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CASE REPORT

GOLDENHAR SYNDROME: A RARE CASE REPORT

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ABSTRACT

Goldenhar's syndrome, also known as oculo-auriculo-vertebral (OAV) spectrum, is a rare condition described initially in the early 1950's by Maurice Goldenhar. It is a complex syndrome characterized by an association of maxillomandibular hypoplasia, deformity of the ear, ocular dermoid and vertebral anomalies. The etiology of this rare disease is not fully understood, as it has shown itself variable genetically and of unclear causes. Here, we describe a 13-year-old male patient with classical signs of the syndrome as pre-auricular tags, microtia, kyphoscoliosis, polydactyly and cleft lip.

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INTRODUCTION

Goldenhar syndrome was described by Maurice Goldenhar as a triad of accessory tragic, mandibular hypoplasia and ocular dermoids (Saxena, 2012). The syndrome encompasses a range features, but the craniofacial features are highly characteristic and make the role of an oral physician very important in the diagnosis of such syndromes. The syndrome consists mainly of mal-development of the first and second branchial arches (Martelli-Junior, 2010). Many authors consider the presence of ear anomalies (microtia and tissue tags) necessary for diagnosis (Adeoye, 2002). Additionally, dentofacial anomalies like facial asymmetry or facial and/or mandibular hypoplasia, dermal epibulbar tumors, palpebral alterations, vertebral anomalies, lateral facial clefts, cardiovascular and renal problems are observed (Pinheiro, 2003).

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1:3500 to 1:5600 incidence of the syndrome is reported. Male to female ratio of 3:2 is seen with male predilection (Tuna, 2011). No geographic or racial predilection is seen as such. Abnormalities are usually unilateral in 85% of cases and bilateral in 10–33% of the cases. The right side is more frequently affected (Gaurkarm, 2013). The etiology of the syndrome is not fully understood with most cases being sporadic (Lakshman, 2017). However, it is possible that abnormal embryonic vascular supply, disrupted mesodermal migration or some other factor leads to defective formation of the branchial and vertebral systems. 1-2 % cases have also been associated with genes autosomal dominant being more common (Jena, 2007). The diagnosis of Goldenhar's syndrome is mainly based on the clinical and radiologic findings (Reddy, 2016). So, here we report a case of a 13 – year old boy who came to our department with microtia, kyphoscoliosis, polydactyly and operated cleft lip which prompted us to make a diagnosis of goldenhar syndrome.

Case Report

A 13-year-old male patient reported to the Department of Oral Medicine and Radiology with the chief complaint of pain in lower right back tooth region of jaw since 4 days. Patient gave history of similar complaint 1 month back but the pain was relieved on taking medications. He was born to unconsanguineous parents. His elder sister was normal. Mother reported an uncomplicated pregnancy on questioning. The patient gave history of difficulty in hearing from the left ear since childhood. He also had also undergone surgery for cleft lip in childhood. On general examination, the built of patient was asthenic. He had a short stature. Patient also had kyphoscoliosis and polydactyly with right thumb which was asymptomatic. A review of other systems appeared to be normal. Mental IQ was also normal.

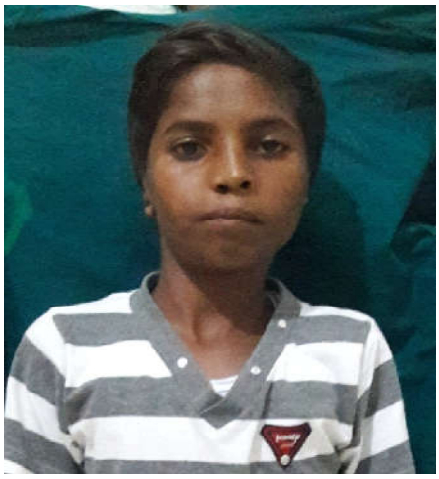
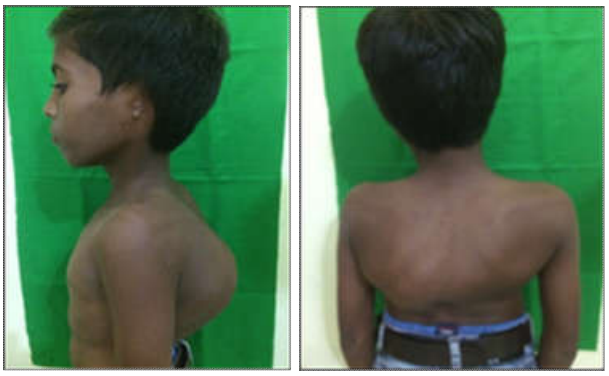


Fig. 1. Face profile showing scar of operated cleft lip and facial symmetry



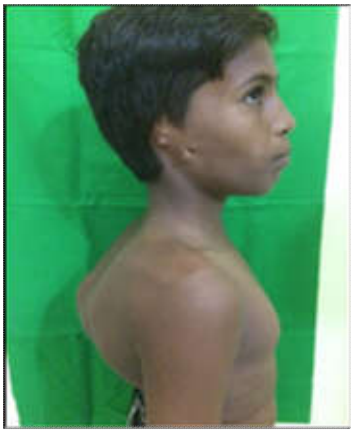


Fig. 2. Kyphoscoliosis and microtia

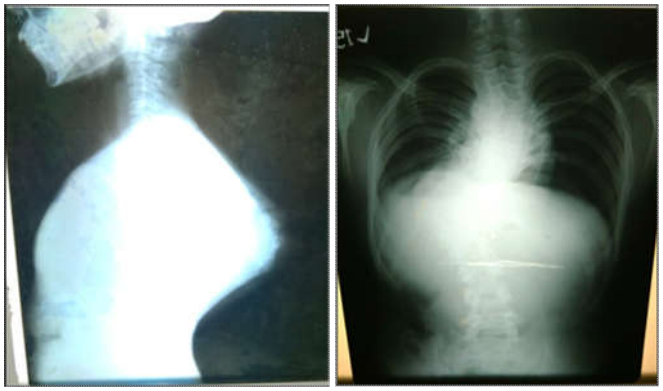


Fig. 3 a. lateral chest X-ray b. AP chest X-ray

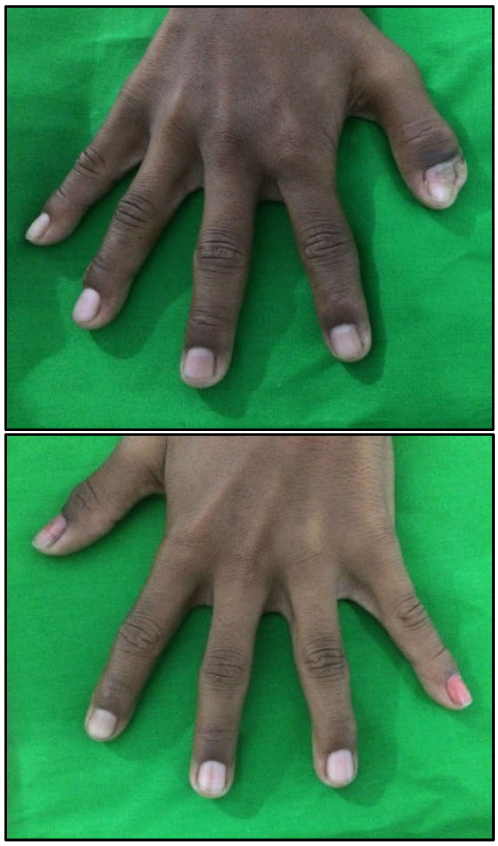


Fig. 4 a. Right hand –attempt in division of right thumb b. left hand

Both the ears were absent (microtia) and tissue tags were present in its place. Only a small 1 mm opening could be seen for right external auditory canal. He had a hearing defect in left ear. On extra oral examination the face was bilaterally asymmetrical. Also hypertelorism could be appreciated. We could see a scar of operated cleft lip in philtrum area. Also a single right submandibular lymph node was palpable roughly round, 0.5 cm in size, soft, non-tender and mobile. Intraoral examination revealed deep occlusal caries with 46 with tenderness on vertical percussion positive. Also there was occlusal caries with 36. There was severe crowding with maxillary anteriors and over-retained 53. Crowding could also be seen with lower anteriors. Panoramic view revealed occlusal caries with 46 with periapical rarefying osteitis. The postero-anterior and lateral chest radiographs confirmed the kyphoscoliosis. The hand – wrist radiograph showed the bony division of the right thumb. The chief complaint of the patient was adhered to by doing root canal treatment with 46. Also a crown was given with 46 and restoration was done with 36.

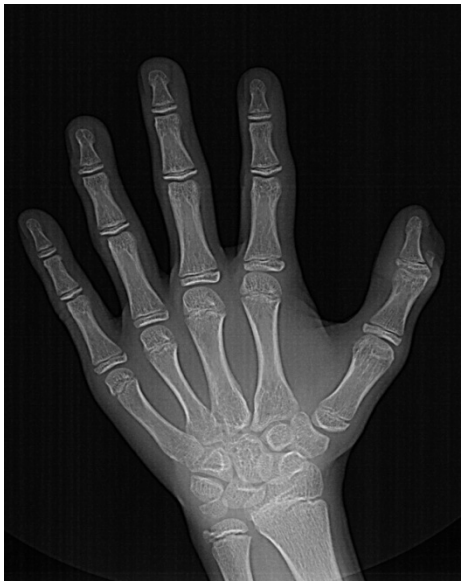


Fig. 5. Radiograph of right wrist there is attempt in division of right thumb

The patient was also sent for ENT evaluation. He was advised with ear prosthesis. But due to poor socioeconomic backgrounds patient refused for the same and also for further investigations. His consent was taken for photographs, clinical examination and radiographic evaluation. Then the patient was lost to follow-up. Hence on the basis of clinical and radiographic evaluation, a diagnosis of goldenhar syndrome was done.

DISCUSSION

Goldenhar syndrome was first described by a Swiss ophthalmologist, Maurice Goldenhar in 1952. The study of this condition is still controversial because of its complexity and broad clinical aspects. Many physicians consider presence microtia as an essential part of diagnosis (Pinheiro, 2003). Our patient had microtia with pre-auricular tissue tags bilaterally. Type of Preauricular appendices and microtia vary but presence is a considered as a rule for diagnosis. But bilateral occurrence is seen only in 30% cases (Pinheiro, 2003). The patient could not hear from left ear because of missing external auditory meatus similar to high prevalence of same reported earlier. Patient also showed kyphoscoliosis as seen in 35-60% of cases (Adeoye, 2002). Fortunately for the patient, it was asymptomatic. Patient also had polydactyly which is again a characteristic feature associated with this syndrome (Alasti, 2009). Although the patient gave history of an operated cleft lip, this alteration is observed in about 5% of the cases. Despite the reported frequency of cardiovascular alterations ranging from 5 to 58%, in this patient no cardiovascular alterations were found (Pinheiro, 2003). There was no family history of same and pregnancy was also uneventful suggesting the occurrence to be sporadic (Saxena, 2012). Our patient was provisionally diagnosed as Goldenhar syndrome but a number of other first and second arch syndromes were considered in the differential diagnosis (Alasti, 2009).

- Klippel – Feil Syndrome – there is presence of peculiar cup - shaped pinna
- CHARGE Syndrome – there were no heart, renal or genital defects in our patient.
- Nager Syndrome – here the patients have short neck due to missing cervical vertebrae as against kyphoscoliosis in our patient
- LADD Syndrome - (lacrimo-auriculo-dento-digital syndrome) the patients present with fused digits rather than polydactyly also the lacrimal glands were normal in our patient.

Treatment of Goldenhar syndrome is very much speculative. It requires a multidisciplinary approach. It is mainly based on the presence of the symptoms. In those with hearing impairment, hearing aids or other treatments may be recommended. Craniofacial and dental abnormalities may require surgical repair. Distraction osteogenesis along with functional orthodontics has successfully been tried in growing age. Plastic surgery to fix the jaw, cheeks, and ears can also be done (Saxena, 2012).

Conclusion

Taken together, our findings indicate that Goldenhar syndrome is very much a clinical entity more than elaborative diagnostic tests with oral diagnostician playing an important role. The treatment of deformities can be done by multiple procedures which require a multidisciplinary team. Also long-term follow-up is important to monitor the growth and development of the patients.

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