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Treacher Collins Syndrome rare disorder in neonate with respiratory failure- A Case Report

Dr. Garima Bansal[1], Dr. Varughese P.V.[1]

1. Department of Paediatrics, Christian Medical College, Ludhiana, Punjab

Abstract: Treacher Collins Syndrome is a rare genetic disorder in which first and second pharyngeal arch is affected leading to disfigurement of face. These children generally have normal development but a major social stigma due to facial changes.[1,2] This is a case report of neonate born with facial deformities sparing the rest of body parts with respiratory failure. He was managed symptomatically as well as multiple teams were involved for correction of deformities. Being a rare genetic disorder, there is a social limitation leading to delay and incomplete treatment, hence education and genetic counselling are important to reassure and correct the dysmorphism. It is important to guide the parents about this disorder for future planning.

Keywords: TCOF 1, POLR, MFD, TCS,

Abbreviations: MFD Mandibulofacial dysostosis; TCS Treacher Collins Syndrome; ABG arterial blood gas; HCO3 bicarbonate; ECHO Electrocardiography; PDA Patent Ductus Arteriosus; PFO Patent Foramen Ovale

INTRODUCTION

Treacher Collins is also known as Mandibulofacial dysostosis (MFD). It is composed of various genetic and clinically heterogenous disorders.[2] It is an autosomal dominant disorder with variable expression and high penetrance but can also be inherited as an autosomal recessive pattern as it seemed in this case report as both the parents were normal. It is caused due to mutation in genes TCOF 1, POLR1C, POLR 1D, POLR 1B depending on inheritance pattern.[3,4]

The commonly observed clinical features in MFD are generally bilaterally symmetric. It involves various parts of face such as: abnormalities of external auditory canal, hearing problems, lateral downward slanting palpebral fissures, coloboma of lower eyelids, lesser eyelashes, hypoplasia of facial bones, cleft palate.[5]

CASE REPORT

A neonate born to 28 year old G1P0 mother at 38 weeks 5 days of gestation by LSCS in view of Meconium stained amniotic fluid grade 3, un-booked pregnancy, born to non-consanguious marriage with no family history of similar disorder. He was born with Apgar 9,9 at 1 and 3 min respectively. In view of peripheral cyanosis, tachypnoea, distress and desaturation after delivery baby was intubated. On presentation he had HR 134 beats/min, RR 84/min, Temperature 97.8 F, RBS 112mg/dl.

His general physical examination showed facial dysmorphism with no other part involved. The clinical features present were anti-mongoloid palpebral fissure, lateral canthal dystopia, defective lower lid on right side along with hypertelorism. He also had B/L microtia grade 3 and non-visualization of external auditory canal with retrognathia, telecanthus and cleft palate (Figure 1, 2).

The complete blood profile at birth was normal. Random blood sugar was normal. ABG showed pH 7.376, pCO2 7.9, pO2 157, HCO3 4.5, SBE -21.2, Sat 99.9%. ECHO showed PDA with left to right shunt, PFO with left to right shunt, trivial TR (Right ventricular pressure 20mm hg) and normal Left ventricular contraction. Chest X-ray showed few infiltrates bilaterally.

He was started on ventilatory support, IV fluids, IV inotrope (Inj. Dopamine) and other supportive care. Diagnosis was made based on clinical findings and team approach. Underlying investigations for organ development and genetic counselling was planned. Family counselling was done and prognosis was explained. Further genetic counselling sessions were planned.


Figure 1

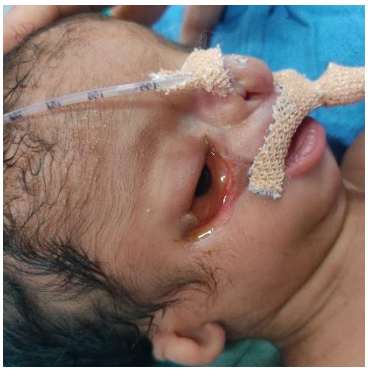
Figure 2

DISCUSSION

TCS requires a multi-disciplinary approach both pre as well as post operatively. It may require input from various health professionals to provide a better life. The primary management is to maintain airway by doing tracheostomy, which arise due to facial dysmorphism and respiratory tract involvement. Following this further facial reconstruction can be done such as for coloboma, microtia, cleft palate. Feeding can be introduced through feeding tube for proper nutrition till the correction surgeries are done. These children may require hearing aids based upon the involvement.[6,7]

This disorder requires multiple reconstruction surgeries which should be performed early in life for better social development of the child and to reduce stigma. The patient described in the case report is a neonate born with facial dysmorphism and breathing problems. It is important to look for underlying organs, genetic pattern and development of the child along with reconstruction surgery. Diagnosis is made based upon the clinical presentation and genetic testing. Due to high cost of genetic testing and social stigma this disorder is generally under treated.

Based on this case report, multi-disciplinary approach can give a better living. As the child can be given a better life with reconstruction surgeries, hearing aids and other supportive therapies it is beneficial to identify and treat early. Importance is of genetic counselling and awareness of the disease for future planning.

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