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Feeding and Swallowing Disorders in 100 children With 22q11.2 Syndrome

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Research Abstract Title

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IRB Number: STUDY00002026

Describe role of Submitting/Presenting Trainee in this project (limit 150 words):

The submitting author, along with the PI, identified the gap in the literature about the topic at hand. She was involved in the data collection, the analysis of the data as well as the literature review and writing of the abstract.

Background, Objectives/Goal, Methods/Design, Results, Conclusions limited to 500 words

Background:

22q11.2 deletion syndrome (22q11DS) is the most common microdeletion syndrome. The syndrome includes a wide range of variable features including congenital heart disease, palatal abnormalities, immune deficiency and learning difficulties. Otolaryngologic manifestations include hearing loss, characteristic facial features and laryngotracheal abnormalities. A few studies have reported dysphagia as a common feature of this syndrome however little is known about the specific feeding and swallowing difficulties that occur in children with 22q11DS.

Objectives/Goal:

The aim of the study is to identify the frequency of feeding and swallowing disorders in a large cohort of pediatric patients with 22q11DS. The goal is to better understand feeding issues in this unique population including symptoms, diagnosis, workup, and treatment strategies.

Methods/Design:

A retrospective chart review of 100 random patients from our 22q11DS patient repository was performed. Patient information was recorded including demographics, history of feeding difficulties, diagnostic modalities, and treatments. The use of alternative feeding methods such as nasogastric tube (NG) or gastrostomy tube insertion were noted.

Results:

One hundred patients (age 3 months-18 years) with 22q11.2 deletion syndrome were included. 41 patients of the cohort were evaluated by occupational therapy or a feeding therapist for dysphagia. One quarter of the patients (n=25) had feeding difficulties requiring at least one swallow study. Swallow study results showed 76% with oral dysphagia, 72% with pharyngeal dysphagia and 48% with laryngeal penetrations. Eighteen patients required hospitalization for feeding related problems including failure to thrive and aspiration. Twenty-six patients required NG feeding and 25 patients underwent gastrostomy. 28 had an operative airway evaluation performed and ten of these were found to have laryngeal cleft that underwent injection/repair.

Conclusions:

This study suggests that feeding difficulties are common in children with 22q11DS and often these patients require swallow studies and airway evaluations. Need for alternative feeding methods was common. Providers should be aware of this increased risk for feeding and swallowing disorders in children with 22q11DS to ensure timely diagnosis and treatment.