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

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Feeding and Swallowing Disorders in 141 Children with 22q11.2 Deletion Syndrome

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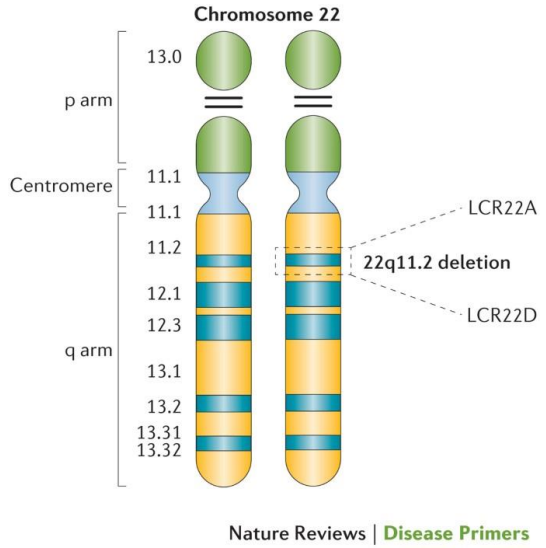
No disclosures

Outline

- Introduction to 22q11.2 deletion syndrome
- Methods
- Results
- Discussion
- Limitations
- Conclusion

Introduction

- 22q11.2 deletion syndrome previously DiGeorge's Syndrome or Velocardiofacial syndrome
- Most common microdeletion syndromes
- Incidence is 1:1000-1:3000 live births
- Wide array of symptoms



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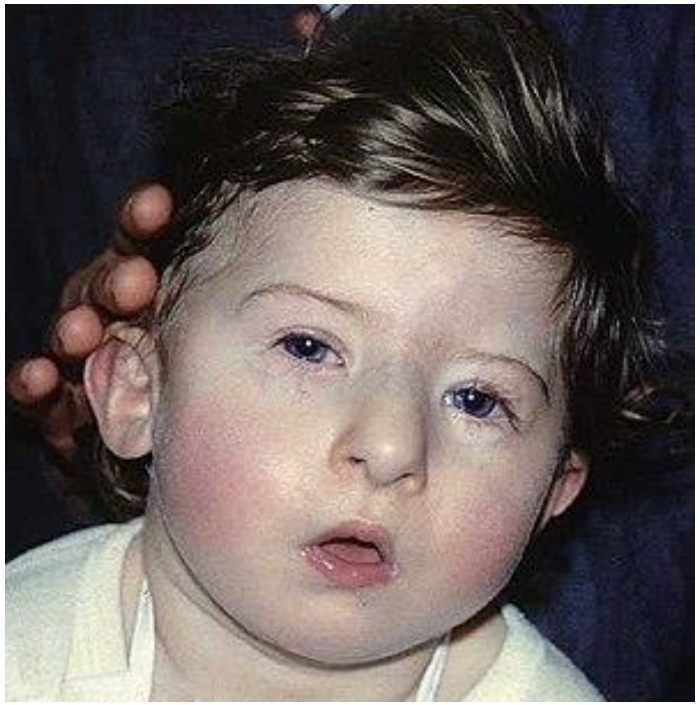
22q11.2 Syndrome

- Cardiac anamolies
- Immunodeficiency
- Hypoparathyroidism/hypocalcemia
- Velopharyngeal insufficiency
- Developmental delays
- Palatal anomalies



22q11.2 and Feeding Difficulty

- Many patients having difficulty with feeding
- Related to cardiac and palatal anomalies
- 22q11.2 patients fail to gain weight at the same rate as patients without the microdeletion
- Difficulty swallowing
- Recurrent aspirations
- Recurrent nasal and ear infections
- Failure to thrive

Goal of Study

- To assess feeding difficulties in our 22q11.2 patient population at CMH
- To assess how these difficulties change with time as patients age
- Need for alternative feeding methods

Methods

- IRB approval
- Retrospective chart review of 166 patients from 22q11.2 multidisciplinary clinic
- Recorded patient with recurrent aspiration, failure to thrive, dysphagia
- Patients with NG and G-tube feeding recorded
- Patients who underwent VFSS had the reports reviewed
- Patients with multiple VFSS followed over time

Results

- 166 charts reviewed- 141 with 22q11.2 deletion syndrome
- 58 (41.1%) were female and 83 (58.9%) were male.

Medical History

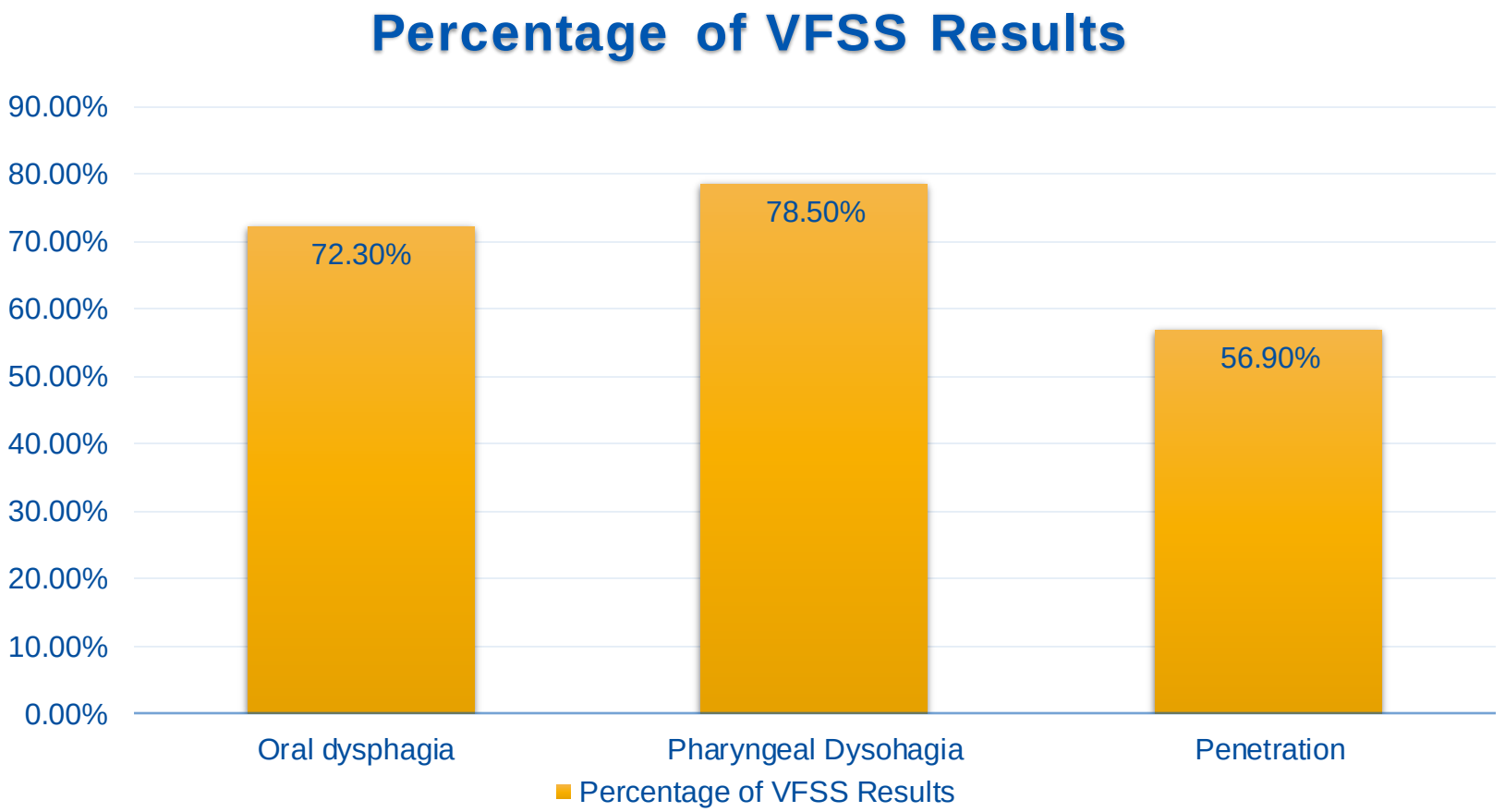
- 85 patients (60.3%) with cardiac history
- 22 patients (15.6%) were known to have overt cleft palates.
- 13 patients (9.2%) had a laryngeal cleft
- 19 patients (13.5%) had laryngomalacia

Speciality seen	Number (%)
OT AND SLP	34 (24.1%)
OT	25 (17.7%)
SLP	6 (4.3%)

Results

- 6 patients (4.3%) had failure to thrive
- 27 patients (19.1%) need admission due to concern for feeding
- **33 patients (23.4%) required gastrostomy tube**

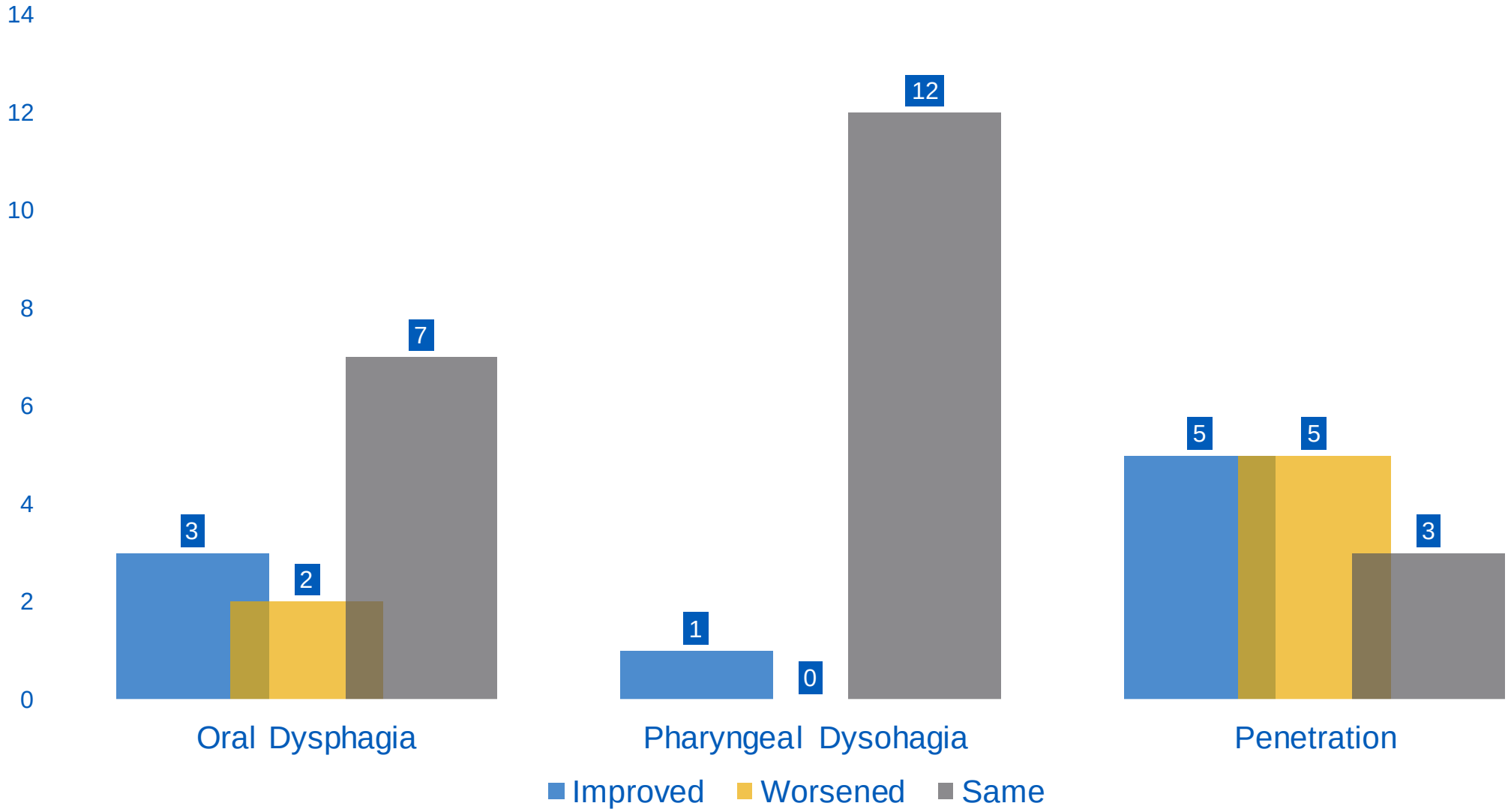
- 65 VFSS reviewed
- Belonged to 39 (27.6%) patients with at least one study



VFSS Outcomes

- 18 patients (27.7%) recommended to have positional changes
- 39 patients (60%) put on thickener
- **18 patients (27.7%) no PO feeding**

Change in VFSS with Time (n=13)



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Discussion

- Different authors have proposed different theories for the difference in feeding patterns of 22q11.2 DS patients

- Eischer et al.
  - 36% of their studied population had difficulty feeding
  - Hyperdynamic movement of posterior pharyngeal wall
  - Cricopharyngeal muscle prominence consistent with incomplete opening and closing of upper esophageal sphincter
  - Poor coordination → trouble feeding

Eicher PS, et al. Dysphagia in children with a 22q11.2 deletion: unusual pattern found on modified barium swallow. *J Pediatr*. Aug 2000

Wong et al

- 41% of their patient population had laryngeal penetration
- Swallowing is a complex mechanism requiring neurologic as well as oro-facial muscular coordination
- Abnormal sucking parameters, abnormal labial seal, difficulty with bolus control and formation as well as pharyngeal nasal reflux

Wong et al., Patterns of Dysphagia and Airway Protection in Infants with 22q11.2-Deletion Syndrome. *Laryngoscope*. 11 2020

- Kapinski et al.,
  - Swallowing requires oro-facial coordination regulated by nerves V, VII, IX, X and XII
  - LgDel mice hindbrains studied after death
  - Mice who had aspirations were found to have deficiency in hindbrain nerve development leading to swallowing difficulties

Kapinski et al., Dysphagia and disrupted cranial nerve development in a mouse model of DiGeorge (22q11) deletion syndrome. *Dis Model Mech*. Feb 2014

Welby et al.,

- LgDel mice feeding disorders did not improve with age similar to our population
- Lifelong concern → need for alternative feeding methods

- Our study → **23.4%** needed G tube

- Campbell et al.→ 16% of patients needed G tube

- Ebert et al.
  - →33% of patients needed G tube
  - →Mean duration 1385 days
  - →Tracheostomy, cardiac surgery, nasopharyngeal reflux, subglottic stenosis, laryngeal web all associated with G tube use
  - →Cleft lip and cleft palate less likely to need G tube

Ebert et al., Percutaneous Enteral Feeding in Patients With 22q11.2 Deletion Syndrome. *Cleft Palate Craniofac J.* 01 2022

Limitations

- Retrospective review
- Small study population with repeat VFSS
- Did not record G-tube use duration

Conclusion

- Feeding difficulties common among this patient population
- These may not resolve with time and might require long term alternative feeding methods
- Repeat VFSS may be needed to better assess patient progression and need for alternative feeding methods
- Information can be useful when counseling patients in 22q11.2 clinic and prior to any intervention
- Need prospective data (possibly multi site)

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