

# Congenital Absence of Uvula: Case Report

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## Abstract

Traditional Uvulectomy has been an age-long practice throughout Africa and certain parts of the Middle East. It is performed as a treatment to prevent throat infections for centuries and is carried out usually during infancy and childhood. Uvula is a fleshy projection hanging at the back of the throat from the posterior margin of the soft palate. Congenital absence of uvula has been observed to be associated with other congenital abnormalities such as cleft palate. This case report concerns a 26 year old male from Nigeria, examined at a health fair conducted at All Saints University School of Medicine in Dominica who on an incidental finding was diagnosed with neither the absence of uvula without any other abnormality nor a history of uvulectomy. To the best of our knowledge, we report the first case in medical literature of the congenital absence of uvula in a patient with no other known abnormalities.

**Keywords:** Uvula; Congenital absence; Congenital abnormality; Traditional Uvulectomy; All Saints University School of Medicine; Dominica; Health Fair.

## 1. Introduction

The uvula is the small teardrop-shaped projection that hangs from the posterior free margin of the soft palate. The uvula has many functions; it helps prevent the soft palate from being forced into the nasopharynx, and resists pressure differences between the mouth and oral part of the pharynx as in coughing or sneezing [1, 2].

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It is formed by the fusion of the palatine shelves during the 7-12<sup>th</sup> week of intrauterine life [1, 2]. It also plays an avid role in the expression of the gag reflex as the uvula is innervated by the vagus nerve (cranial nerve X) which supplies the motor activity of gagging whilst the glossopharyngeal nerve (cranial nerve IX) carries the sensory innervation [3]. Many also believe that the uvula assists with speech as it produces a salivary secretion that lubricates the throat while talking. The glossopharyngeal nerve also helps with palatal movement as it innervates the stylopharyngeus muscles, damage to this nerve may result in a complication called glossopharyngeal palsy [3].

## **2. Case report**

A 26-year-old Nigerian male visited the health fair for a regular checkup with the complaint of sore throat since one week. Upon first impression the patient was alert and had no difficulty in speech. Intraoral examination of the patient by lowering the tongue using a tongue depressor revealed slight erythema and swelling of the palatopharyngeal arches and complete absence of uvula was observed (Figure 1). The patient denied any sort of oral surgery including uvulectomy. The patient admitted to previously drinking but had stopped 2 years ago. He also admitted to smoking marijuana a couple of times in his earlier years of life. The remaining history and physical examination was unremarkable.

Physical examination results:

Blood pressure: 126/84 mm Hg.

Pulse: 88 beats/minute

Weight: 70 kg, Height: 179 cm

Body Mass Index (BMI): 21.8 (Normal weight: 18.5 - 24.9)

Oral Examination:

Palatopharyngeal arches display erythema and swelling on inspection.

Gag Reflex positive using tongue depressor.

## **3. Discussion**

There have been common reports of bifid and trifid uvulas along with cleft palates [4]. This is due to the failure of the merging process in the soft palate during intrauterine life [5]. The congenital absence of the uvula is still yet to be investigated. In many parts of the world uvulectomies are performed as traditional surgical practices. This practice is well documented in African countries such as Kenya, Sierra Leone, Tanzania, Ethiopia and Nigeria during ethnic identification rituals [6]. The patient denied any involvement with this traditional practice of uvulectomy. The uvula plays an important role in the gag reflex, being innervated by both the glossopharyngeal nerve and vagus nerve. When the gag reflex was tested on the patient, it showed positive

response for the reflex confirming the integrity of both the glossopharyngeal and vagus nerve. The glossopharyngeal nerve also innervates the muscles of the oral pharynx; this was examined by observing the symmetric elevation of the soft palate when the patient said “ah”, no abnormality was present [3].



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

#### 4. Conclusion

Most of the congenital absence of uvula cases are accompanied with other abnormalities such as cleft palate or cleft lip [5]. This case study is unique because this patient does not only have any other abnormality but also comes from a community where uvulectomy has been a common practice for centuries. It can be concluded that information regarding congenital absence of the uvula can be of high importance not only from an embryological stand point but also from an evolutionary aspect. We recommend that further research accounting for the prevalence of congenital absence of uvula be carried out in areas where traditional uvulectomies have been common practice. We believe further research may yield more cases if we consider evolution to play a role.

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