

**Prader-Willi Syndrome:**  
A Comprehensive Training

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# Background/ Experience

- Education- Elementary and Special Education.
- Dissertation and research has focused on PWS
- Completed hundreds of trainings and consultations with schools, non-PWS specific group homes and community agencies.
- Have personally worked with over **500** people with PWS.
- Have presented about PWS at the state, national, and international level.
- Written various articles for PWSA-USA and research journals about PWS
- Author of a children's book- Penny A Tale of Lost Currency

# Agenda

- What is Prader-Willi Syndrome? (PWS)
- History and Phases of PWS
- Diagnosis and Testing for PWS
- PWS and the Hypothalamus
- PWS Characteristics
- Nutrition and PWS
- Behavior and PWS
- Summary of Managing PWS
- PWS Resources
- Questions/ Comments/ Discussion

# What is Prader-Willi Syndrome?



# What is Prader-Willi Syndrome?

- Prader-Willi Syndrome is a genetic condition.
- Prevalence- 1:12,000 to 1:17,000
- PWS occurs:
  - - equally in males and females
  - - affects all races
  - - the most common genetic cause of obesity
- Failure to receive active genes- **father's chromosome 15**  
**(15q11-q13)**

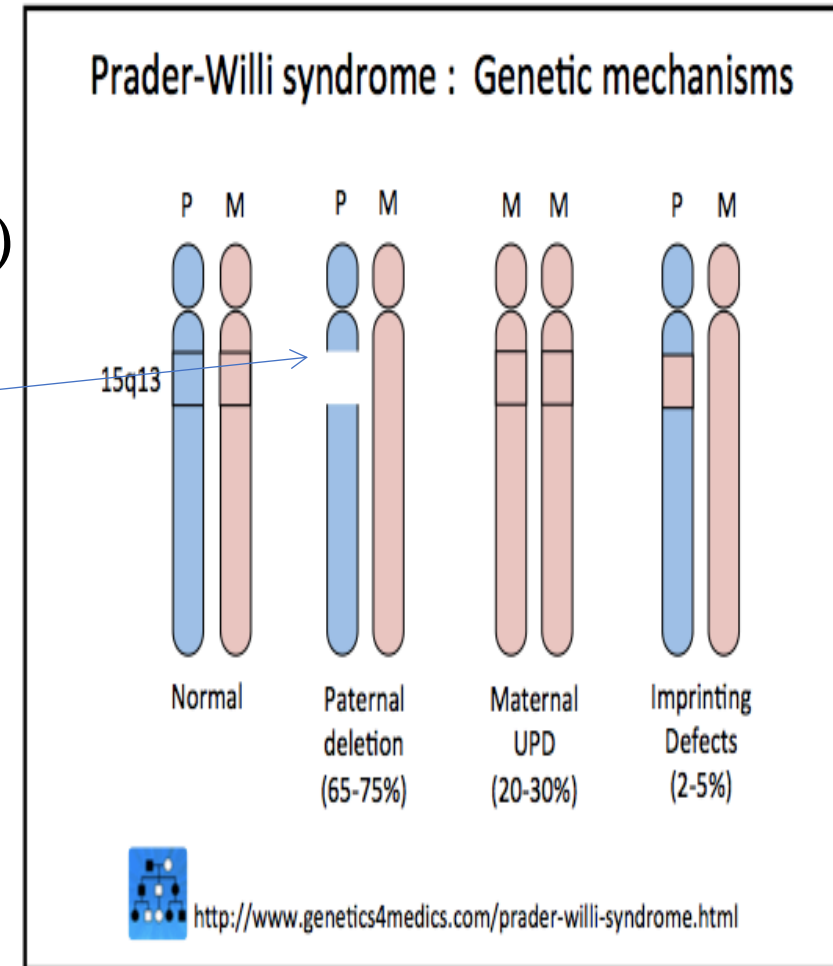
# What is Prader-Willi Syndrome?

- PWS is rarely hereditary.
- PWS rarely occurs more than once in a family.
- Previously, life expectancy for a person with PWS was short.
- However, with better management, individuals with PWS are living into their 70's.
- There are individuals that die at a young age due to mismanagement of the syndrome.

# What is Prader-Willi Syndrome?

- **Paternal deletion**

- Loss of a small piece of chromosome 15 (father)
- Genetic information missing
- Nothing the father did or did not do
- Can't be prevented
- About 70% of the PWS population



# What is Prader-Willi Syndrome?

- **UPD**

- No deletion on Chr. 15

- *Uni-parental*- one parent

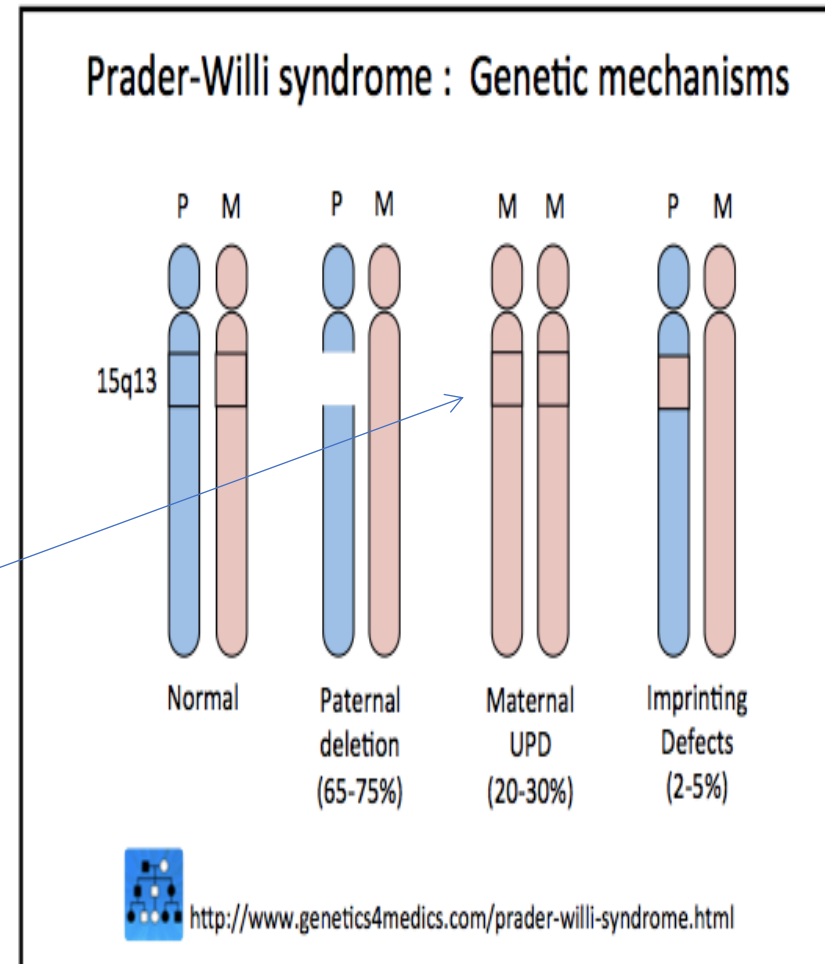
*Disomy*- two chromosome bodies

- Both Chr. 15's- mother  
“uni-parental disomy”

- Structure is normal

- Wrongly distributed

- About 25% of the PWS population

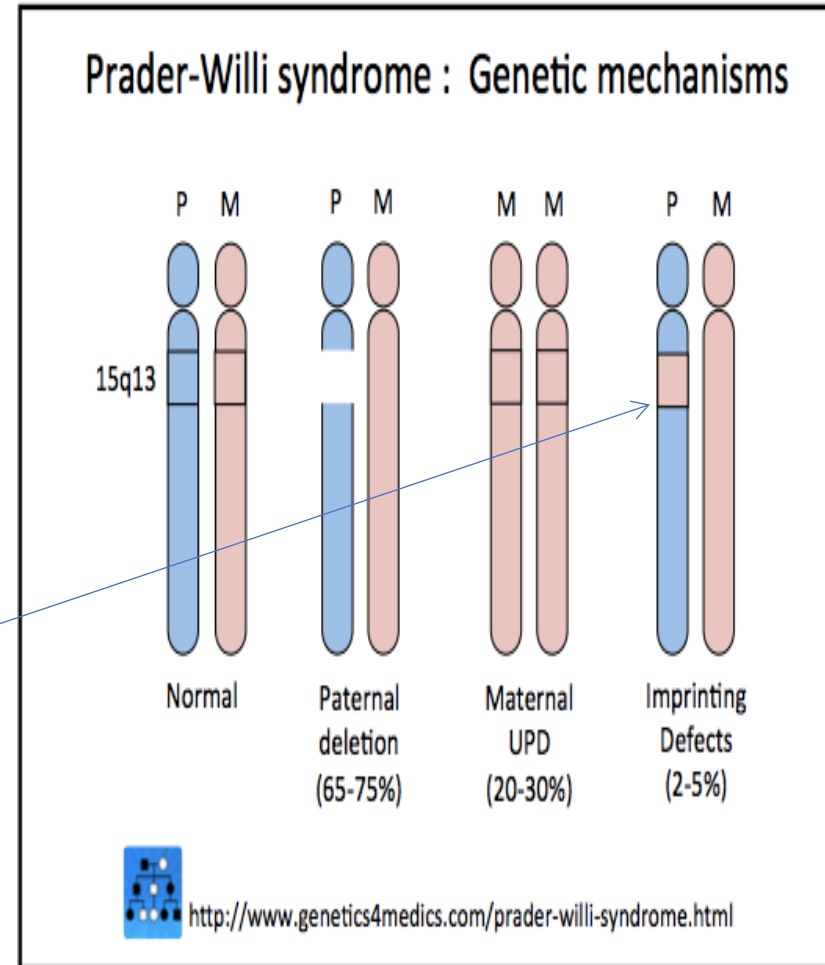




# What is Prader-Willi Syndrome?

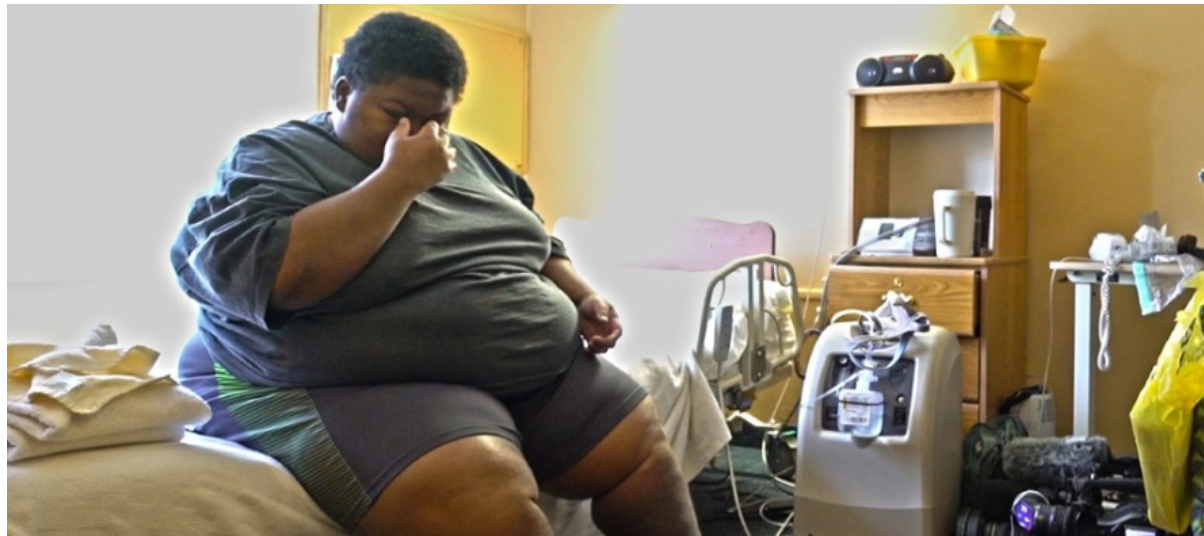
- **Imprinting Defect**

- Small percentage of cases causes the paternal chromosome 15 genetic material (although present) to be inactive.



# The History of PWS

- PWS was first described by Swiss doctors (Andrea Prader, Alexis Labhart, and Heinrich Willi (1956))
- Clinical characteristics= 9 children they examined
- Historically- 2 stages of PWS
- Currently- multi-phase syndrome with sub-phases



# PWS- Phase 0

## PWS- Prior to birth

- Occurs in utero
- Characteristics are:
  - decreased fetal movements
  - decreased birth weight
  - decreased length



# PWS- Phase 1

- **PWS- PHASE 1- (0-2 years 8 months old)**

Infant is hypotonic and not obese

- **Sub-phase 1a**

poor appetite, feeding difficulties, failure to thrive

- **Sub-phase 1b**

growing steadily on the normal curve; improved appetite



# PWS- Phase 2

## Sub-phase 2a

(2 years 8 months 4 ½ years old)

Weight increases above normal

No significant caloric intake

No increased appetite

No increased interest in food



## Sub-phase 2b

(4 ½ years old-8 years old)

Abnormal increased

not insatiable appetite

Overweight- obese if diet

not regulated



# PWS- Phase 3

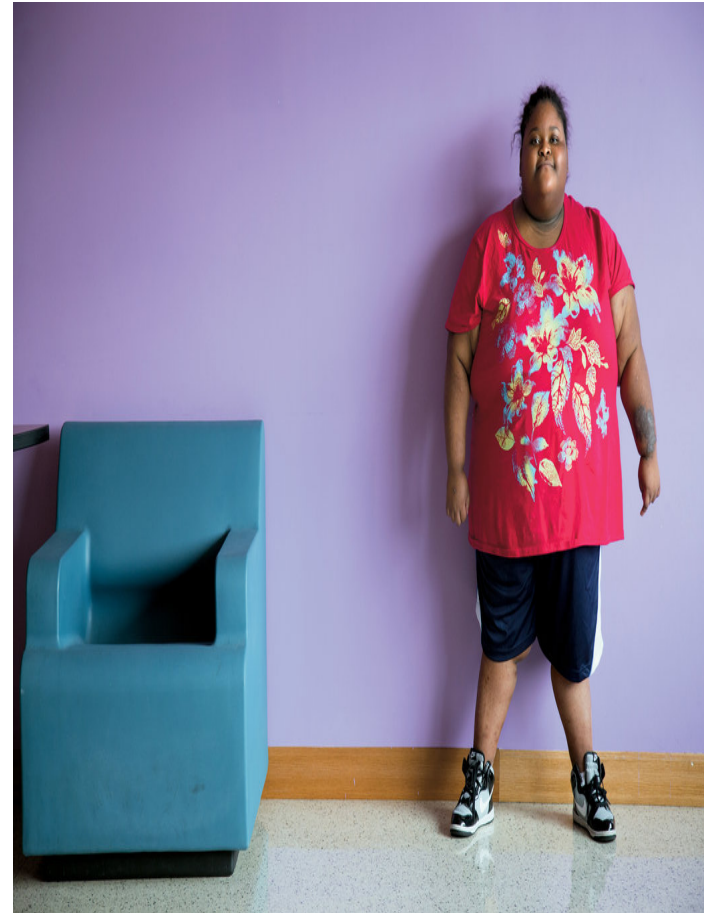
- **8 years into adulthood**
- Hyperphagia- excessive eating and weight gain
- Insatiable appetite
- Intense or relentless food-seeking
- Classic phase of PWS
- It may appear as early as 3 years of age or as late as 15 years of age.





# PWS- Phase 4

- Occurs later into adulthood
- Individual may no longer have an insatiable appetite and can feel full
- Significant improvement in appetite and weight control  
Only occurs in the minority of persons with PWS
- **Most stay in Phase 3**



# Diagnosis and Testing for PW

## **DNA methylation analysis** - >99% accuracy

(Also detects Angelman Syndrome- same region/ chromosome 15)  
*Prader-Willi/ Angelman Methylation Panel*  
*(Can detect all genetic subtypes)*

## **FISH test (Fluorescence in situ hybridization)**

Confirms a deletion if present- does not reveal the size of the deletion  
Does not detect UPD or an Imprinting deletion

## **Chromosome Microarray**

Detects UPD 15- 70% accuracy  
Does not detect the Imprinting defect 15/ PWS

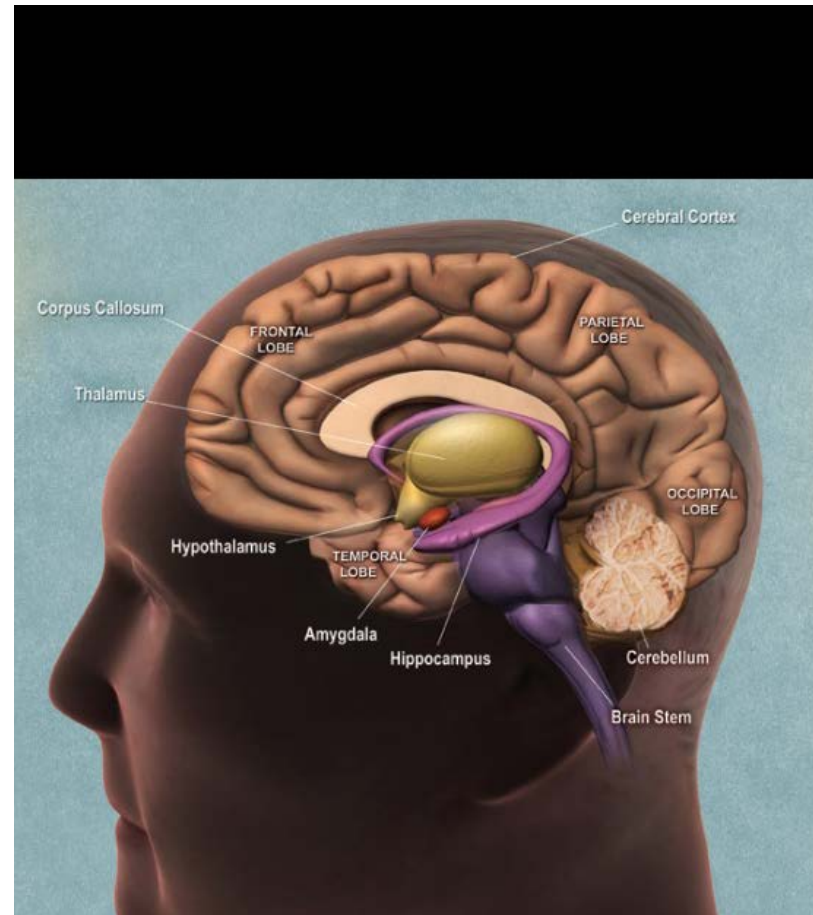
## **UPD and Imprinting Defect Testing**

Specialized DNA test  
Requires blood from the patient and parents for accurate interpretation



# PWS and the Hypothalamus

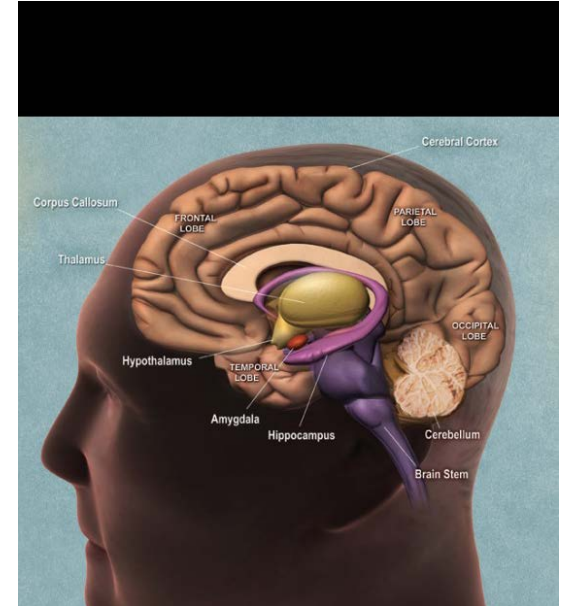
Dysfunction of various hypothalamic systems may be the basis of a number of symptoms in Prader-Willi syndrome.



# PWS and the Hypothalamus

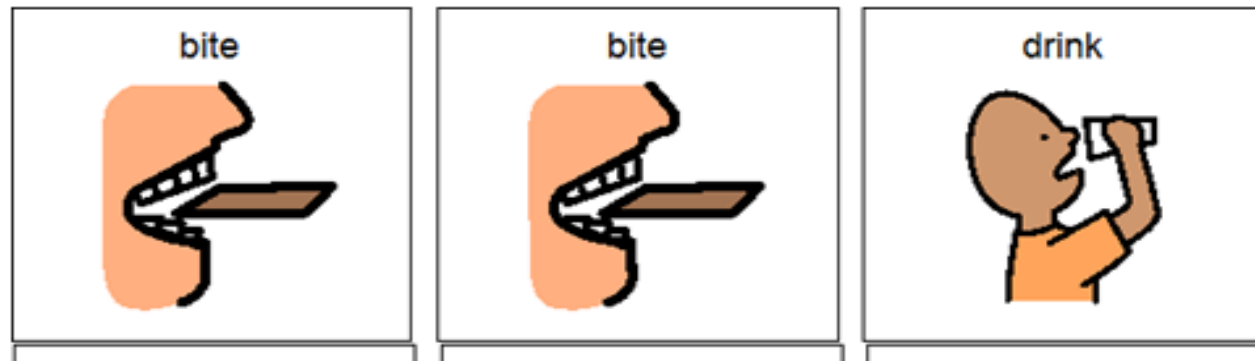
Hormones interact with the pituitary gland

- Physical and emotional stress (stress hormone)
- Thyroid stimulating hormone
- Reproductive hormones
- Behaviors and emotions
- Regulates water levels- signals kidneys to absorb water
- Body temperature
- Circadian rhythms
- Satiety and hunger
- Growth hormone



# Additional Characteristics

- Shorter stature- if not treated with growth hormone
- Small hands and feet- if not treated with growth hormone
- Poor swallowing- PACE and CHASE strategy
- Irregular body temperature- sickness/ infections
- High pain threshold or abnormal response to pain.



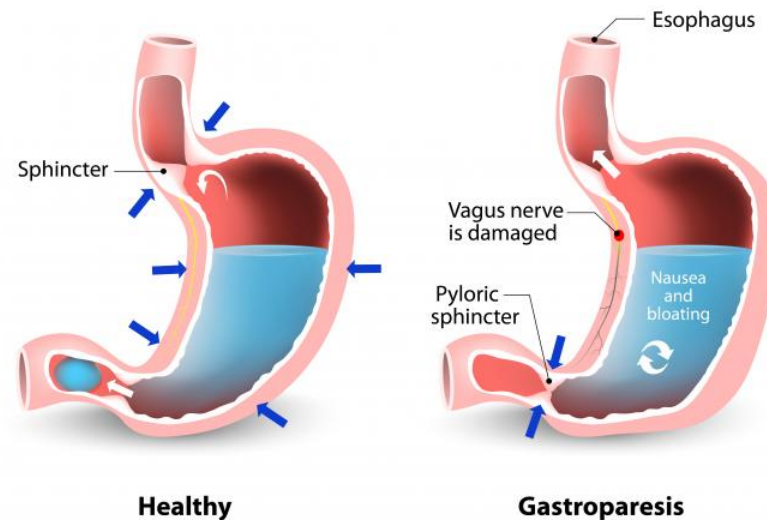
# Additional Characteristics

- Easy bruising
- Curvature of the spine- scoliosis
- Skin picking, cellulitis, rectal picking
- Lymphedema- Swelling in an arm or leg caused by a lymphatic system blockage
- Behavior issues
- Bowels empty slowly- constipation and GI problems
- Immature sexual development and delayed puberty
- Diabetes (associated with significant obesity/ Type II)

# GI Issues and PWS

- **Gastroparesis**- a condition where the movement and digestion of food in the stomach is slowed or stopped.
- A **low calorie diet** should continue but **fiber should be reduced** when slow gastric emptying is diagnosed or suspected.

## GASTROPARESIS



# Additional Characteristics

## **PWS CAN BE LIFE THREATENING!**

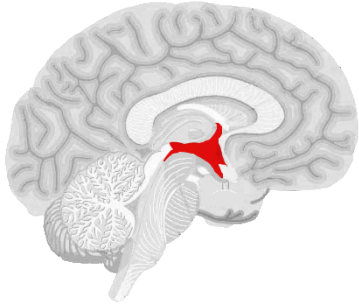
- No vomiting reflex- mostly likely due to low muscle tone and hypothalamic dysfunction
- Seek medical treatment immediately if vomiting
- Death can result for a variety of reasons:
  - 1- respiratory failure
  - 2- choking episodes
  - 3- stomach necrosis or rupture



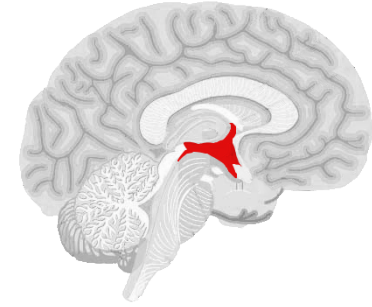
# NUTRITION and PWS







# NUTRITION and PWS



- Persons with PWS display abnormal development of the brain connections that normally trigger satiation, telling the body it has gotten enough to eat.
- They can **NOT** learn to control the urge to eat
- There is no appetite suppressant that will help
- No level of disgust or distaste- will hinder food consumption if presented with the opportunity



# NUTRITION...WEIGHT...PWS

- Decreased REE- Woman and Men with PWS need lower calories  
*(600-1000 calories per day without gaining weight)*
- Persons with PWS = low muscle tone
- Burn 1/2 or 1/3 of the calories than typically developing people
- Increased fat mass compared to weight and body mass index (BMI)  
Ex: 290 lbs      4 ft.= 88.5 BMI



# Cognitive Ability and PWS



# Cognitive Ability and PWS

- Mild to moderate intellectual disability
- 40% mild ID and 20% moderate ID
- IQ estimates fall between 50 and 85/ Mean IQ of 60.
- Some individuals with PWS have borderline ID.
- Intellectual disability= IQ score 70 or below, deficits in adaptive functioning and on-set before 18 years old

# Cognitive Ability and PWS

Higher cognitive level  $\neq$  control over food

Higher cognitive level  $\neq$  control over negative behaviors



# Behavior and PWS



# Behavior and PWS

- PWS has temperamental and behavioral characteristics
- These characteristics are so commonly present that they become part of the PWS behavioral phenotype
- Behavioral phenotype = PWS personality
- Psychiatric and behavioral symptoms can be organized across five domains

# Behavior and PWS

- Food related behaviors
- Cognitive rigidity
- Oppositional/ defiance
- Anxiety/ insecurity
- Skin picking





# Causes of Behaviors in PWS

- unplanned food exposure
- thinking about food
- an upcoming social event
- change in schedule
- a change in the expected meal
- a social situation or interaction
- purposeful meltdowns with someone who they know will *“give in”*





# Domain #1- Food Related Behaviors

- overeating typical food, eating frozen food, food sneaking/ foraging, manipulating to get food, shoplifting or stealing food, breaking locks, verbal/ physical aggression to obtain food, food thinking





# What is **FOOD SECURITY**?



- Food security = managing food related behaviors
- Food security = a measure of the availability of food and the individual's ability to access these foods in socially acceptable ways
- Food security is not just about keeping food out of reach
- It is also about keeping food out of sight and mind.



# What is **FOOD SECURITY**?



<b>No Doubt</b>	<b>No Hope</b>	<b>No Disappointment</b>
<p>Predictable routine Food is scheduled Person with PWS can relax and think less about food Menu predictability reduces anxiety</p> <p>*Dr. Forster and Dr. Gourash- Pittsburgh Partnership</p>	<p>No unplanned extras outside of the schedule meal times No opportunities to gain access to food Chances to obtain food cause stress</p> <p>*Dr. Forster and Dr. Gourash- Pittsburgh Partnership</p>	<p>Expectations are made that will be reliably carried out No other expectations have been raised so there is no disappointment when there are not realized or fulfilled</p> <p>*Dr. Forster and Dr. Gourash- Pittsburgh Partnership</p>



# What is **FOOD SECURITY**?



- Lock all areas that contain food
- Locking reassures the person with PWS that they can't obtain the food- it is not a punishment
- Locking sets the environment up for success
- Provide an alternative setting for scheduled mealtime



# What is **FOOD SECURITY**?



- Provide a structured predictable routine for meals (menu is posted, a written schedule shows when the next meal will occur, the same alternative is given if menu is disrupted)
- Adult supervision should be provided during mealtimes
- Food should be secured during transit in the community.



# What is **FOOD SECURITY**?



- Plan for food exposure that is not typical- field trips, special events, holiday parties, birthdays
- Food should not be used as a reward or during instruction
- Person with PWS should not be “punished” for obtaining unlocked food
- Food security creates improved physical/mental health



# What is **FOOD SECURITY**?



- Not observing “food related behaviors”?
- These behaviors don’t always happen immediately!
- Behaviors could be related to “food thinking”
- Poor expressive language skills = negative behaviors

## **Example:**

12-year-old student with PWS eating in the cafeteria

Afternoon behaviors in the classroom- physical and verbal aggression  
removed him from the cafeteria = behaviors decreased



# What is **FOOD SECURITY**?



- What if food IS accessed in the environment?
- Small portions of food = do not take food!



Large portions of food = take food and be prepared to deal with behaviors that go along with taking food





# Domains #2-4

## Cognitive Rigidity, Oppositional Defiance and Anxiety/ Insecurity

- Could look like worry
- Constant need for reassurance
- Questions about schedules/ expectations
- Some physical agitation-rocking and fidgeting
- Can lead to tearfulness
- Uncooperative behaviors
- Aggressive behaviors
- Shutting down- person not responsive/ not able to be redirected



# Domains #2-4

## Cognitive Rigidity, Oppositional Defiance and Anxiety/ Insecurity

1. Visuals
2. Calendars
3. Social Stories
4. 3 card system
5. Re-direction
6. Prompting to complete a highly preferred activity
7. Positive behavior support

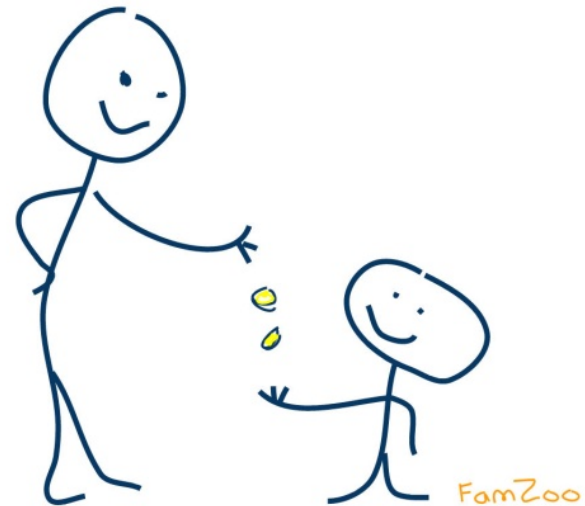


# Positive Behavior Support

- Create a program that includes highly preferred incentives and rewards

**Example:** Tommy engages in physical aggression. When he has “safe hands” during 30 minute increments, he gets a sticker/ check mark, etc. When he gets 4 stickers before 12:00 noon, he gets 15 minutes of computer time.

- *Computer time is the motivator*
- *No punishment- he just doesn't EARN reward*
- *The PWS brain is not wired for consequences*



## Daily Schedule/Record of Accomplishments

Name John Bloom Date 5/17/2019 Staff: Day: Jane Evening Patrick

2 Target Behavior Goals: #1 Safe hands #2 Nice words

Goals:

8:00-10:00 X= 15

11:00-1:00 X= 15

## SCHEDULES/ VISUALS/ REWARD SYSTEMS

<u>TIME</u>	<u>ACTIVITY</u>	<u>Attend</u>	<u>Participate</u>	<u>Cooperate</u>	<u>Behavior #1</u>	<u>Behavior #2</u>
8:00-8:30	BREAKFAST	X	X		X	X
8:30-9:00	Morning announcements	X	X	X	X	
9:00-9:30	Physical Therapy	X	X	X	X	X
9:30-10:00	Physical Therapy	X	X	X	X	X
10:00-10:30	Reward- Computer Time					
10:30-11:00	Occupational Therapy					
11:00-11:30	Occupational Therapy					
11:30-12:00	Free Time					
12:00-12:30	LUNCH					
12:30-1:00	LUNCH					
1:00-1:30	Reward- Computer					

# Domain #5- Skin Picking

- Provide an alternative activity to replace picking
- Offer rewards for behavior you want to see- no picking
- Don't punish for picking
- Don't talk about picking
- Redirection
- Medication may be beneficial in severe cases



# Supporting PWS Behaviors

- **It is often impossible to stop a meltdown!**- Remain calm, think before you speak/act, speak in a calm voice. Avoid yelling! Try not to show fear, anger, or frustration.
- **Keep yourself out of harms way-** move dangerous objects away  
Seek assistance from others if needed
- **Give time and space to calm-** retreat to a designated area until calm
- **If yelling is happening, refuse to speak.**  
Praise when the calm voice is used/redirect.



# Supporting PWS Behaviors

- **Move past the episode!**- Do not punish or debrief the episode later in the day- “What could you have done better when you were mad earlier today?”
- **Avoid reigniting the anger or frustration!**- do not discuss it the following day
- **Never give in to unreasonable demands**
- **Let the meltdown run its course!**- When the person is calm, redirect and move on.



People with PWS are.....

**HARD WIRED**

*PWS Behaviors decrease but they aren't eliminated!*

*Stable behaviors do not equal cured behaviors!*

*Supports can't be removed!*



# Summary of Behavior- PWS

Behavior management with PWS is based on:

- total food security/ limited food exposure
- positive behavior support and verbal praise
- reward systems
- environmental accommodations and modifications
- preparation
- predictability
- schedules
- visuals
- student specific safety plans
- thinking outside of the box about behavior and ways you manage behaviors
- determine if food security or total food security is needed

# PWS Resources

- Prader-Willi Syndrome Association- USA- School Section

<http://www.pwsausa.org>

<https://www.pwsausa.org/medical-issues-a-z/>

<http://www.pwsausa.org/shop/>

- International Prader-Willi Syndrome Organization

<https://www.ipwso.org/for-teachers-cpit>

- Foundation for Prader-Willi Research

<https://www.fpwr.org/>

# Contact Information

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# Questions

