Abnormal Videofluoroscopic Swallow Studies (VFSS) in Infants with Prader-Willi Syndrome Indicate a High Rate of Silent Aspiration

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Background

- Prader-Willi Syndrome (PWS) is a genetic syndrome due to loss of expression from genes within the PWS imprinted region at chromosome 15q11.2-13
- Characteristics include hypotonia and feeding difficulties in infancy with later development of hyperphagia and obesity
- Feeding and swallow dysfunction may lead to the use of a feeding tube such as a nasogastric or gastric tube
- Growth hormone improves tone, body composition, and height and can be started in infancy but there have not been published reports which document improvement in feeding after starting treatment
- Mortality in children with PWS include those due to respiratory illness (61% incidence), choking (5% incidence), and sudden unexplained death (17 % incidence)[1]
- Swallowing abnormalities, or dysphagia, increase the risk of respiratory complications as well as choking
- There are no specific guidelines in evaluation and management of feeding difficulties in infancy
- Despite well-described feeding difficulties in infants with PWS, there are no published reports of formal swallow studies describing swallow physiology and aspiration risk

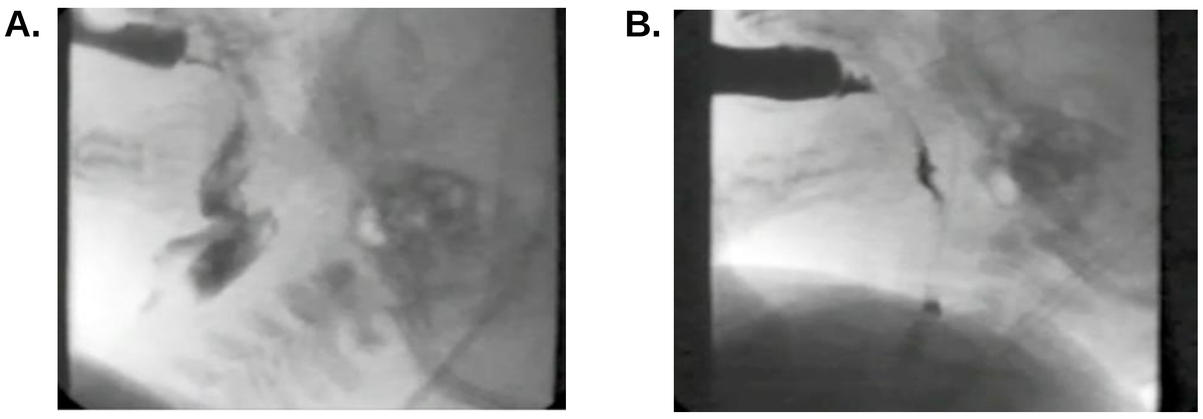

Figure 1. Images from a VFSS (A) Aspiration during swallow, (B) Pharyngeal residue present after swallow.

Objective and Hypothesis

- To evaluate the swallowing function of infants with PWS seen at Seattle Children's Hospital (SCH) with VFSS obtained for clinical indications of poor feeding
- We hypothesize that VFSS will diagnose pathology missed by clinical observation and may help determine feeding safety in PWS infants

Methods

- VFSS results of infants followed in the interdisciplinary Seattle Children's Hospital (SCH) PWS clinic between October 2014 - April 2016 were reviewed
- IRB approval obtained from SCH

Results

- Six infants with genetically confirmed PWS underwent 10 VFSS. Please see characteristics in Table 1.
- 60% of studies were done while the infant was on growth hormone. Average age of growth hormone initiation was 2.5 months.

Acknowledgement

We would like to thank Robin Glass, MSOTR/L, IBCLC for her consultation as well as provision of VFSS images.

Reference:

1. Tauber M, Diene G, Molinas C, Hebert M. 2008. Review of 64 cases of death in children with Prader-Willi syndrome (PWS). American journal of medical genetics Part A 146A(7):881-887.

Results

Table 1. Subject Characterizations at Time of Initial VFSS

	Age	Female/Male	Genetic Subtype	Growth Hormone (age of start)
1	7 mo	Female	UPD	Yes (2 mo)
2	7 mo	Male	Deletion	Yes (3 mo)
3	6 mo	Male	Deletion	Yes (3 mo)
4	3 wk	Female	Imprinting Center	No
5	14 mo	Male	UPD	No
6	6 mo	Male	Deletion	No

- All studies showed pharyngeal dysphagia and silent aspiration (Table 2)
- Most studies (80%) showed abnormal pharyngeal clearance (Table 2)

Table 2. VFSS Results N (%)

Aspiration by Consistency			Silent Aspiration	Pharyngeal Residue
Thin Liquid	Thick Liquid	Puree		
10 (100%)	6 (60%)	2 (20%)	10 (100%)	8 (80%)

- One subject underwent 5 studies over 14 months (see Table 3)
- Studies showed improvement over time but was still abnormal at 15 months old

Table 3. VFSS for Subject 4

Age	Growth Hormone (age of start)	Aspiration by Consistency			Pharyngeal Residue
		Thin Liquid	Thick Liquid	Puree	
3 wk	No	Yes	No	N/A	No
3 mo	Yes (2 mo)	Yes	No	N/A	Yes
7 mo	No	Yes	Yes	Yes	Yes
10 mo	Yes (7 mo)	Yes	Yes	Yes	Yes
15 mo	Yes (7 mo)	Yes	Yes	No	Yes

Conclusions

- VFSS showed pharyngeal phase dysphagia with silent aspiration in all infants, which may have been unconfirmed with only clinical observation
- Abnormalities were present despite early initiation of growth hormone
- Unrecognized swallow dysfunction may be a contributor to morbidity in PWS
- Further longitudinal studies are needed to characterize swallowing function in PWS over time
- Comprehensive evaluation should occur before starting oral feeds in infants with PWS. VFSS can be a useful clinical tool in this decision.

Disclosure:

PS is involved in the multi-center study "Randomized, Double-Blind, Placebo Controlled, Phase 3 Trial of ZGN-440 (Subcutaneous Beloranib in Suspension) in Obese Subjects with Prader-Willi Syndrome to Evaluate Total Body Fat Mass, Food-related Behavior, and Safety Over 6 Months)" sponsored by Zafgen, inc.

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