Treacher -Collins syndrome
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CASE REPORT

ABSTRACT

INTRODUCTION
Treacher Collins syndrome (TCS) is a rare syndrome inherited as autosomal dominant. The affected children may vary in severity ranging from minimal features as slanting of palpebral fissures to major features of craniofacial development such as hypertelorism, micrognathia, maxillary hypoplasia, high arched palate, conductive hearing loss, external ear abnormalities and narrow nostrils. We report a case of Treacher Collins syndrome with term gestation with polyhydramnios and fetus with micrognathia on ultrasound examination with previous two babies and father having the same disorder.

Case Report
A 30 year old, unbooked G3 P2 L1 D1, postcesarean pregnancy, term gestation with polyhydramnios with breech presentation with features of TCS admitted for institutional delivery at Government General Hospital, Kakinada, Andhra Pradesh, India. Her scan on admission revealed single fetus in breech presentation with micrognathia and AFI 25cm. she delivered a live male baby weighing 2.2kg with features of TCS through cesarean section and inspite of airway support the baby died on first postnatal day.

Conclusion
This case report confirms the need to identify the antenatal women with physical malformations and early ultrasound scan in women with previous H/O anomalous children, and polyhydramnios so that termination can be planned early.

Key Words
Polyhydramnios , Treacher Collins Syndrome, Micrognathia, Anti mongoloid slant of eyes
ended in an emergency caesarean section for cephalopelvic disproportion and a live female baby of weight 3kg was delivered. This baby now aged about 3years is having facial anomalies like low set ears, maxillary and mandibular hypoplasia, right external auditory canal atresia and conductive deafness. Both the pregnancies were full term without complications in the antenatal and postnatal periods. In the present pregnancy she had two antenatal checkups and did not undergo any ultrasound scans till admission. On examination she was ill built and moderately nourished with antimongoloid slant of palpebral fissures, maxillary and mandibular hypoplasia, high arched palate, low set ears with complete left auditory canal atresia and partial atresia of right auditory canal with conductive deafness.

Systemic examination was normal. On abdominal examination the uterus was over distended with polyhydramnios and a single, mature fetus in breech presentation with fetal heart sounds 138/min. Haematological investigations and renal profile was normal. Ultrasound revealed a single live fetus of 35 wks with BPD 9mm, FL 6.7mm in breech presentation showing micrognathia. AFI 25cm and grade III placenta. Elective lower segment caesarean section was done. A live male baby weighing 2.2 kg with features of TCS was born and admitted in NICU with airway support. The baby died on 1st postnatal day due to respiratory problems. The patient’s father was also having the features of TCS.
DISCUSSION

TCS is an autosomal dominant disorder where 40% of the affected patients have previous family history and the remaining 60% of cases arising as a result of a denovo mutation. TCOF1 is the only gene currently known to be associated with TCS. It has been postulated that TCS represents a defect of blastogenesis that could be attributed to interference in cephalic neural crest cell histidifferentiation. Prenatal testing of the disease can be done by---

Molecular genetic testing: DNA analysis of fetal cells by aminocentesis at 15-18 weeks of gestation or chorion villous sampling at 10-12 weeks of gestation to detect TCOF1. Ultrasound: In pregnancies at risk finding of polyhydramnios, microcephaly and abnormal facial features is possible.

Preimplantation genetic diagnosis: in families where disease causing mutation has been identified in an affected family member.

In new born with TCS airway management is required because of narrowing of the airway and severe micrognathia. Neonatal death is usually associated with obstructive sleep apnea as a result of malformations. If after delivery, early diagnosis allows treatment to improve aesthetic appearance, it improves the social life of affected individuals.

CONCLUSION

This case suggests the importance of having awareness of physical abnormalities and early ultrasound scanning in affected members of TCS so that termination can be planned early.

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Please cite this paper as: Lavanya K Sarella, Prabodh K Dakey, Kamala C. Treacher -Collins syndrome. Inter J Medical Sci Res Prac 2014; 1 (3): 105-107.

ACKNOWLEDGEMENTS
Nil

PEER REVIEW

Double Blinded externally peer reviewed.

CONFLICTS OF INTEREST
Nil

FUNDING
Nil