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# A SINGLE CASE STUDY ON VELOCARDIAL FACIAL SYNDROME(VCFS)

Clinical Research

**Akshaya S** BASLP,

**P. S. Divya*** Clinical supervisor, MERF-Institute of Speech and hearing (P)Ltd, chennai-28
*Corresponding Author

## ABSTRACT

Velocardiofacial Syndrome(VCFS) is a genetic condition characterized by abnormal pharyngeal arch development. The general Characteristics of VCFS include cleft lip palate (CLP) usually of the soft palate, heart problems, typical face, eye problems, feeding problems associated with nasal regurgitation, palatal differences, middle ear infections, and low-calcium (hypoparathyroidism).However, not all the symptoms may be evident in all patients. An 8 year old male child with presence of hyper nasality in VCFS not associated with CLP. Speech and language profiling was carried out using Teaching and Assessing Language for Kids (TALK),Articulatory skills (Hindi Articulation Test),Nasal emission and Intelligibility using Universal Speech Parameters and VPI screener. This study highlights the importance of subjective measures of nasality in children with velocardiofacial syndrome.

## KEYWORDS

### CASE REPORT

An 8 year old male presented to the department of speech language pathology with the complaint of nasalized speech. Pre-Natally, the child's mother had measles and high fever by 1.5 months of gestation. Peri-Natally, No complications was reported to be present. Post-Natally the child had undergone surgery for Patent Ductus Arteriosus (PDA) by 25 days after birth and has undergone surgery for hernia by 4 years of age. All the articulatory structures except Hard palate (High Arched Palate) was observed to be structurally normal and functionally adequate for speech production.Medical History reveals Nasoendoscopy was done at a private hospital (02/05/15) - Nasopharynx : Grade-1 Adenoids; Normal Soft palate movement; Adequate Closure VP Sphincter. Vision :The child has undergone surgery for cataract at 2 years of age. Audiological findings reveals the child has Moderate loss in the right ear and mild loss in the left ear.Motor development was observed and reported to be delayed. Speech and Language development was reported to be normal but substitution of nasals was predominant. No Significant behavioral history was reported to be present except Bedwetting. Pre-Linguistic Skills such as Eye contact, Sitting tolerance, Mutual eye gaze, Attention and concentration and Working memory were observed to be good.Speech and language profiling was carried out using Teaching and Assessing Language for Kids (TALK)was observed to be Age Appropriate.

### Speech Skills

Speech and language skills was assessed and the profiling is done in **Table.1**

**Resonance** was assessed using Universal speech parameters and the profiling is done in **Table.2**

**Table 1 represents the speech skills wherein articulation, voice, prosody and fluency was noted.**

| | |
|---|---|
| **Articulation**<br>• SODA errors were present<br>• Omission of /s/ [in all positions]<br>• Distortion of /s/ [occasionally]<br>• Nasalization: Present<br>• Weak oral pressure consonants<br>**Consistency of errors :** consistent in syllable, word and sentence level.<br>**Stimulability** : Good | **Voice**<br>• Pitch: Appropriate<br>• Loudness : Approprite<br>• Quality : Normal<br>• **Resonance: Rhinolalia Aperta [Hyper Nasality]**<br>**Hyper nasality score:**<br>Word level: mild(1)<br>Sentence level: moderate(2)<br>**Overall Hypernasality in child's speech:**<br>*Moderate degree(2)* |
| **Prosody**<br>Stress : Appropriate<br>Rhythm : Appropriate<br>Intonation :Appropriate<br>Rate of speech :Normal | **Fluency**<br>No significant complications related to fluency of speechwas observed. |

**Table 2 represents the hypernasality scores using VPI Screener.**

| | | |
|---|---|---|
| **VPI SCREENER** | **HYPERNASALITY**<br>Hypernasality is perceived as pervasive and draws attention to itself and away from the message.There is increased nasality heard on high and low vowels.Most vowels retain their identify.Speech is socially unacceptable. | **SEVERITY**<br>**Hyper nasality score:**<br>Word level: mild(1)<br>Sentence level: moderate(2)<br>**Overall Hypernasality in child's speech:**<br>*Moderate degree(2)* |

### Discussion :

This case study highlights the presence of Moderate level of nasality in the VCFS child with no association with cleft lip and/or palate and from the review of literature there has been documented wherein presence of nasality attributed to the fact that there can be differences in the velopharyngeal closure timings due to defect in speech motor control and reduced thickness of superior pharyngeal constrictor muscle, generalized hypotonic, or motor delays.

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