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(CASE REPORT)

Congenital absence of uvula with velopharyngeal insufficiency and its implications

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Abstract

Isolated nonsyndromic cleft palate may be associated with absence of uvula. It is very rare to see the congenital absence of uvula without any other associated congenital anomaly.

Velopharyngeal insufficiency (VPI) is a relatively infrequent disorder in children which usually presents with disordred speech development and recurrent respiratory infection due to improper closure of mouth cavity from the nose during productive speech and swallowing.

Important causes of VPI in children include congenital deformity of soft palate, pharynx and /or uvula.

Other causes like cerebral palsy, neuromuscular dysfunction including congenital myopathy may cause secondary VPI, so-called velopharyngeal 'incompetence'.

Without early diagnosis and timely intervention, the affected children can develop significant speech impairment and social- intellectual suffering.

We describe here a girl child of 8 years without any facial or limb dysmorphism who presented with impaired speech and language development and recurrent upper respiratory infection from VPI resulting from congenital absence of uvula. She eventually developed poor social interaction for which counselling was conducted. She was also treated with speech therapy and referred to pediatric plastic surgeon for a possible uvuloplasty

Keywords: Uvula; Soft palate; Velopharyngeal insufficiency; Speech therapy; Counselling

1. Introduction

VPI is defined as an abnormality in the reflex neuromuscular coordination in palato- pharyngeal synchrony during swallowing and productive speech.

Congenital anomalies of soft palate like cleft palate, bifid or aplasia/ hypoplasia of uvula are not infrequently found in clinical practice. The median raphe is an important part of the uvula which strengthens the attachments of tensor veli palatini and levetor veli palatini during elevation of soft palate while swallowing or phonating.

One or more of these anomalies are liable to produce velopharyngeal insufficiency eventually. Isolated absence of uvula is a relatively rare congenital deformity.

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Left untreated, babies and children with cleft palate may have dental problems, ear infections and hearing problems, feeding difficulties, or unintelligible speech which may affect child's self-esteem and social relationships and many such children find it difficult to attend school, communicate fluently.

Here we describe an 8 years old girl who presented with history of non- development of speech and of recurrent upper respiratory infection who on inspection was found to have complete absence of uvula without any facial or limb dysmorphism. She eventually developed poor social attachments with her peers and relatives and required counselling.

2. Case report

An 8 years old girl child presented with hypernasality and unintelligible speech. She also used to keep her isolated from social gathering and peer- play.

She was the 2nd child born to her non -consanguineous parents. The first child (boy) did not have any illness and was developmentally normal.

Mother of the patient died about 2 years after giving birth to her. Cause of her death was unexplained.

Any history of maternal illness during pregnancy was denied by her father.

Routine medications in antenatal period had been taken and institutional delivery had been conducted at term by Caesaean section. Immediate neonatal period was uneventful. No document was available to know about her oral cavity after birth.

The girl received all routine vaccinations duly. Her father was unaware of pneumonia and influenza vaccines.

During her infancy and childhood she had recurrent cough and cold which usually resolved spontaneously. She had one choking episode while swallowing solid oily food in her school.

Her developmental milestones had been normal and scholastic performance was average, being a student of 3 rd standard.

On examination her hight and weight for age were within normal limit.

No sign of any acute or chronic malnutrition was evident. No apparent facial or limb dysmorphism was present. Inspection of the oral cavity revealed a median cleft of the soft palate and complete absence of uvula (fig.1). The median raphe was also absent.

Her voice was hypophonic with nasal tinge and words were barely audible from a distance of more than 50 cm. Fluency, comprehension and repetition were normal. Her vitals and oxygen saturations were normal.

There was no significant pallor. Cyanosis, jaundice, edema, clubbing were absent.

Her respiratory, gastrointestinal, cardiovascular and neurological examination clinically did not show any abnormality.

Thus a diagnosis of congenital absence of uvula with Velopharyngeal insufficiency was made.

She was referred to plastic surgeons for a possible reconstructive uvuloplasty. Speech therapy was instituted and psychological counselling was conducted for her impaired social interaction.

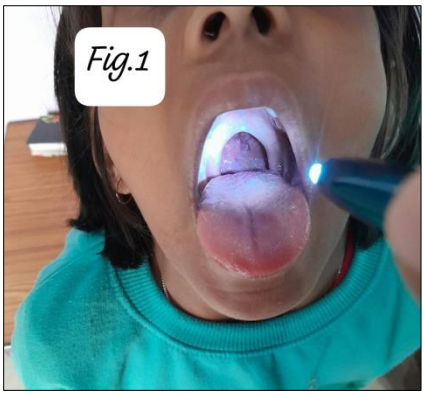

Figure 1 Patient's oral cavity showed absence of uvula. No history of prior palatal surgery

3. Discussion

Isolated nonsyndromic cleft palate may be associated with absence of uvula. It is very rare to see the congenital absence of uvula without any other associated congenital anomaly (1,2).

Velopharyngeal insufficiency (VPI) denotes a disorder of the velopharyngeal coordination which normally seals the nasopharynx from the oropharynx during sucking, swallowing, blowing, vomiting and speech articulation (3).

The etiology of VPI is multifactorial, mostly idiopathic with associated palatal defect , but familial autosomal dominant occurance of isolated cleft palate can occur, usually in midline (4,5).

Visual assessment of velopharyngeal portal is the most effective way of determining portal function during phonation. Direct methods involve viewing the movements of the palate and pharyngeal walls via nasal endoscopy (6).

VPI can occur secondary to structural deficits, neurological disorders, faulty learning, or as a syndromic finding (7).

Our patient attended the OPD with the complaint of low grade fever, cough and dysphonia due to a probable upper respiratory infection. On routine examination of oral cavity there was complete absence of uvula with a soft palate cleft in the midline. The median raphe was also absent.

She and her father were not aware of the defect in her oral cavity as no health care giver earlier saw the defect and communicated to them. Probably her mother also overlooked it. When asked about the mumbling of speech they admitted her inability to produce optimally audible voice from her early childhood. Her father denied of any problem during her breast feeding. But she had 1 episode of choking while eating solid food few months back which was overlooked again.

Her impairment of speech like hypernasality and clagginess of voice were present causing a "whispering" and unintelligible quality.

She eventually developed introvertness for her low volume speech and kept herself isolated from her peers.

Immediate counselling was conducted with her father, speech therapy was instituted and she was referred to pediatric and plastic surgeon for a possible reconstruction of the anomaly. Her poor social interaction was taken care of by psychological counselling and assurance. .

4. Conclusion

Untreated cleft palate may have devastating consequences on a child's self-esteem and social relationships and many such children find it difficult to attend school and communicate freely.

All potential parents should be made aware of and counselled about congenital anomalies in their children, particularly the defects in oral cavity which might have long term implications without timely intervention.

The primary care pediatrician has an essential role in making a timely diagnosis, acute care, anticipatory guidance, and appropriate referral. Speech therapy, if instituted early can significantly compensate for the anatomic defect

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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