5-2022

Feeding And Swallowing Disorders In Children With 22q11.2 Deletion Syndrome

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Objective

The aim of the study is to identify the frequency of feeding and swallowing disorders in a large cohort of pediatric patients with 22q11DS. It also identifies how these difficulties progress as patients age.

The goal is to better understand feeding issues in this unique population including symptoms, diagnosis, workup, and treatment strategies.

Introduction

22q11.2 deletion syndrome (previously known as DiGeorge's syndrome or Velocardiofacial Syndrome) is one of the most common chromosomal microdeletion syndromes. The incidence in the literature ranges between 1:1000-1:3000 livebirths. [1,2]

These patients present with a wide array of symptoms including, but not limited to, cardiac anomalies, immunodeficiency, velopharyngeal insufficiency, hypoparathyroidism and developmental delay.

Feeding difficulties are commonly reported in this patient population, however, little is known about the pathogenesis and the incidence amongst patients. While these feeding difficulties may be directly related to the patients' cardiac status or abnormal palates, a great number of patients will develop significant difficulties with feeding as an isolated problem. Children with 22q11.2 deletion syndrome fail to gain weight at the same rate as children without the microdeletion.[3] They commonly suffer from difficulty swallowing and recurrent aspirations which may lead to recurrent nasal sinus infections, respiratory as well as ear infections.[4] The patients will often have recurrent hospitalizations, failure to thrive and might eventually require gastrostomy tube feeding.

The authors of this study aim to describe the incidence of feeding difficulties in a population of 22q11.2 deletion syndrome patients and their progression with time. They also looked at the need for alternative feeding methods and the treatments they received in order to better shed light on this common implication of the syndrome.

Methods

After approval from the institutional review board, a retrospective chart review was performed of 166 patients followed in the 22q11.2 deletion multidisciplinary clinic in a tertiary care pediatric hospital.

The charts were searched for any evidence of feeding difficulties which included recurrent aspiration, failure to thrive and reported dysphagia. For the sake of ease, the term feeding difficulties in our paper refers to any patients who had oral or pharyngeal dysphagia, aspirations, failure to thrive due to feeding intolerance or aversive feeding patterns. The need for alternative feeding methods such as nasogastric tube feeding or gastrostomy tube feeding were noted. Patients who underwent video fluoroscopic swallow studies (VFSS) were identified and the findings recorded. The reports of the VFSS were reviewed rather than the films. Patients who underwent more than one VFSS study had their results followed over time and changes noted. Patients who underwent surgical laryngoscopies as well as injection laryngoplasties were recorded.

The data collected was reported in percentages unless otherwise specified. The information was then used in a descriptive manner to determine the incidence of feeding difficulties and how they were treated in our institution.

Table 1. Patient characteristics.

	Yes (%)	No (%)
Concern for feeding difficulties	39.2	60.8
Gastrostomy tube	23.4	76.6
Laryngomalacia	11.4	88.6
Laryngeal cleft	7.8	92.2
Failure to thrive	4.3	95.7

Results

141 patients had 22q11.2 deletion syndrome
There was a total of 65 VFSS that were reviewed. These belonged to 39 patients who had at least one VFSS performed during the time they were followed in our institution. Thirteen patients had more than one swallow studies that were reviewed separately to follow the pattern of swallowing difficulties they had over time. Of the total VFSS reviewed, 72.3% had oral dysphagia and 78.5% had pharyngeal dysphagia. 56.9% experienced penetrations into the larynx.
Of the 13 with repeat studies and oral dysphagia, 7 patients (53.8%) had no change, 3 patients had improvement in symptoms, whereas 2 patients had worsening of symptoms. Pharyngeal dysphagia seemed to improve in only 1 of the 13 patients.
Penetrations resolved in 5 patients followed over time. The remainder of the patients either had worsening or unchanged penetrations.

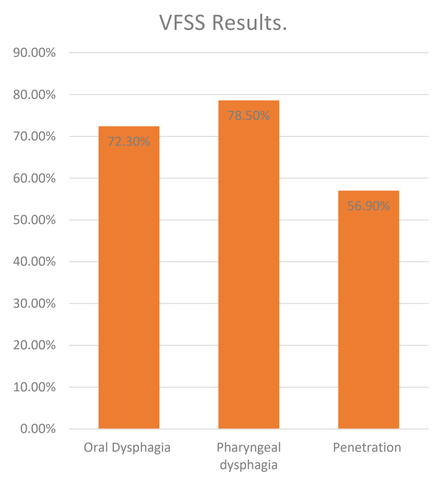

Chart 1. Percentage of patients with different VFSS outcomes.

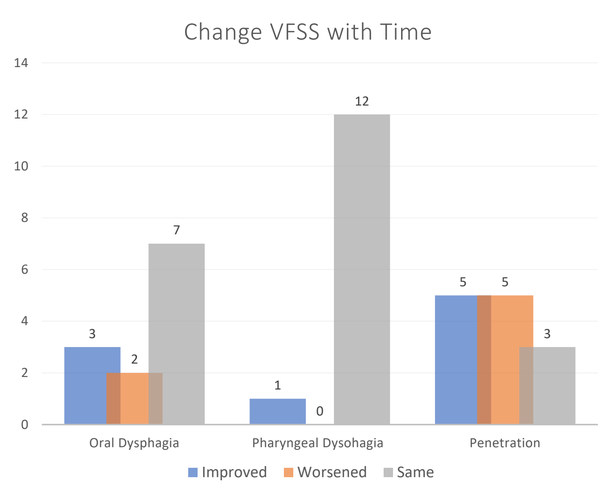

Chart 2. Changes over time in patient with repeat VFSS

Discussion

Several authors have noted that patients with 22q11.2 deletion syndrome suffered from more feeding difficulties than non-syndromic children with similar anomalies and proposed different mechanisms for these differences.

Eischer et al. proposed that the cricopharyngeal muscle prominence and poor muscle coordination resulted in these difficulties. Patients with 22q11.2 deletion syndrome have also been found to have abnormal sucking parameters, abnormal labial seal, and difficulty with bolus control.

Swallowing is a complex mechanism requiring neurologic as well as oro-facial muscular coordination. The development of these oro-facial muscles is dependent on several lower cranial nerves and regulatory factors. The *LgDel* mice hindbrains were studied showing they had a deficiency in these factors and nerve regulators causing difficulty in swallowing in these mice.

Welby et al., using the *LgDel* mouse model, were able to demonstrate that oropharyngeal dysphagia and feeding difficulties persisted as the mice grew, implying that feeding difficulties were not only a childhood complication in this patient population but could be a lifelong concern. This is demonstrated in our patient population as well, as we noted that the majority of children with repeat VFSS did not improve over time.

Conclusions

Although feeding difficulties are common amongst the 22q11.2 deletion syndrome population, they are not well described thus far. In this paper we were able to show the prevalence of feeding disorders amongst our patient population.

Most importantly, we were able to show that feeding difficulties did not improve as patients grew older. This had previously been demonstrated in the *LgDel* mouse model. This is important when counselling families about alternative feeding methods such as nasogastric or gastrostomy tubes and the potential for long term use.

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