Medical and Psychiatric Management of Prader-Willi Syndrome

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Introduction

- Prader-Willi Syndrome (PWS) is a rare neurodevelopmental disorder which occurs in roughly 1 in 22000 births. (1)
- PWS is caused by abnormal parent-specific imprinting within the Prader-Willi critical region 15q13 on chromosome 15. (2)
- Most cases of PWS are due to loss of a segment of the paternal chromosome 15. (2)

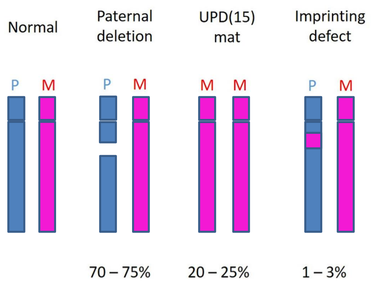

Figure 1. Genetic mechanisms of Prader-Willi Syndrome. The three main mechanisms that result in PWS are paternal deletion, maternal uniparental disomy (UPD) of the 15th chromosome, and imprinting defects. (2)

Case Report

19 yo male with PWS, moderate intellectual disability, Generalized Anxiety Disorder, Impulse control disorder, Major Depressive Disorder with comorbid diabetes. He is currently treated with Fluoxetine 60 mg daily, Oxcarbazepine 300 mg twice a day, Lithium 600 mg twice a day, Bupropion XL 300 mg daily, Metformin 2000 mg daily, NovoLog 36 mg daily, and Lantus 33 mg daily. His height was 183 cm and his weight was 152 kg with a BMI of 45, indicating morbid obesity.

Overview of Prader-Willi Syndrome

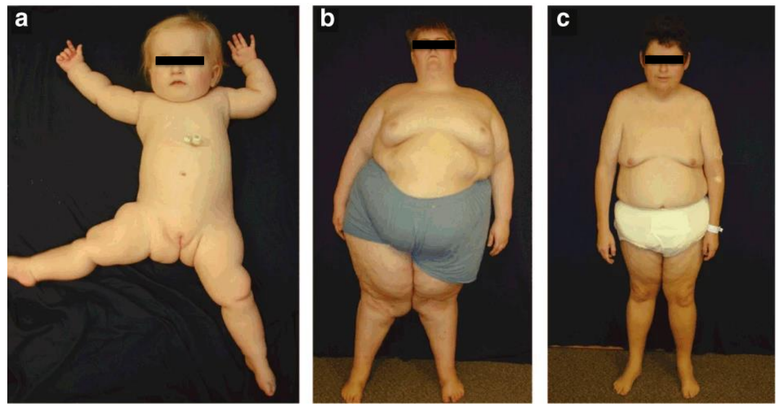

Image used with permission from (2).

Figure 2. Patients with PWS. PWS presents with multiple comorbidities.

- Management of PWS requires multidisciplinary care.
- PWS presents in infancy as hypotonia and failure to thrive.
- Children with PWS classically have the following characteristics:
  - developmental and cognitive delay (4)
  - defects in satiety leading to obsession with food, hyperphagia, and obesity (5)
  - Hypogonadism leading to sex hormone deficiency and impaired sexual development (5)
  - Growth hormone deficiency leading to short stature (5)
  - Obstructive sleep apnea secondary to obesity (5)
  - Osteoporosis or scoliosis from deficient calcium intake (5)
- Life expectancy of a patient with PWS depends on obesity and comorbidities. Medical supervision becomes increasingly important after age 40. (3)

Psychiatric considerations of PWS

Figure 3. Child Behavior Checklist DSM orientated scales

	PWS Mean T score	Control Mean T score
DSM oriented scales		
Affective problems	66.22	60.08*
Anxiety problems	60.37	60.88
Somatic problems	63.05	52.00*
Attention deficit/hyperactivity problems	63.53	63.13
Oppositional defiant problems	61.58	58.58
Conduct problems	62.50	63.17

Table reproduced from (1)

Figure 3. Children with PWS have more severe affective and somatic problems compared to controls (2). *p<0.05. Affective disorders include Major depressive disorder and Bipolar disorder. Somatic problems are physical complaints such as pain, weakness, or shortness of breath.

References

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