TREACHER COLLIN SYNDROME

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Treacher-Collins syndrome is one of the rare craniofacial deformities. Most of the cases of this deformity have been reported in continental and French literature. One case of Treacher-collins syndrome was reported in Indian literature (Kumar and Sood 1962) hence the case is being reported.

Case History

Patient a 2½ years female child, presented in the Plastic Surgery outpatient department of L. L. R. & Associated Hospitals, Kanpur for the deformity of face and ears.

On examination it was noted that the child had an average built. She had antimongoloid slant of the palpebral fissures. The palpebral fissures were small. Notching of the outer third of lower lid was present. Eye lashes were absent in the lower eye lid. Both the zygomatic arches were underdeveloped. The mandible was also underdeveloped. External auditory meatus could not be seen but the external auditory canal was palpable. Malformation of both the external ears was present. She also had macrostomia, a high arched palate and abnormal dentition.

She was the 3rd child of her parents. She was born premature. The other two children of the parents, one male and one female were absolutely normal. There is no family history of any congenital abnormality on either sides of the parents. The child was weak at the time of birth and had delayed milestones.

Fig. 1  Front view

Fig. 2  Side view.

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Discussion

Treacher-collins syndrome is reported in the literature under a number of names—Mandibulo-facial dysostosis, Fischhochrosis klein syndrome and Berry syndrome. The first historical reference to this entity was published in 1889 by Berry and Treacher-collins reported two case 11 years later. It is peculiar that the entity is named after a person following a case report.

Rogers (1964) analysed 200 cases of this syndrome and did not suggest any statistically significant higher incidence of the syndrome in either male or female. His figures do suggest that the syndrome may be genetically transmitted primarily by the female of the affected line whereas in our case the family history is negative for any such craniofacial deformity. The complete-fually developed syndrome consists of all or most of the following features:

1. Antimogoloid slant of the palpebral fissures.
2. Notching or colobomas of the lateral portion of the lower lids with deficient or absent eye lashes of the medial two thirds or four fifths of the lower eye lid.
3. Hypoplasia or underdevelopment of the facial bones especially of the malar bone and the mandible.
4. Bilateral deformity of the ears, usually microtia, with or without an external auditory canal. In some cases absence of ossicles resulting in deafness.
5. Macrostomia, a high arched palate and abnormal dentition.
6. Blind fistula or dimples between the ears and the angle of the mouth.

The case under report had most of the features constituting the syndrome namely, beak shaped nose, antimongoloid eyes with notching of lower eyelids, absent cilia, hypoplasia of zyomatic arches micrognathia, microtia, macrostoma with high arched palate and abnormal dentition.

The familial incidence of this syndrome has also been reported by Zwemeliller and Stur. In our case the family history is not suggestive of any familial incidence. The patient belonged to an upper middle class so nutritional factors which may be responsible for it cannot be accepted. Warkany (1956) and DeUsche (1955) demonstrated that riboflavin deficiency produced dentofacial anomalies particularly micrognathia in rats.

Summary

A case of treacher-collins syndrome (Mandibulo facial dysostosis) is reported.

References:

