



# PWS Training for Professional Providers

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## *Genetic and Medical Overview*



## Objectives

**What is Prader-Willi syndrome**

**Genetics of PWS**

**Medical Alerts**

**Q&A**



## Overview of PWS and Genetics

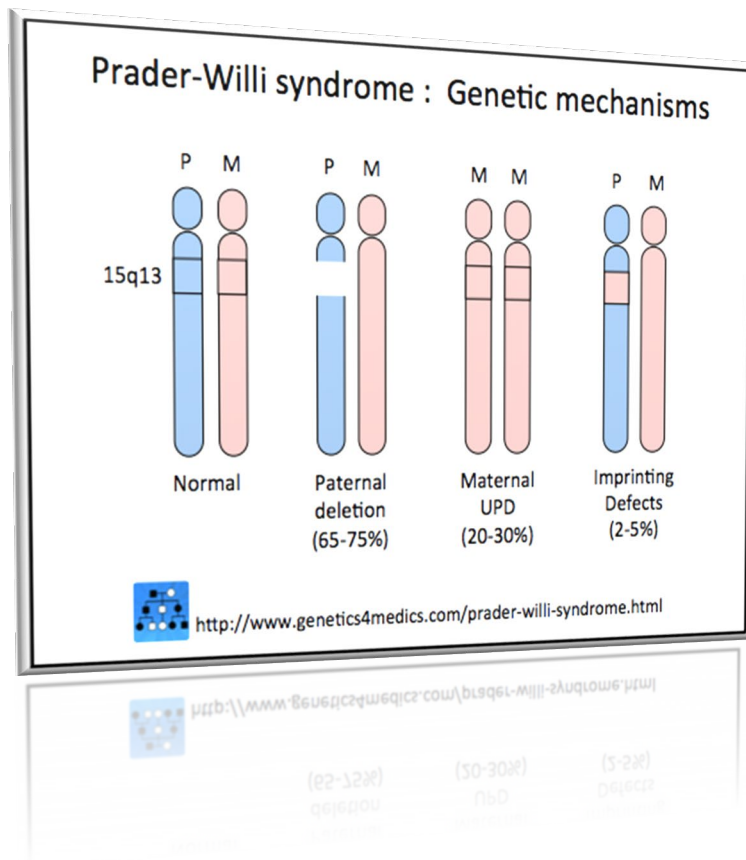
Prader-Willi syndrome (pronounced “Prah-der”) is a non-inherited genetic disorder that results from a flaw on Chromosome 15

PWS occurs randomly in 1 in 15,000-20,000 live births, or approximately 35,000 people in U.S. Babies throughout the world are born with PWS. PWS does not discriminate; it affects all genders and races equally

Previously, life expectancy for a person with PWS was short. However, with earlier diagnosis and better management, individuals with PWS are living into their 70s



## Overview of PWS and Genetics



UPD (uni-parental disomy): both chromosome 15s come from mother, none from father

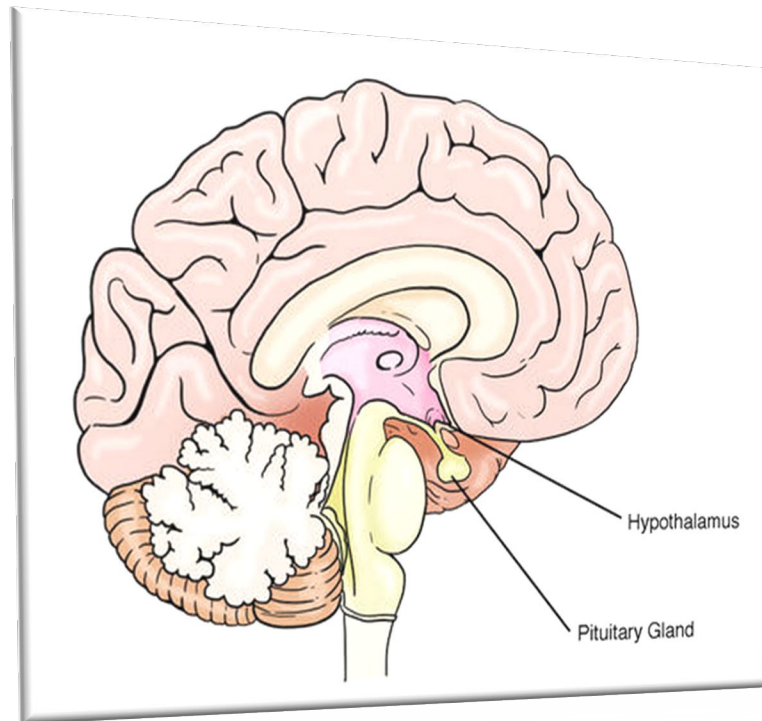
Paternal Deletion: loss of a small piece of chromosome 15 from the father

Imprinting Defect: both chromosome 15s are present, however the 15q11-q13 region of the paternal chromosome is inactive





## Hypothalamic Dysfunction

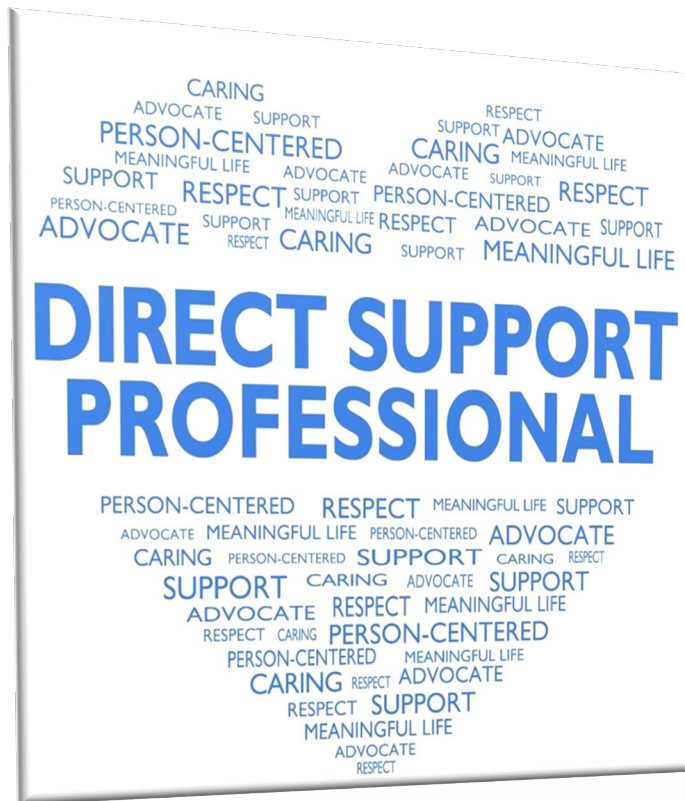


PWS symptoms  
are due to a  
dysfunction of the  
hypothalamus

Although hypothalamic  
dysfunction is believed  
to cause the symptoms  
of PWS, it is not yet  
clear how the genetic  
abnormality causes  
the dysfunction



## The Role of Direct Care Professionals



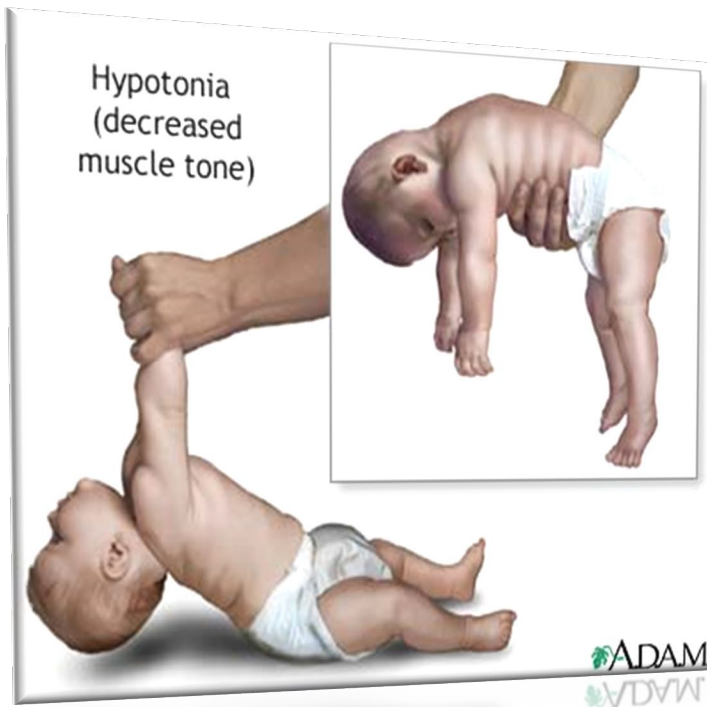
Do **NOT** be afraid  
to speak up and  
advocate on their  
behalf

**YOU** are a critical  
part of the health  
care team

**YOU** know the  
individual with PWS  
better than the  
healthcare provider  
evaluating them



## Multi-Phase Syndrome – Phase 1



PWS occurs in  
utero

Reduced prenatal  
movement and  
growth

Floppy or rag doll  
appearance in  
infancy

Low muscle tone  
(hypotonia)

Excessive  
sleepiness

Poor weight  
gain

# GENETICS AND MEDICAL OVERVIEW



## Phase 2: Ages 2 years – 8 years

Weight increases as normal without increased appetite and transitions to an abnormal increased insatiable appetite. Individuals become overweight/obese if caloric intake is not regulated.

## Phase 3: 8 years into adulthood

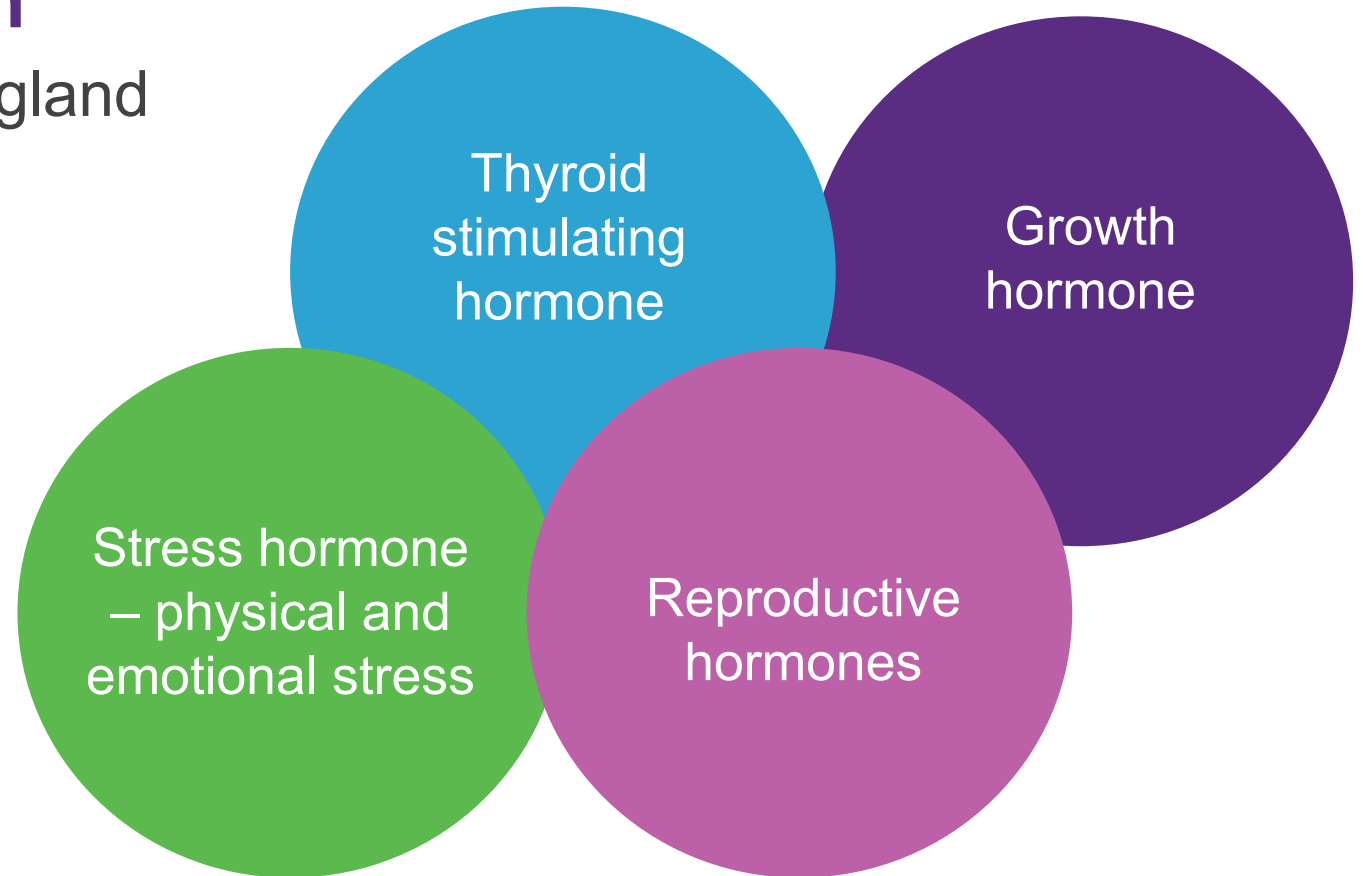
Hyperphagia – excessive eating and weight gain. Insatiable appetite, intense food seeking.





## Hypothalamic Dysfunction

Hormones interact with the pituitary gland





## Hypothalamic Dysfunction

Hypothalamic dysfunction affects their ability to:



Control appetite

Regulate their body temperature

Swallow and digest food properly

Control emotions

Learn and comprehend

Have strong bones

Hyperphagia  
varies from  
person to  
person

In addition to hyperphagia, people with PWS also have very low metabolism

There is no FDA approved treatment available for hyperphagia yet

Behavior  
modification cannot  
be effectively used  
to address  
hyperphagia

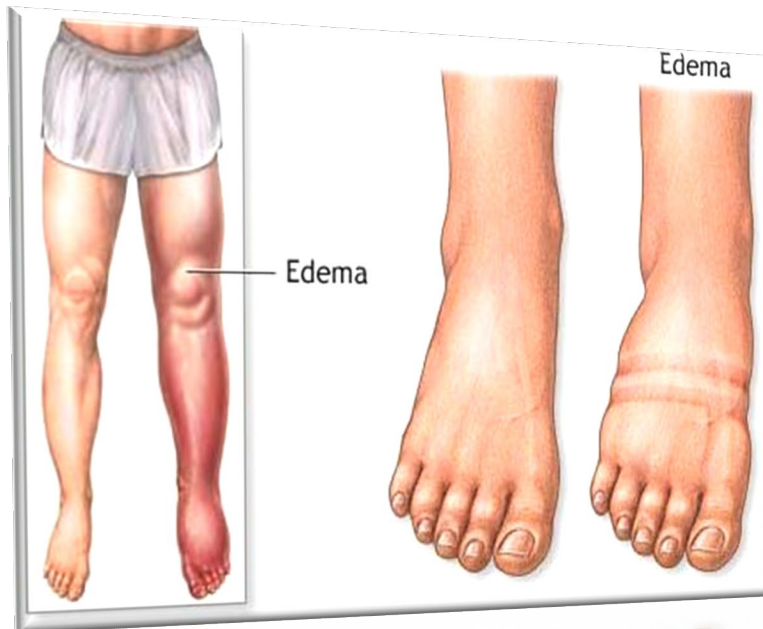
One food binge  
can lead to death:  
stomach rupture,  
stomach or colon  
necrosis

People who have hyperphagia do not reach satiety – the message of fullness does not reach the brain

Food security is a necessity!!! Motion detectors, locks and alarms are needed



## Life Threatening Obesity



People with PWS may still gain a lot of weight

Those who are obese may experience serious health conditions including:

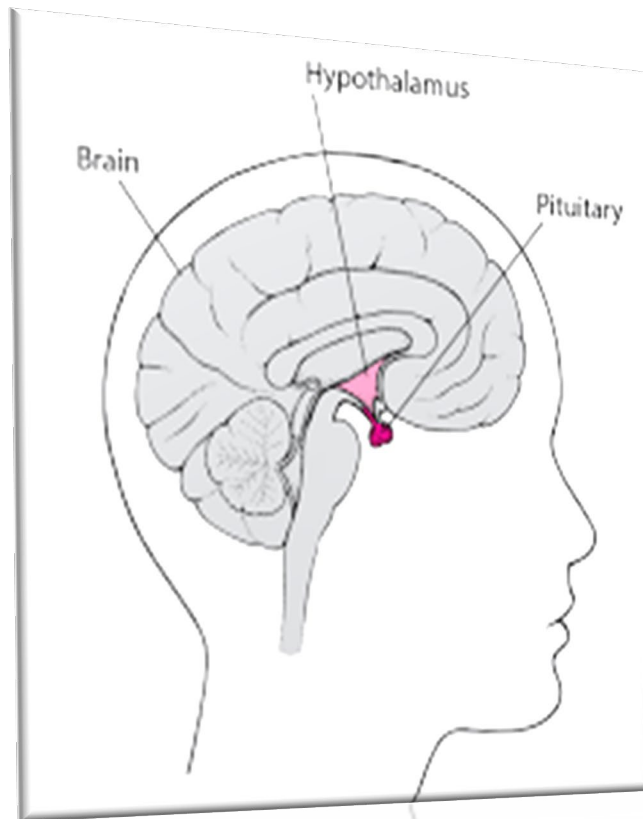
Heart and circulation problems such as high blood pressure and edema

Respiratory concerns – physical restraints are discouraged

Diabetes



## Central Adrenal Insufficiency (CAI)



Adrenal glands make three hormones: cortisol, androgens and aldosterone

CAI is caused by the lack of pituitary Adrenocorticotrophin Hormone (ACTH) which acts upon the adrenal gland hormones

A stress dose of cortisol may be warranted after surgery or in times of stress





## Medications – Adverse Reactions



Metabolism of medication is impaired

People with PWS may have unusual reactions to standard dosages of medications

Extreme caution is needed when using psychotropics and narcotics



## Increased Pain Tolerance

Lack of typical pain signals may mask infection and injuries

Complaints of pain should be taken seriously

Significant fractures can occur from simple falls. Monitor for decreased movement, guarding and behavior changes

# GENETICS AND MEDICAL OVERVIEW



## Advocacy Tips For High Pain Threshold



**Insist on x-ray**

**Medical Alerts  
Booklet**

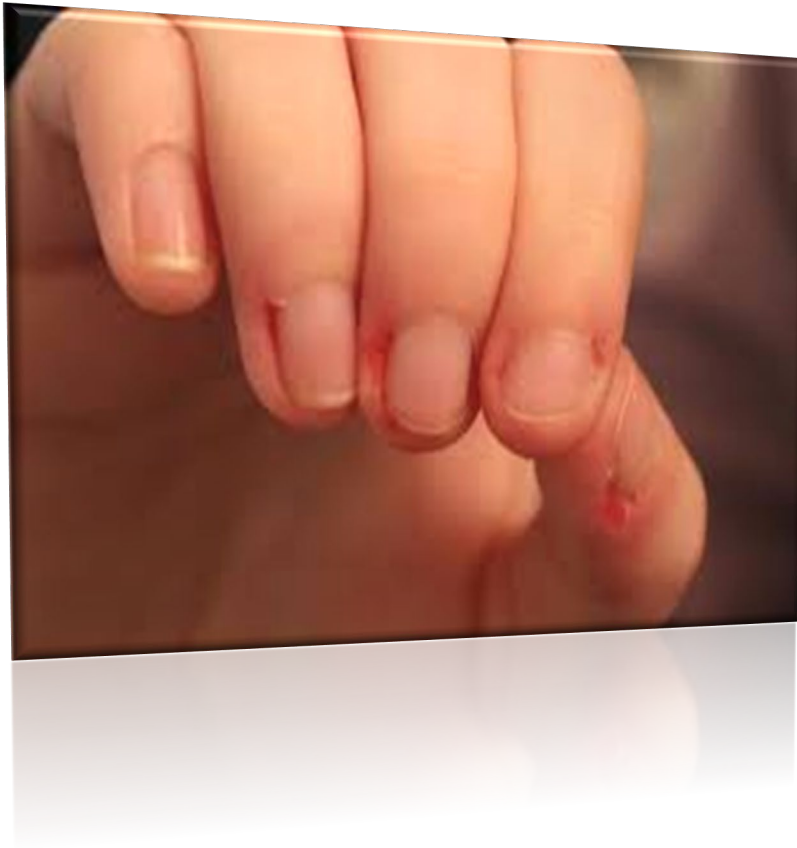
**Monitor Every  
4 hours for 24  
hours**

**Immediate  
Need**

**Persistent  
Pain**



## Skin Lesions and Bruises



Skin picking is common in PWS and can lead to open sores and infections

Communication is key

Skin picking lesions and bruising can lead to suspicion and allegations of physical abuse

Individuals with PWS bruise very easily. All bumps, falls and restraints must be reported to nursing personnel for skin checks



## Swallowing and Choking

**PACE  
AND  
CHASE**

All employees  
should be trained  
in the Heimlich  
maneuver

Provide foods  
with moisture i.e.  
soup, sauces

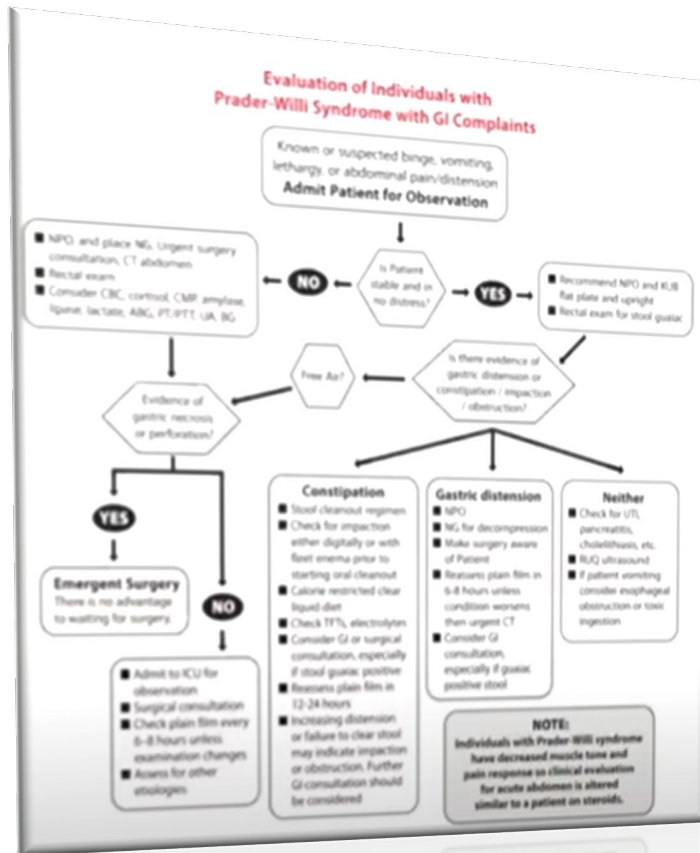
Most people with  
PWS have low  
saliva production  
contributing to  
swallowing  
difficulties

A 2016 study showed  
that food / medication  
was left in the throat of  
every study participant  
without their  
knowledge





## Gastrointestinal Concerns



Gastroparesis:  
slow emptying of  
stomach contents –  
can lead to bloating  
and distention

Vomiting – vomiting  
is rare in PWS and  
should be treated  
as a medical  
emergency

High fiber and fatty  
foods are difficult to  
digest – limit these  
types of foods

Remain upright  
after eating

Avoid large  
meals

Bring the GI  
algorithm with  
you to ER



## Gastrointestinal Concerns

### Constipation



Poor muscle tone  
and sluggish  
contraction of the  
muscles in the  
intestinal system

Low physical  
activity

Poor water /  
fluid intake

Medications  
can cause  
constipation

Not enough  
fruits or  
vegetables



## Gastrointestinal Concerns









Individuals with PWS may have bowel movements daily and still be constipated

Changes in bowel habits should be reported to nursing

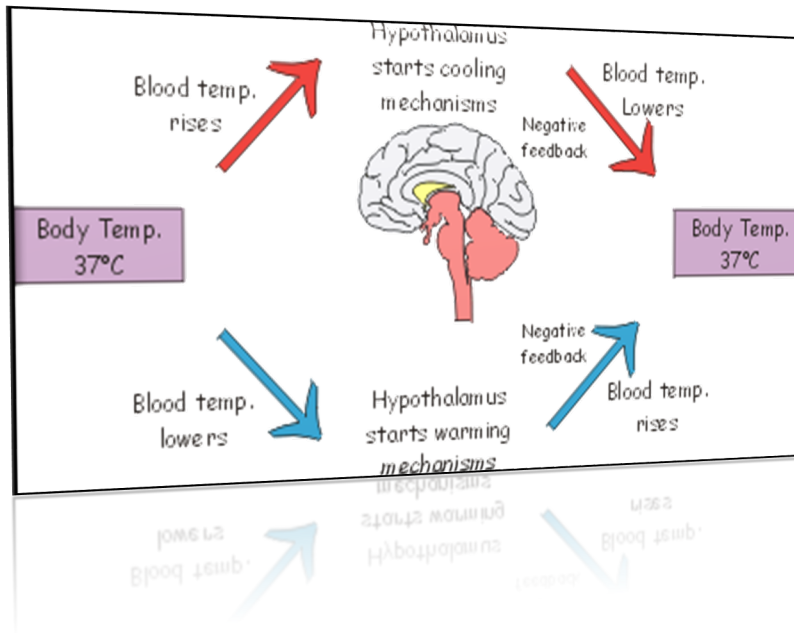


## Bristol Stool Chart

BRISTOL STOOL CHART			
	Type 1	Separate hard lumps	Very constipated
	Type 2	Lumpy and sausage like	Slightly constipated
	Type 3	A sausage shape with cracks in the surface	Normal
	Type 4	Like a smooth, soft sausage or snake	Normal
	Type 5	Soft blobs with clear-cut edges	Lacking fibre
	Type 6	Mushy consistency with ragged edges	Inflammation
	Type 7	Liquid consistency with no solid pieces	Inflammation



## Body Temperature Regulation



May overheat easily

Lack of fever when ill – this cannot be a measurement for illness

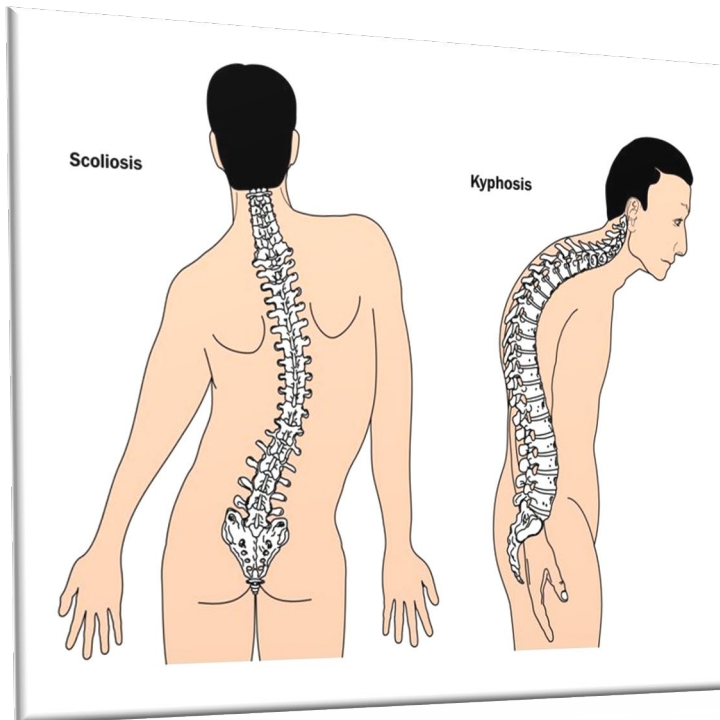
Your hypothalamus regulates your body temperature

Most people with PWS have a lower baseline body temperature i.e. 96-97





## Osteopenia / Osteoporosis



High risk for  
scoliosis and  
kyphosis

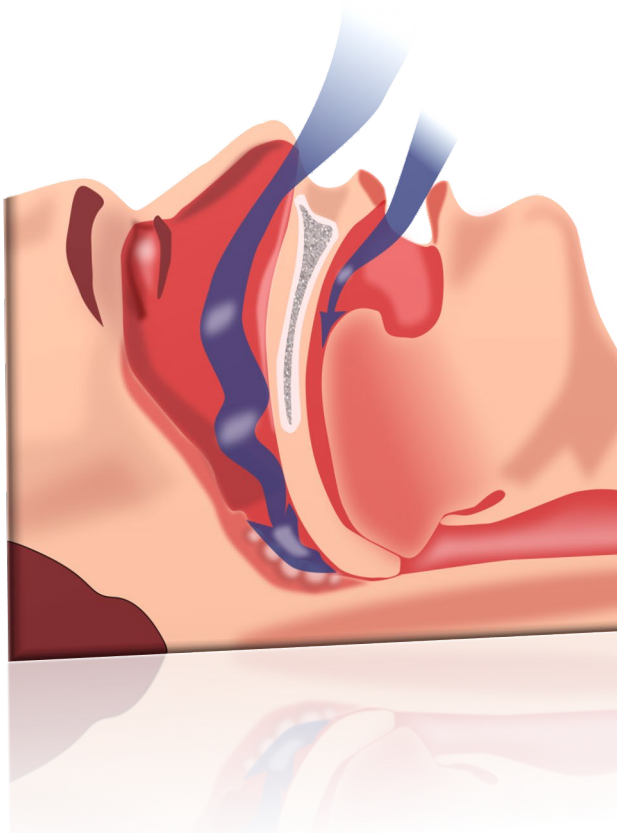
Orthopedist are  
crucial members of  
the health care  
team

Hormone  
deficiencies cause  
poor bone density

Medication /  
hormones are  
needed to strengthen  
bones i.e. calcium,  
growth hormone,  
estrogen and  
testosterone



## Respiratory Concerns



Obstructive Sleep Apnea – due to a blockage in airway, no air enters the lungs

Central apnea – the complete stopping of breathing during sleep

Hypotonia, weak chest muscles, swallowing abnormalities and sleep apnea are all common

Central hypoventilation – disorder of decreased breathing rate or depth, particularly during sleep



## Respiratory Issues are the Number One Cause of Death in PWS



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- Obesity
- Sleep Apnea
- Weak Chest Muscles
- Scoliosis
- Medications
- Choking



## Tips for Respiratory Concerns



**Manage  
Weight**

**Food Security**

**Sleep Studies**

**CPAP/Bi-PAP**

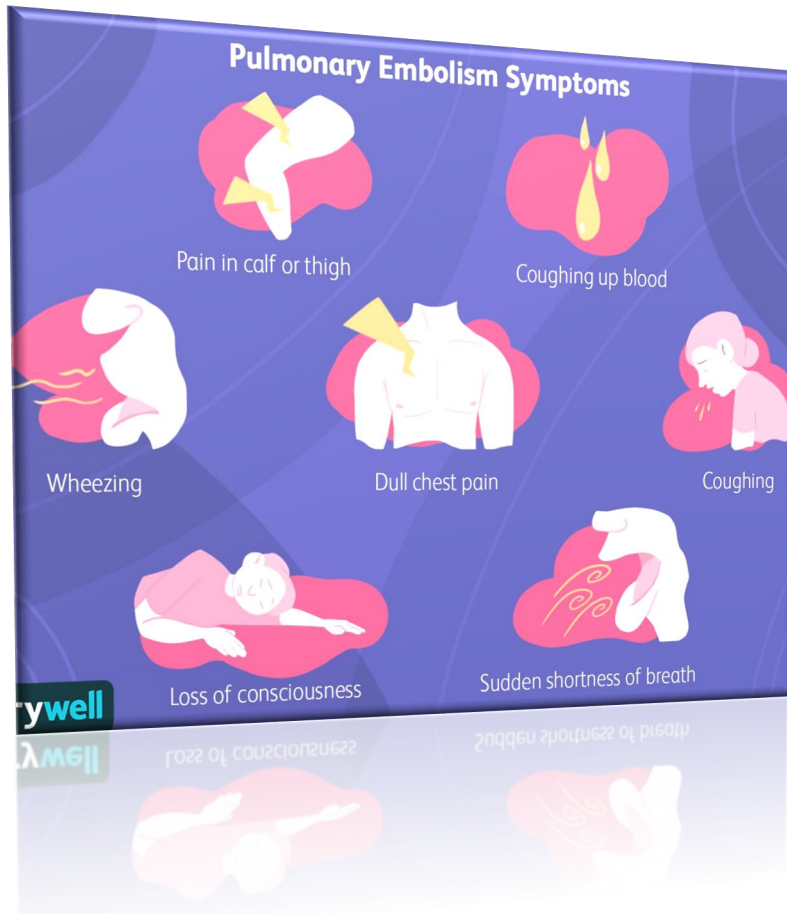
**Pulse Oximeter**

**Start Low &  
Go Slow**

**Peer-to-Peer**



## Pulmonary Embolism



Individuals with PWS are at higher risk for pulmonary embolism (blood clot in lungs)

DVT prophylaxis should be considered for all obese individuals





## Hospitalizations – Special Considerations



Extra support, advocacy and education of the hospital staff

Many diagnostic tests require fasting – food security **MUST** be in place

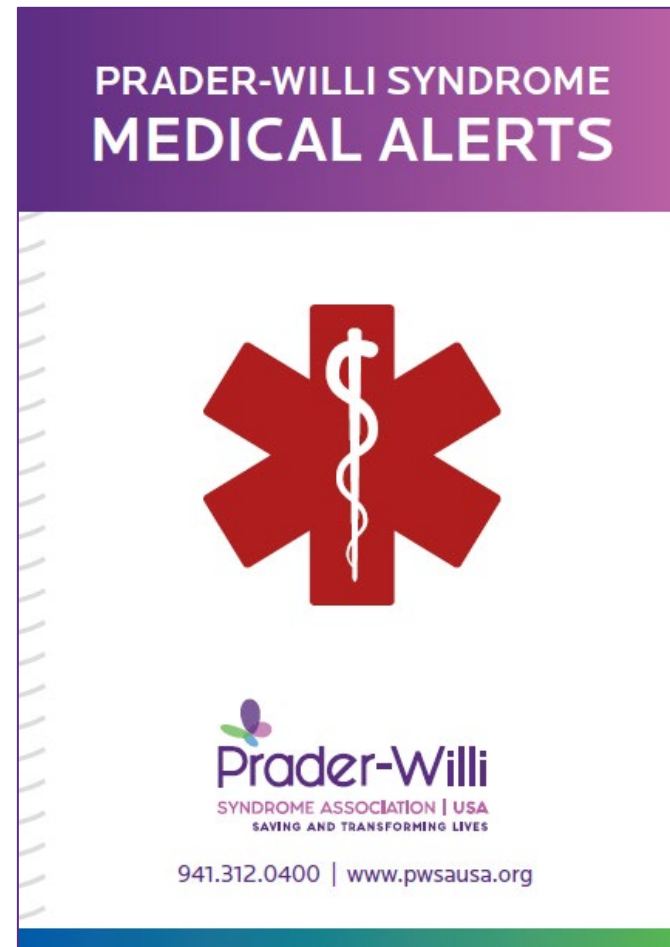
It is not uncommon for people with PWS to gain weight while hospitalized

Many people with PWS have cognitive limitations and may not be able to provide accurate information about their medical history

# GENETICS AND MEDICAL OVERVIEW



## PWSA | USA Medical Alerts Booklet





## Open Discussion

### Q & A



# RESOURCES

**PWSA | USA Medical Alert booklet**



[www.pwsausa.org](http://www.pwsausa.org)



[Info@pwsausa.org](mailto:Info@pwsausa.org)



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