ORIGINAL RESEARCH PAPER

Neonatology

KEY WORDS: Treacher Collins Syndrome Craniofacial development, neonates, rare disorder.

A CASE OF TREACHER COLLINS SYNDROME IN A NEONATE IN A TERTIARY CARE HOSPITAL IN ASSAM

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ABSTRACT

Treacher Collins Syndrome (TCS) or Mandibulofacial dystosis is a rare autosomal dominant disorder of craniofacial development. The congenital malformation is associated with dysmorphogenesis of first and second brachial arches. This condition affects an estimated 1 in 50,000 people. This article describes the clinical features of TCS in a neonate who was referred to the Department of Pediatrics.

INTRODUCTION

Treacher Collins Syndrome (TCS) or Mandibulofacial dystosis is a congenital disorder of craniofacial development that occurs with an incidence of 1 in 50,000 live births[1]. Dysmorphogenesis of first and second brachial arches due to interference during the development is deduced to be the cause of TCS[2]. Malar hypoplasia, mandibular hypoplasia, anti-mongoloid palpebral fissures, malformation of auricular pinna, coloboma of the lower eyelids and cleft palate are among the frequent clinical presentations[3].

CASE REPORT

A 2 day old neonate was seen in our emergency department as a case of birth asphyxia. Baby was referred from a primary health centre for respiratory distress.

History

The baby was born vaginally, at full term, birth weight was 2.6 kg. Baby did not cry at birth. Baby was resuscitated at the point of delivery. Baby was referred to our hospital at day 2 of age with complaints of respiratory distress. She was the first born child of her parents. Her parents had a non-consanguineous marriage. There was no history of exposure to known teratogenic agents or of maternal disease.

The baby was received in NICU. Baby had a respiratory rate of 75 per min, SpO_2 of 85% at room air. She had subcostal retractions; was put on oxygen inhalation by nasal prongs at 2L/min. Baby did not have convulsions and was started on tube feeding.

Physical Examination

Baby had a characteristic appearance with antimongoloid slant of the palpebral fissures, malar hypoplasia , malformed and small rudimentary ears (Fig.). Maxillae, mandibles and zygomatic bones were hypoplastic giving the baby a bird-like appearance .She had a tuft of hair extending upto the face. Baby did not have lid colobomas. The baby had a dolicocephalic head. Introrally, she had a high arched palate.
The baby had a patent airway, was gradually weaned off oxygen support. Systemic examination was unremarkable. Routine investigations including haemogram, liver and kidney function tests, Chest Xray were within normal limits. Xray PA view of the skull revealed hypolplastic maxilla and mandible with underdeveloped zygoma.

Based on the clinical and radiological findings, a diagnosis of Treacher Collins Syndrome was considered.

There was no family history of any similar phenotype and the case was thus considered to be that of sporadic origin.

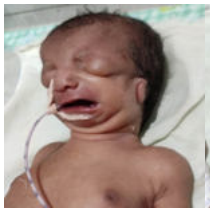

Fig. 1

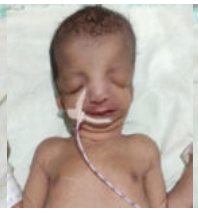

Fig. 2

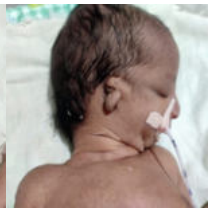

Fig. 3

Figures 1-3 showing the anti-mongoloid slant of the palpebral fissures, micrognathia, small rudimentary ears, extension of the tuft of hair upto face, dolicocephalic head and the bird like appearance.

DISCUSSION

TCS, alternatively called MFD, is an autosomal craniofacial development disorder related to the chromosomal region 5q32-q33.1, presenting typical facial aspects, clinical and genetic heterogeneity[4]. This syndrome was described by Thomson, Treacher Collins, Berry , and Franceschetti and Klein[5]. Dr. E. Treacher Collins, an ophthalmologist, described two children with malar hypoplasia; since then the condition has been named after him[5].

TCS exhibits autosomal dominant inheritance with variable penetrance[6]. "Treacle" (encoded by TCOF 1) is involved in ribosome biogenesis pathway, through its interaction with upstream binding factor (UBF) is central to neural crest cell proliferation and to normal cranio facial development. Mutations in the TCOF1 gene reduces the amount of treacle leading to depletion of neural crest cells in pharyngeal arches 1 & 2, thereby resulting in cranioskeletal hypoplasia[1,6,7].

40% of the cases are associated with previous family history while the remaining 60% of the cases are thought to arise as a de novo mutation[8]. Our case did not report with a familial history of the syndrome.

CONCLUSION

An affected parent of either sex will transmit the defect to 50% of offspings in accordance with Mendelian laws[8]. This emphasises the importance of genetic counselling to affected individuals and their families.

There is no cure for TCS. Treatment is aimed at the specific needs of each individual. Early detection and multidisc iplinary approach can produce excellent results for complete restoration of the form and function of the patient.

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Conflicts of interest
There are no conflicts of interest.

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