

Case Report

Congenital Absence of Uvula: A Rare Case Report

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Abstract

The uvula is a fleshy projection hanging at the back of the throat from posterior margin of the soft palate. Congenital absence of uvula is a very rare entity in general population. Congenital absence of uvula has been seen to be associated with other congenital abnormalities such as cleft palate, various genetic conditions, and immunodeficiencies states. This case report concerns a 35-year old female, who on incidental finding was diagnosed with neither congenital absence of uvula without any other abnormality nor a history of uvulectomy. We report this rare case of congenital absence of uvula in a patient with no other known abnormalities.

Why this case?

There is very limited literature on congenital absence of Uvula. It may co-exist with various genetic abnormalities pre-disposing the patient to a potentially difficult airway in Emergency Department. It can also lead to ambiguity in airway assessment using modified Mallampati (MMP) Class, classification.

Keywords: Uvula; Congenital absence; Abnormalities

Introduction

Uvula is a soft flap of tissue that hangs down at the edge of the soft palate. It is formed by the fusion of the palatine shelves during the 7th to 12th week of intrauterine life [1]. The uvula has two main functions: Firstly during swallowing, the soft palate and the uvula move together to close off the nasopharynx and prevent food from entering the nasal cavity. It has also been proposed that the large amounts of thin saliva produced by the uvula serve to keep the throat well lubricated and have role in speech [2]. The congenital absence of the uvula is very rare in general population. There are only few case reports of this entity in medical literature [3,4].

Case Presentation

Thirty-five years old women came to our emergency department with chief complaints of fever and dry cough for the last three days. On examination, patient was conscious and alert, vitals were stable and systemic examination was within normal limit. On examination of the oral cavity for sore throat, it revealed absent uvula. On further probing the patient regarding the same, she gave the history of absence of this organ since birth and thorough examination did not show any additional deformity like cleft palate in the oral cavity. However, it showed slight congestion of posterior pharyngeal wall and with intact gag reflex tested by using tongue depressor. There was no scarring in soft palate to provide any evidence of uvulectomy. The patient had no significant past medical history, no recurrent throat infection, otitis media, snoring or airway surgery. He had no history of immune deficiencies, or dysmorphic features.

Discussion

The cause of absent of uvula is classified into congenital and acquired. The congenital absence of the uvula is infrequent in the general population. Jorgenson et al. reported only one case of the absent uvula in 2258 oral examinations in neonates [5]. Congenital absence of uvula can be associated with genetic conditions such as Apert syndrome, anhidrotic ectodermal dysplasia, cerebrocostomandibular syndrome, and hyperimmunoglobulin E syndrome [6]. These genetic conditions may be associated with factors causing difficulty in airway management in emergency department such as rheumatoid arthritis is associated with anhidrotic ectodermal dysplasia, fusion of cervical vertebra and cleft palate in Alpert syndrome and micrognathia, palate malformations and glossoptosis in hyperimmunoglobulin E syndrome [5,6]. Bifid or absent uvula is associated with non-Syndromic isolated cleft palate also has been reported in the literature [7]. An acquired absent uvula may be secondary to surgery or cultural practices of removing uvula for chronic cough and throat infection as in Sub-Saharan Africa. It is the diagnostic signs of rhinoscleroma, caused by *Klebsiella pneumoniae* spp. *rhinoscleromatis* and if found early, it is treatable [8].

But in our patient, there was no associated cleft palate or cleft lip also patient denied any history of oral surgery, or recurrent respiratory infection. It is very rare to see the congenital absence of uvula without any other associated congenital anomaly.

Importance in the Emergency Department: Absent uvula is rare in the general population, and the cause should be looked for, as it may co-exist with various genetic abnormalities pre-disposing the patient to a potentially difficult airway. Thus, emphasis should be given to visualization of other oropharyngeal structures while managing the airway in ED.

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Figure 1: Oral cavity with an absent uvula, no scarring to provide any evidence of uvulectomy.

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