TREACHER COLLINS SYNDROME: A CASE REPORT

Jacob Antony Thoppil[1], Manish Kumar Tailor[2], Yogesh Aseri[3], Digvijay Singh Rawat[4], B. K. Singh[5], P. C. Verma[6]

[1]Junior Resident, Department of ENT, Jawaharlal Nehru Medical College, Ajmer, Rajasthan.
[2]Senior Resident, Department of ENT, Jawaharlal Nehru Medical College, Ajmer, Rajasthan.
[3]Assistant Professor, Department of ENT, Jawaharlal Nehru Medical College, Ajmer, Rajasthan.
[4]Assistant Professor, Department of ENT, Jawaharlal Nehru Medical College, Ajmer, Rajasthan.
[5]Senior Professor and HOD, Department of ENT, Jawaharlal Nehru Medical College, Ajmer, Rajasthan.
[6]Senior Professor and HOD, Department of ENT, Jawaharlal Nehru Medical College, Ajmer, Rajasthan.

ABSTRACT

BACKGROUND

Treacher Collins Syndrome is a congenital disorder of craniofacial development of the head and neck region. This is a case report of a patient who presented in our outpatient department with complaints of severe hearing loss, deformed face, external ears and malaligned teeth. With further evaluation, a diagnosis of Treacher Collins syndrome was made.

KEYWORDS

Treacher Collins Syndrome, Mandibulofacial Dysostosis, Conductive Hearing Loss, Coloboma Iris, TCOF1 Gene.

HOW TO CITE THIS ARTICLE: Thoppil JA, Tailor MK, Aseri Y, et al. Treacher Collins syndrome: a case report. J. Evid. Based Med. Healthc. 2016; 3(86), 4719-4722. DOI: 10.18410/jebmh/2016/994

BACKGROUND

Treacher Collins Syndrome[1-3] is an autosomal dominant disorder of craniofacial morphogenesis with high penetrance and variable expressivity.[1] The essential features of this syndrome were described by Treacher Collins, a British ophthalmologist, in the year 1900, but the first extensive description of the condition was given by Franceschetti and Klein in 1949 in which they coined the term mandibulofacial dysostosis.[1] It is estimated that the frequency of TCS is 1 in 50,000 live births.[2] Approximately, 60% of the autosomal dominant occurrences arise as de novo mutation.[4] Genetically, the treacle gene (TCOF1) is mutated. It is found on chromosome 5q31.3-32 and encodes a serine/alanine rich nucleolar phosphoprotein responsible for the craniofacial development.[1,2]

CASE REPORT

A 25-year-old female presented to our outpatient department with severe hearing loss, deformed face, external ears and malaligned teeth. Tuning fork tests were performed. A pure tone audiometry and BERA were done. These audiological investigations confirmed a case of severe conductive hearing loss. Further examination revealed downward slanting of eyes, depressed zygomatic arches, sunken cheek bones, deformed external ears (which were operated twice), coloboma of lower eyelids. There were no signs of mental retardation. Palate was high arched and uvula was normal. There was no history of similar complaints in the family.

A noncontrast high-resolution CT scan of skull and face clearly demonstrated maxillofacial deformity with prominent mandibular hypoplasia. Based on clinical and radiographical findings, she was diagnosed with TCS.

IMAGES

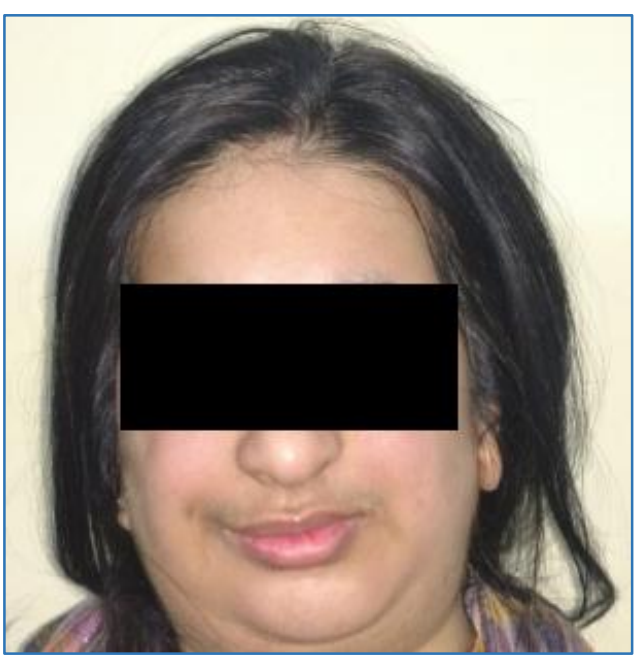

Image 1

Financial or Other, Competing Interest: None.
Submission 21-09-2016, Peer Review 28-09-2016,
Acceptance 11-10-2016, Published 27-10-2016.
Corresponding Author:
Dr. Jacob Antony,
Room No. 224, Residents Hostel,
Jawaharlal Nehru Medical College, Ajmer, Rajasthan.
E-mail: jacobbhaimbbs@gmail.com
DOI: 10.18410/jebmh/2016/994

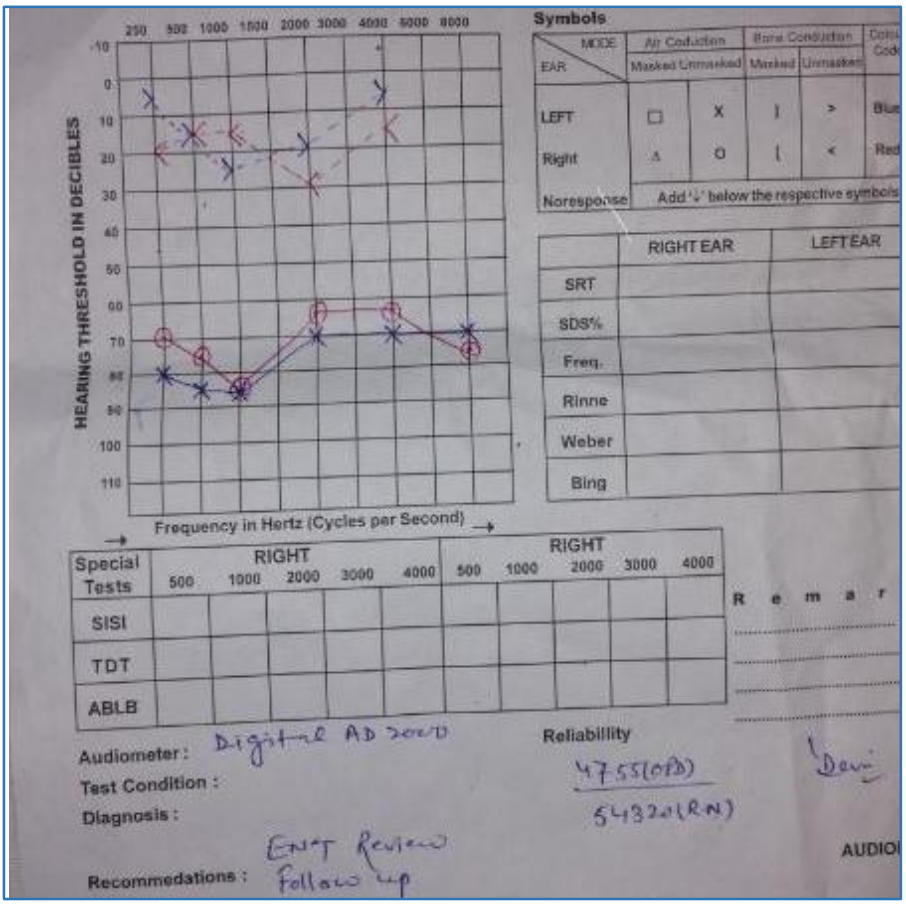

Image 2. (Audiogram)

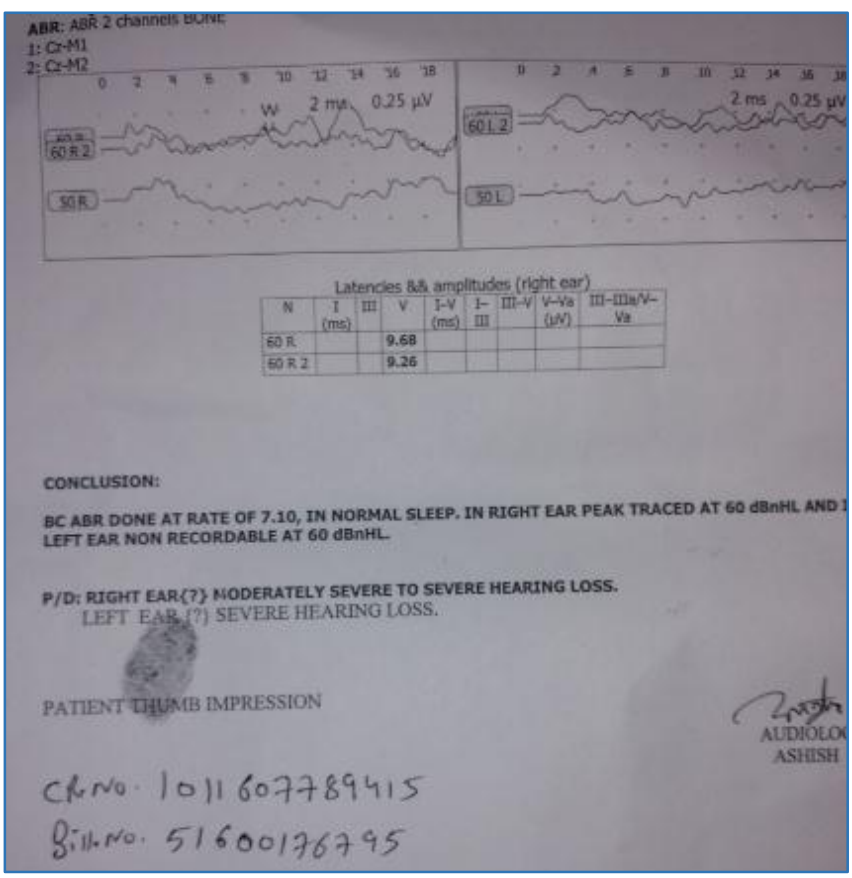

Image 3. (BERA)

DISCUSSION

Treacher Collins Syndrome (TCS)

Is a condition that affects the development of bones and other tissues of the face. The signs and symptoms vary greatly ranging from almost unnoticeable to severe. It is characterised by numerous bilateral symmetrical developmental anomalies derived from first and second branchial arches.[5,6] It is also called as Franceschetti-Klein Syndrome or mandibulofacial dysostosis. This was described for the first time by Thompson (1846).[5,7,8] However, the syndrome is named after Edward Treacher Collins, the English surgeon and ophthalmologist who described two cases in 1900. Treacher Collins syndrome is an autosomal dominant disorder with variable expression. More than half of all cases are thought to be new mutations because there is no family history of the disease.

TCS is caused by mutation in genes TCOF1 (>80% cases),[1,2] POLR1C, POLR1D. In some cases, the mutation is not detectable. Symptoms of this disorder vary greatly ranging from almost unnoticeable to severe.[9] It is most noticeably characterised by abnormalities of the head and face. These include down slanting eyes with notched lower lids,[2,10] sunken cheekbones[5,7] and jawbones, pointed nasal prominence,[11,12] broad mouth[5] and high-arched palate.[11,13] They also show malformation of the auricular pinnae[13] and conducting hearing loss[13,14] and preauricular hair extension.[13] A minority of those affected with TCS may have cleft lip and/or palate.[14]

Differential Diagnosis

- Oculoauriculovertebral dysplasia.
- Goldenhar's syndrome.
- Nager's acrofacial dysostosis (Similar Facial Features).[3]

Investigations

- The earliest possible diagnosis is by chorionic villus sampling (if there is a family history).
- Diagnosis may also be made at midtrimester antenatal ultrasound.[9]
- Postnatally, diagnosis is essentially made on clinical features. A thorough assessment must be made for all associated features especially those affecting breathing and complications, e.g. conductive hearing loss.

Management

The spectrum and degree of deformities are extensive and therefore the nature and intensity of management are also very variable.

- Affected children and their families may need a great deal of support.
- Hearing and speech: hearing aids and speech therapy.

Surgical

- In severe cases, the airway must be evaluated and secured from birth. Either positioning alone or tracheostomy is required to manage the airway and a gastrostomy required for feeding.
- Operative correction of cleft palate maybe necessary.
- Operations of choanal atresia or mandibular lengthening are performed at the age of 2 to 3 years or later.
- The timing of bone and soft tissue reconstruction will vary, but bone reconstruction should usually precede soft tissue corrections.
- Autogenous tissues, e.g. ribs or iliac bone should be used and synthetic materials avoided.
- Soft tissue reconstruction includes correction of lower eyelid coloboma and ear reconstruction.

CONCLUSION

By clinical and radiological evaluation, we came to the conclusion that this female is a case of Treacher Collins Syndrome. Most of the clinical findings of the patient matched with that of classical findings of Treacher Collins Syndrome. From otorhinolaryngology point of view, there was bilateral conductive hearing loss, which was confirmed using audiometry and BERA. But, findings like cleft palate, bifid uvula were not noted. The patient was operated twice for external ear deformity. The prenatal testing for mutation of the TCOF1 gene can reduce the incidence of TCS.

REFERENCES

1. Martelli-Junior H, Coletta RD, Miranda RT, et al. Orofacial features of Treacher Collins syndrome. Med Oral Patol Oral Cir Bucal 2009;14(7):E344-348.
2. Trainor PA, Dixon J, Dixon MJ. Treacher Collins syndrome: etiology, pathogenesis and prevention. Eur J Hum Genet 2009;17(3):275-283.
3. Marszalek B, Wojcicki P, Kobus K, et al. Clinical features, treatment and genetic background of Treacher Collins syndrome. J Appl Genet 2002;43(2):223-233.
4. Neville BW, Damm DD, Allen CM, et al. Text book of oral and maxillofacial pathology. 3rd edn. Philadelphia: WB Saunders Elsevier 2009:45-46.
5. Anil S, Beena VT, Ankathil R, et al. Mandibulofacial dysostosis: a case report. Aust Dent J 1995;40(1):39-42.
6. Madhan R, Nayar S. Prosthetic management of a patient with Treacher Collins syndrome. Ind J Dent Res 2006;17(2):78-81.
7. Dubey AK, Gupta RK. Treacher Collins syndrome: a report on two cases. MJAFI 2006;58:67-68.
8. Arvystas M, Shprintzen RJ. Craniofacial morphology in Treacher Collins syndrome. Cleft Palate Craniofac J 1991;28(2):226-230.
9. Farzana I, Afroza A, Rukunuzzaman MD, et al. Treacher Collins syndrome- a case report. Bangladesh J Child Health 2008;32(1):33-36.
10. Tay MT, Quah BL. Mandibulo-facial dysostosis--the eye signs of a case study. Singapore Med J 1991;32(5):365-367.
11. Dhayagude S, Parakh A, Ramchandran PA. Case report of a child with Treacher Collins syndrome posted for craniofacial reconstruction. The Indian Anesth Forum 2010;2:1-5.

12. Hertle RW, Ziylan S, Katowitz JA. Ophthalmic features and visual prognosis in the Treacher Collins syndrome. Br J Ophthalmol 1993;77(10):642-645.
13. Marres HA, Cremers CW, Marres EH, et al. Ear surgery in Treacher Collins syndrome. Ann Otol Rhinol Laryngol 1995;104(1):31-41.
14. Alexander, Sherry P. Treacher Collins syndrome. J Ind Acad Oral Med Radiol 2010;22:49-52.