Prader-Willi Syndrome Choking Prevention and Education: A Practice Change
Amy Moffett, MSN, RN, CPNP and Kathryn Anglin MSN, RN

No Conflict of Interest

Objectives
- Understand pathophysiology behind the increased choking risk for persons with PWS
- Discuss how a quality improvement program can lead to practice change, thus improving patient outcomes
- Engage participants in a simulated training session mimicking the education that has been provided to patients and families in our clinic
Introduction

- PWS is a complex genetic disorder caused by the absence of the normally active paternally expressed gene on chromosome 15q11q13
- First identified in 1956
- Estimated prevalence: 1/10,000 to 1/30,000 births

Genetics of PWS

- Paternal Deletion - 70%
- 25% (UPD)
- Imprinting Defect - 5%
- Defect - 5%
- 25% (UPD)
- Imprinting Defect - 5%

Clinical Features of PWS

- Hypotonia
- Feeding difficulties
  - Failure to thrive early on then hyperphagia
- Developmental delay
- Short stature
- Hypogonadism
Clinical Features of PWS

- Compulsive-like behaviors
- Low IQ
- Behavior issues (tantrums, stubbornness, compulsive)
- Skin and rectal picking

Clinical Features of PWS

- Paternal Deletion - 70%
- 25% (UPD)
- Imprinting Defect - 5%
- Defect - 5%
- 25% (UPD)
- Imprinting Defect - 5%
Clinical Features of PWS

- Paternal Deletion - 70%
- Imprinting Defect - 5%
- Other - 25% (UPD)

Choking Risk in General Population

- Choking is an important public health problem for children
- Choking is the leading cause of morbidity and mortality among children, especially ages 3 years and younger
- Food, coins, balloons, and toys are the primary causes

Prevention of Choking Among Children Committee on Injury, Violence, and Poison Prevention

Choking Risk in General Population

- Choking prevention standards on foods and toys are addressed in the United States (US) under:
  - Child Safety Protection Act
  - The Consumer Product Safety Improvement Act
  - Regulation of the US Product Safety Commission (CPSC)
- Pediatricians, dentists, and other health care providers should provide choking prevention counseling as anticipatory guidance

Choking Risk in General Population

Choking on Food

- Choking on food causes the death of ~1 child every 5 days in the US
- Hotdogs accounted for 17% of food-related choking among children younger than 10 years old
- Latex balloons accounted for 29% of deaths from 1972-1992

Choking Risk in General Population

Choking on Food

- Food poses an important and under-addressed problem for US children
- ~66-77 children younger than 10 years of age die from choking on food each year
- >10,000 ED visits annually can be attributed to choking on food among children 14 years and younger
- Children <4 years old and children with chewing/swallowing disorders are at greater risk of food-related choking
Choking Risk in PWS

Deaths Due to Choking in Prader-Willi Syndrome

American Journal of Medical Genetics (2006)

- Brief survey 1999 made available by the Prader-Willi Association (USA) bereavement program
- Demographic information available on 178 deceased PWS individuals. Cause of death available on 152 individuals
- 54 families completed questionnaires later in 2004


Choking Risk in PWS

From the Family Questionnaires

- 34% of PWS subjects had a history of choking
- Choking was listed by families as the cause of death in 12 subjects (7.9%). Average age at death was 24 years
- Only 2 individuals were less than 8 years of age*
- Higher incidence in males (11 of 12 subjects)


Choking Risk in PWS

Gastrointestinal History

- Choking incident 34%
- Required Heimlich maneuver 6%
- Decreased vomiting during viral illness 79%

Additional Information

- Only 3 persons reported receiving GH at the time and 10 persons took GH at some time in the past

Choking Risk in PWS

Data suggests that the risks associated with choking are different in the PWS population compared with others.

• Poor oral/motor coordination
• Generalized hypotonia of the pharyngeal musculature may also lead to swallowing difficulties
• Poor gag reflex
• Hypotonia
• Hyperphagia

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Choking Risk in PWS

Poor oral/motor coordination
Generalized hypotonia of the pharyngeal musculature may also lead to swallowing difficulties
Poor gag reflex
Hypotonia
Hyperphagia
Choking Risk in PWS

- Viscous saliva
- Decreased volume of saliva
- Decreased mastication
- Consuming large quantities of food
- Eating habits: foraging, stealing food, and the consumption of inedible foods / non food items

Choking Risk in PWS

- Increased abdominal girths in individuals with PWS may predispose to increased intragastric pressure and gastroesophageal reflux
- The presence of gastroesophageal reflux and esophagitis are predisposing factors to the development of esophageal food impactions in the general population

Choking Risk in PWS

Survival Trends from the Prader-Willi Syndrome Association (USA) 40-Year Mortality Survey

- The largest study to date regarding the causes of death in PWS utilizing the PWSA (USA) syndrome-specific mortality survey database spanning the past 40 years
- Analyses and interpretation of the data limited by the reliability of death reporting based upon the availability and knowledge of family members / access to confirmed genetic status / autopsy reports

Choking Risk in PWS

- Survival estimates for PWS have significantly increased (particularly for cardiac deaths among females, and thrombotic causes and GI-related mortality)
- Choking is more common in males due to rapid eating when sneaking food, but also due to lack of saliva and ineffective swallowing

Choking Risks in PWS

- The results of the 40-Year Mortality Survey provide useful insight into risk factors, mortality trajectory over time and areas of need
- Family members, care providers and health care professionals should be made aware of these risk factors and causation of death to improve the longevity and quality of life of persons with PWS

Choking Risk in PWS

Results:

- Need for increased awareness of the risks in PWS mortality
- Close monitoring and supervision during meals, outings and near roadways to reduce the risk of traffic collision from food seeking (e.g., darting into the road to reach a restaurant)
- Strict oversight of food access and quantity are critical to prevent excessive unsupervised food consumption, choking and gastric rupture which can occur from eating quickly
Choking Risk in PWS

**Results:**

- Chewing and swallowing may be compromised in PWS by decreased salivary secretions, dental caries, poor muscle tone and lack of vomiting
- Lifelong weight control /prevention of life-threatening obesity
- Implementation of preventive measures, specialized training and better awareness by families and group home care providers including: training for use of the Heimlich maneuver, supervised meals, food security along with food preparation and diet modification

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**Quality Improvement**

- A quality improvement (QI) program involves systematic activities that are organized and implemented by an organization to monitor, assess, and improve its quality of health care
- QI programs are essential to organizations:
  - Improved patient health (clinical) outcomes that involve both process outcomes (e.g. provide recommended screenings) and health outcomes (e.g. decreased morbidity and mortality)
PWS Choking Prevention and Education

Resources Given to Families

PWS Registry Data

Hidden Gut Issues
Resources Given to Families

- We explain that we are teaching them choking prevention and choking first aid, **but not certifying them**
- We give them a brochure on local class offerings if they would like to sign up for a full CPR class
- We also give them a reference sheet on CPR for their child’s age group, as we are mainly concentrating on choking first aid during our teaching sessions

Simulated Training Session

Outcomes
Outcomes

Prader-Willi Syndrome Education
Average Score Per Question (n=28)

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<tr>
<td>Informed</td>
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</tr>
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<td>Comfortable</td>
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</table>

Of the families who have received education, on the pre-test question regarding “Awareness of increased risk of choking in the PWS population”, 4 families indicated that they were not at all aware and 5 families indicated that they were only somewhat aware, prior to teaching/training.

Outcomes

Percent of Prader-Willi Syndrome Patients Who Have Experienced a Choking Episode
Oct 2018-Mar 2019 (n=28)

- Yes: 33%
- No: 67%

Percent of Prader-Willi Syndrome Patients Who Required First-Aid for a Choking Episode
Oct 2018-Mar 2019 (n=28)

- Yes: 11%
- No: 89%

Practice Change

- PWS individuals are at higher risk of choking. Therefore, it is imperative that caregivers are educated on choking prevention techniques and choking first aid.
- Establishing a choking education program is cost effective and does not pose a significant burden/workload to the organization.
- NCH Section of Endocrinology elected this QI project as the QI metric for 2019.
Family Feedback

• One mother who was already certified in CPR said that it was very helpful to specifically concentrate on choking prevention and choking first aid, since this is a smaller segment of the entire CPR certification course

• A father who is a physician still found the resources and training helpful as it was specifically tailored towards children with PWS

• Families appreciated their older children knowing how to make the universal choking sign

Future Implications

• Implementing choking questionnaire into the electronic medical record as a screening question at intake

• Swallow studies for high risk patients

• Ongoing education

• Impact of growth hormone therapy on choking risk and episodes

Acknowledgements

• We are forever grateful for our PWS clinic patients and their families. It is an honor to travel this journey with each and everyone of them

• We would like to express special thanks to Dr Kathryn Obrynha, and our QI guru Don Buckingham who gave us this golden opportunity to do this wonderful project on choking in the PWS population
Questions

References

• American Heart Association. (2016). Family & Friends CPR [DVD video]