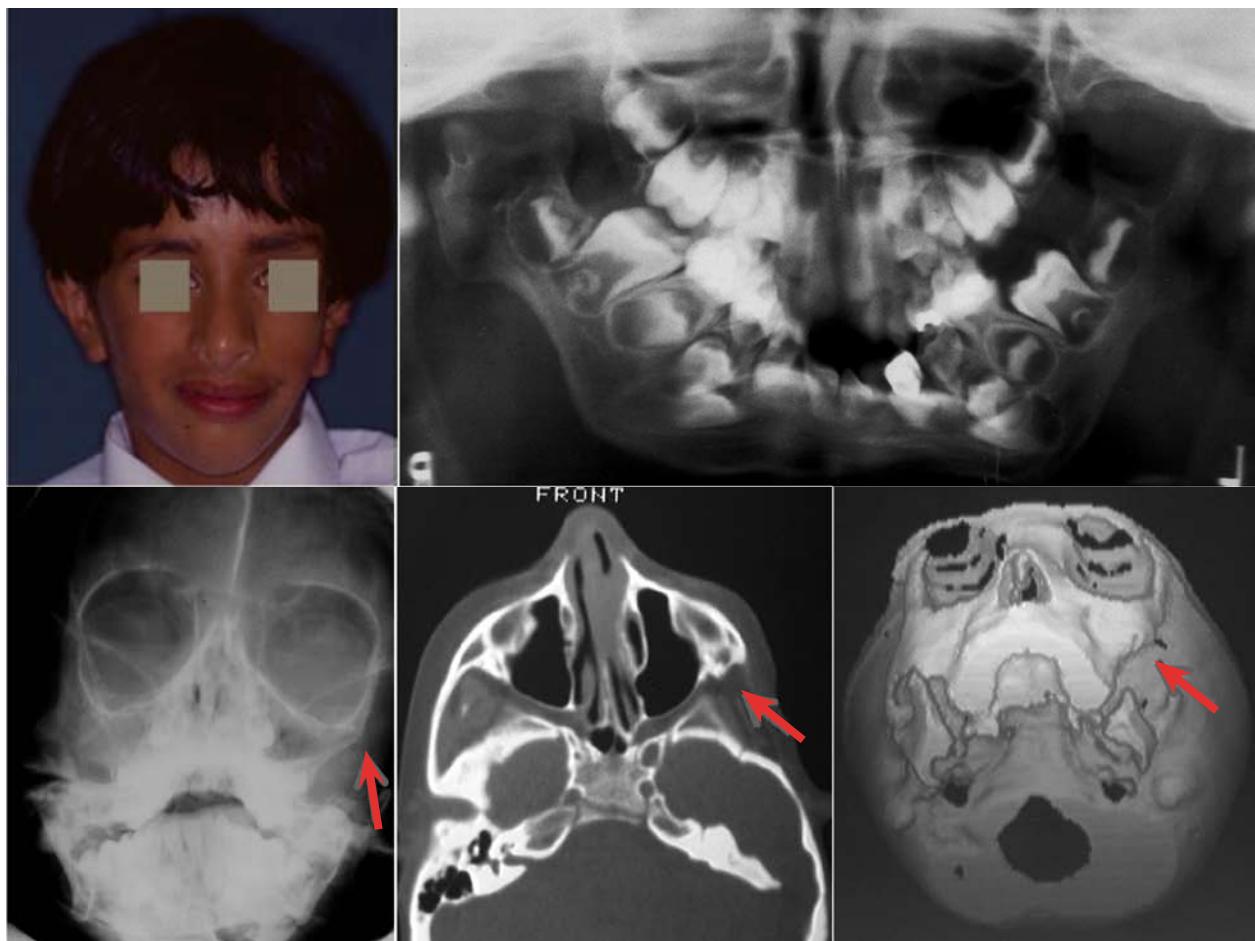


Maxillo-facial radiology case 123

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CJ Nortjé

This six year old patient presented at the Faculty with the main complaint that he was not happy with his facial appearance. His medical history indicated that he had almost complete atresia of his left ear and complete absence of the middle ear, resulting in total deafness on the left side. He had palsy of the temporal branch of the facial nerve, which resulted in an inability to close the left eye. What are the most important radiological features and what is your diagnoses?



INTERPRETATION

The pantomograph show bilateral mandibular hypoplasia with very short rami and an absent zygomatic arch on the left side (arrows). A diagnosis of Mandibulofacial Dysostosis was made. Mandibulofacial syndrome is a group of head and face malformations, most often hereditary in pattern and generally dominant in transmission. The condition is thought to result from failure in differentiation of maxillary mesoderm at the 50mm stage of intrauterine development (i.e., at about 2 months). The syndrome was first described by Thomson in 1847, and is also described as the Treacher Collin's syndrome. The major components of the syndrome are: absence or deficiency of the medial eyelashes, abnormalities of the external and middle ear, hypoplasia or agenesis of the malar bone and macrostomia. The palate is noted to be high or cleft in over 40 percent of the patients and the mandible is almost always hypoplastic. Blind fistulae may be present between the angles of the ears and the commissures of the lips and arid, atypical tongue-shaped processes of the hairline may be seen extend toward the cheeks. The lower border of the body of the mandible is often pronouncedly concave. Because of poor development of the maxilla, the mandibular hypoplasia and the high or cleft palate, dental malocclusion is frequent. The characteristic facial appearance is described as "birdlike" or "fishlike." The cranial vault is normal and most patients have normal longevity. Differential diagnoses, Goldenhar syndrome and Apert syndrome.

Reference

1. Farman AG, Nortjé CJ & Wood R E: Oral and Maxillofacial Imaging, 1st Ed, Mosby. St. Louis, Missouri, 1993,129-131
2. McKenzie, J. & Craig, J.: Mandibulo-facial dysostosis (Treacher Collins syndrome). Arch. Childh. 1955;30 391-395