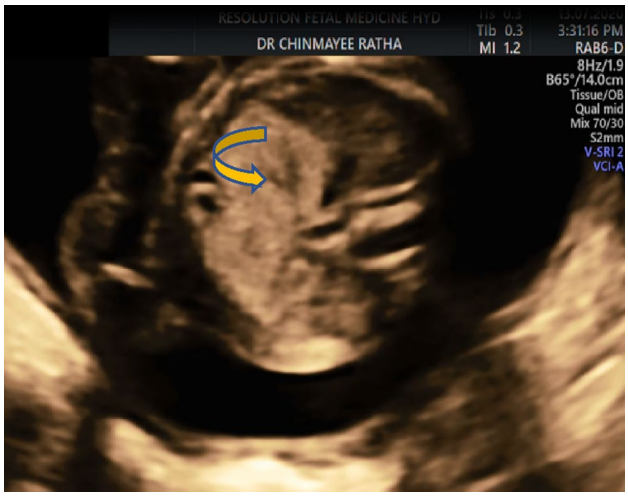

Fig. 9 Echogenic bowel as appreciated here can be diagnosed only by turning down the gains and switching the harmonics off. It is diagnosed if it is as echogenic as the bone (like the iliac bone).

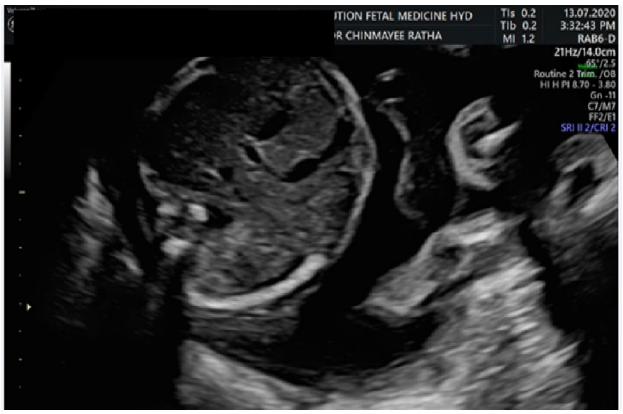

Fig. 10 Absent stomach bubble.

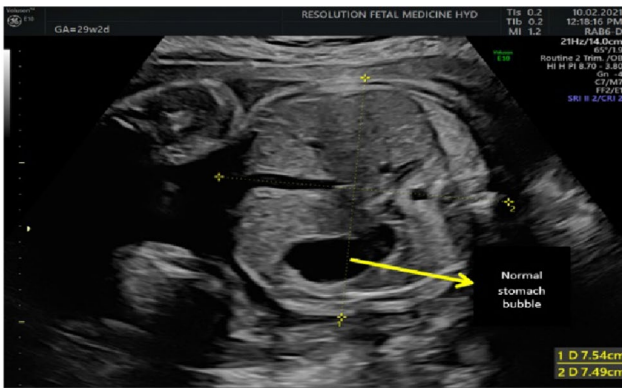

Fig. 11 Picture showing appearance of stomach bubble in a normal fetus (yellow arrow shows the normal appearance of a stomach bubble).

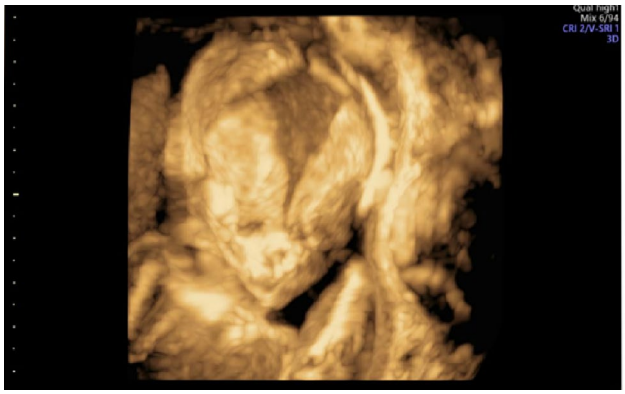

Fig. 12 3D picture: the orbits are not visualized, the nasal projection is not seen.

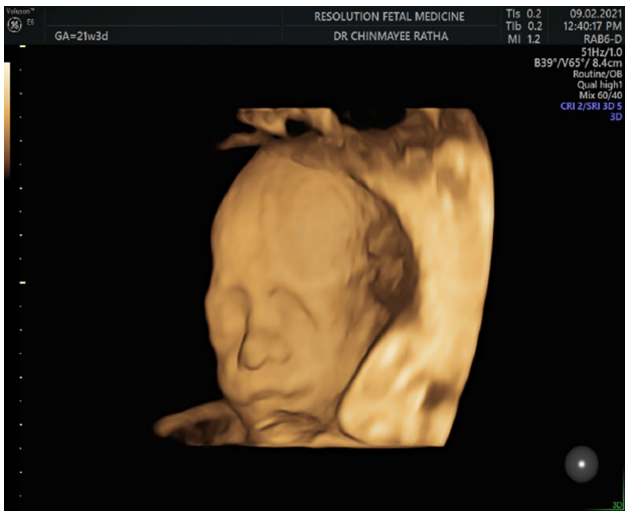

Fig. 13 Picture showing view of fetal face of a normal fetus at 19 weeks for comparison.

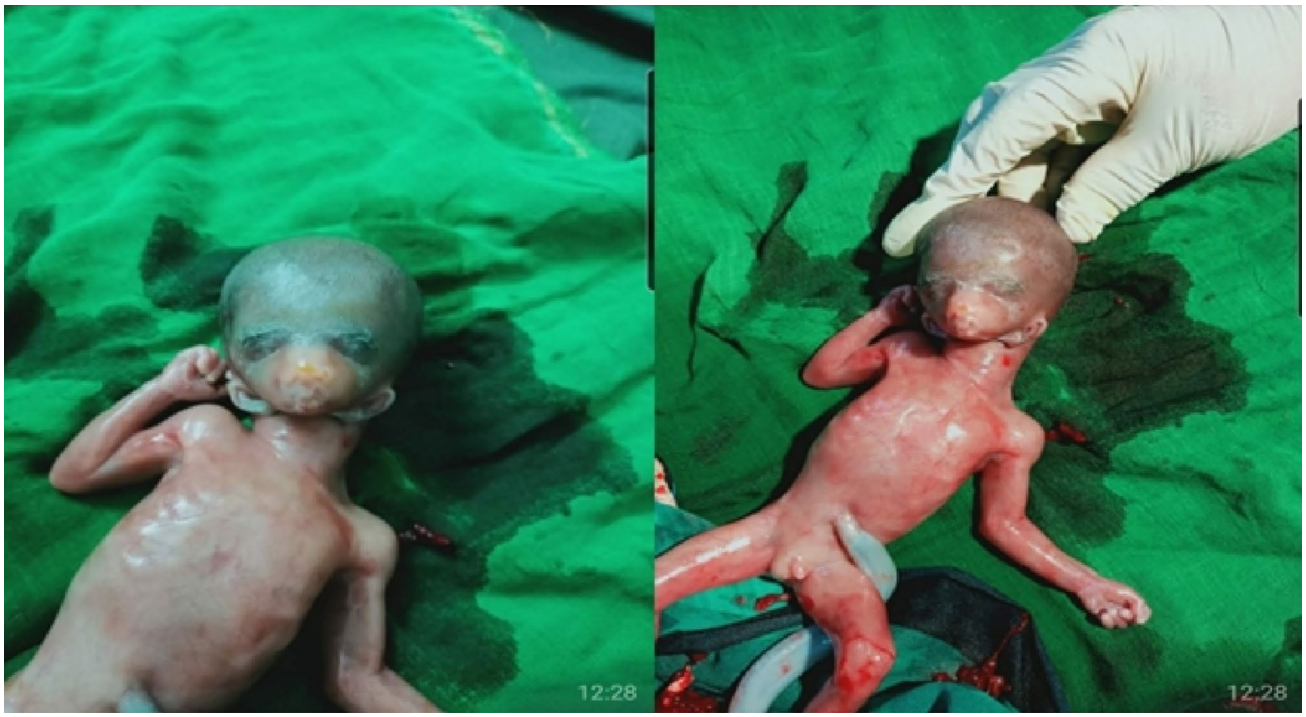

Fig. 14 Post termination picture. The picture shows low set ears almost meeting in the midline (synotia). Anophthalmia (absence of the orbits), microstomia (very small oral aperture), agnathia (absent mandible) and arhinia (absent nose).

Declaration

Conflict of interest The authors have no conflicts of interest or any vested interests to declare.

Consent Patient consent was taken for this publication.

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About the Author

Dr. Shreya Goenka Presently, a trainee for ICOG certificate course in Fetal Medicine. She has many academic achievements and is an enthusiastic obstetrician. She has authored chapters in various books and has been active in the academic events of the local Obstetric and Gynecological society. She has hopes of providing the benefits of fetal medicine to all pregnant women by training newcomers and wishes to specialize in fetal echocardiography.

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