TREACHER COLLINS SYNDROME: A CASE REPORT
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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.


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 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. It is also called as Franceschetti-Klein Syndrome or mandibulofacial dysostosis. This was described for the first time by Thompson (1846). 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), POLR1C, POLR1D. In some cases, the mutation is not detectable. Symptoms of this disorder vary greatly ranging from almost unnoticeable to severe. It is noticeably characterised by abnormalities of the head and face. These include down slanting eyes with notched lower lids, sunken cheekbones and jawbones, pointed nasal prominence, broad mouth and high-arched palate. They also show malformation of the auricular pinnæ and preauricular hair extension. A minority of those affected with TCS may have cleft lip and/or palate.

Differential Diagnosis

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

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.
- 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

