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Case Study
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A Rare Case of Crouzon Syndrome with Choanal Atresia

Gautam Bir Singh and Devanshu Kwatra*

Department of Otorhinolaryngology, Lady Hardinge Medical College & Associated Hospitals, India

Abstract

A rare case of crouzon syndrome with choanal atresia is pictographically depicted. With this case, we illustrate a rare entity that present in an extremely rare manner.

Keywords: Crouzon syndrome; Choanal atresia; FGFR2

Case Discussion

Crouzon Syndrome (CS) is a rare genetic autosomal dominant disorder (prevalence rate: 1/25000 live births) first reported by French Neurosurgeon Octave Crouzon in 1912 [1]. It is caused by mutation in the Fibroblast Growth Receptor 2 [FGFR2] gene [1,2]. CS is characterized by craniosynostosis. The major clinical features are brachycephaly, ocular proptosis, an underdeveloped maxilla, midface hypoplasia and mandibular prognathism [1,2]. CS is distinguishable from other craniosynostosis syndrome like: Pfeiffer's, Apert, Carpenter and Jackson-Weiss etc. by lack of hand/or foot abnormalities [2].

This case (2 months male child) of CS presented with bilateral Choanal Atresia (CA) (Figure 1 and 2). CA is an uncommon occurrence in pediatric patients (incidence 1 in 5000 to 9000 live births) and is rarely seen in CS [3]. As CA associated with CS can cause obstructive sleep apnea and respiratory obstruction leading to cor pulmonale and death, it should be managed aggressively [3,4]. This case was managed successfully by endoscopic surgical resection [3].

Figure 1: Clinical photograph of the patient [stents in nasal cavity] with his father [also having Crouzon syndrome].

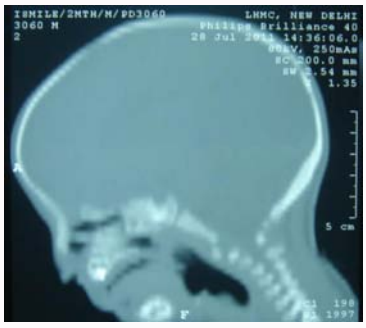

Figure 2: CT scan of the patient showing choanal atresia [Saggital section].

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Correspondence:
Gautam Bir Singh, Department of Otorhinolaryngology, Lady Hardinge Medical College & Associated Hospitals, Shaheed Bhagat Singh Marg, New Delhi -110001, India, Tel: +91-95129-4012368/4007550; Mobile: +91-9818836242;
E-mail: gbsnit@yahoo.co.in

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