
Prader-Willi Syndrome

— By Carmela de Leon —

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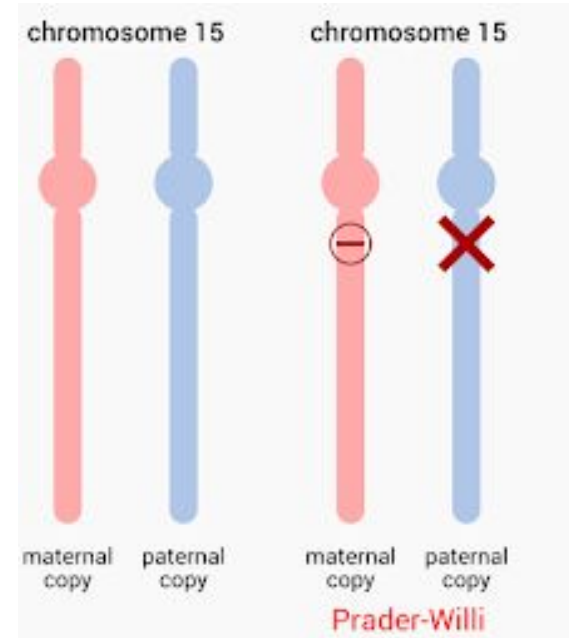
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Neurodevelopmental Variation

There are two causes of PWS:

1. Deletion: the expression of genes on paternal chromosome does **not** occur
2. Imprinting Error: lack of expression of genes on paternal chromosome

PWS affects **1 in 5,000** males and **1 in 8,000** females (Butler, 2019)



PWS has components that affect the child physically, developmentally, intellectually, and social-emotionally. These include:

- metabolism
- appetite
- growth
- cognitive functioning
- behavioral functioning



Characteristics of PWS



**Hormone
deficiencies**

**Cognitive
impairment**

**Hypothalamic
satiation
issues**

**Self
injurious
behavior**

**Muscle
weakness**

**Speech
issues**

**Obsessive
compulsive
disorder**

**Underdeveloped
sex organs**

PWS and Nutrition

Phase 0 - fetus - has restrictions on growth & minor movement

Phase 1 - infancy - poor sucking + hypotonia (ages 0-9 months)

Phase 2 - infancy - weight gain & interest in food (ages 9 to 36 months)

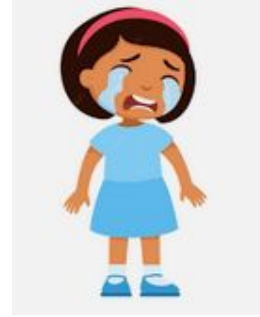
Phase 2a - 3 to 5 years - weight gain + significant change in appetite

Phase 3 - ages 5 to 13 - hyperphagia, food seeking, lack of satiety



Implications of PWS

- Life-threatening obesity
- Communication or speech issues
- Seizures
- Developmental delays



- hypotonia
- Intellectual disability
- Maladaptive behaviors
- Social isolation

Theory of Mind

Individuals with PWS are at a high risk for social exclusion and isolation

They may experience:

- Anxiety
- Depression
- Loneliness
- Cognitive decline
- Sensitivity to perceived social threats



Lycoming-Clinton v. Department of Public Welfare

WHO

An individual by the name of T.T. (who had a diagnosis of PWS)

WHAT

T.T.'s was denied disability resources due to his IQ scores from two assessments

WHERE

Pennsylvania

WHEN

2005

WHY

One psychologist gave him a score of 83 and the other a score of 100

RULING

The courts deemed that T.T. was eligible for services because IQ was not the only determining factor when assessing for a disability. They referenced the IDEA and the different components that make up an intellectual disability.



PWS support team:

Medical team
(nurse,
physician)

Social
worker

nutritionist

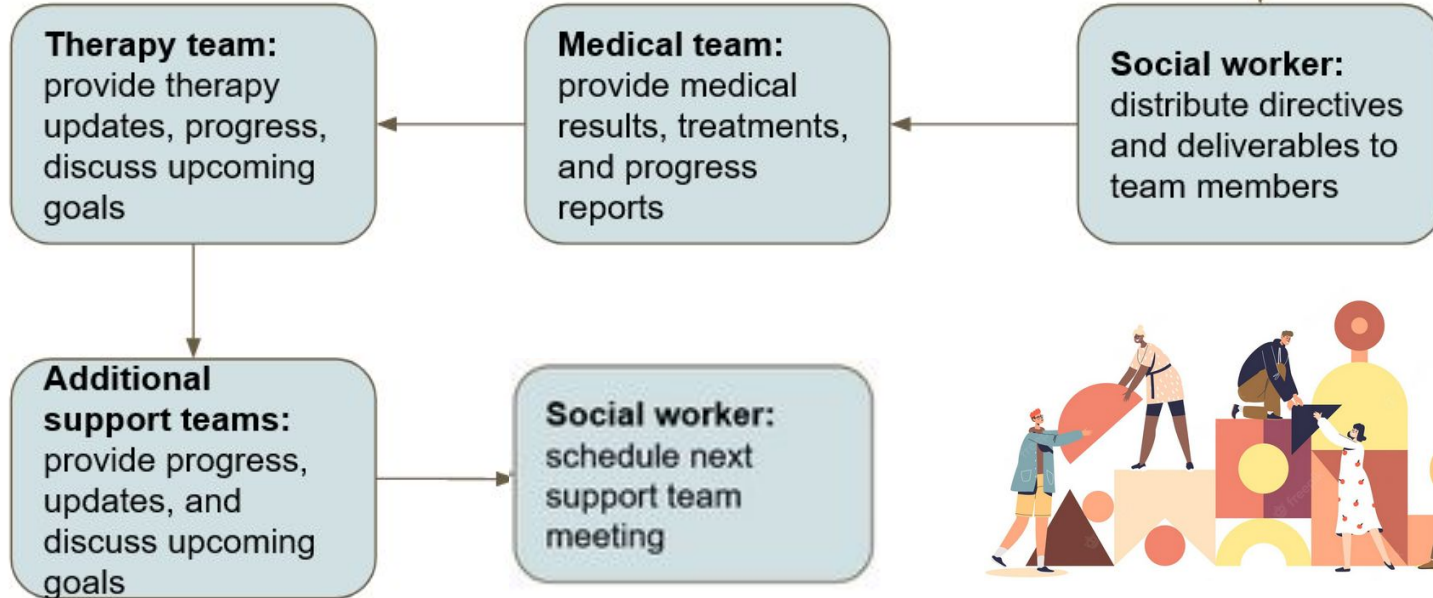
Medical specialist
(endocrinologist)

therapists

psychologist



Care Plan MODEL



Evidence Based Interventions: Therapies

Therapy Type	Purpose
Physical therapy.....	To practice motor development
Occupational therapy.....	To practice fine motor skills
Speech and language..... therapy	To practice pre-language imitation skills (making sounds), and language skills
Behavioral therapy.....	To practice adaptive behaviors and coping skills

Medical Care

Human growth hormone
(HGH)

Endocrinologist for hormone
imbalance

Surgical care

Orthopedic physician for
scoliosis

Medication (such as modafinil)



Care Plan Example

Goal #1 - Weight

Weight Goal: 19 pounds

Date:	Nov. 5, 2022
Current Weight:	15 lbs, 2 oz.
Weight Incr. Since Last Week:	2 oz.
Notes:	Infant still struggling to adjust to new nipples, discussions of feeding tube with parents to occur on 11/8/22
Date:	Nov. 12, 2022
Current Weight:	15 lbs, 4 oz.
Weight Incr. Since Last Week:	2 oz.
Notes:	Spoke with parents reg. feeding tube, they would like to try the nipples out the rest of the month and then use the tube if the infant does not make it to 16 lbs. by EOM



Romans 5:3-5 states that “we also rejoice in our afflictions, because we know that affliction produces endurance, endurance produces proven character, and proven character produces **hope**.”

there's light at
the end of the
tunnel



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