Prader-Willi Syndrome

Prader-Willi syndrome is caused by maternal imprinting after the deletion of a gene located on the long arm of chromosome 15. This condition is characterized by hyperphagia, truncal obesity, hypogonadism, neonatal hypotonia, intellectual disability, and facial characteristics such as almond shaped eyes, a thin upper lip, and a narrow bifrontal diameter.



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Mechanism

Maternal Imprinting

Mother Imprinting a Stamp

Imprinted genes are those wherein one copy is silenced and the other is expressed. The outcome depends on whether the gene was inherited from the mother or father. Imprinted genes are those wherein one allele is silenced, meaning it's not expressed, while the other allele is expressed. Disease can occur if a mutation occurs on the active allele while the other allele is imprinted since there will no longer be a functional copy of the gene in that region. In Prader-Willi syndrome, the maternal allele is imprinted, or silenced, at baseline. The disease then results if the paternal allele is mutated or deleted. A separate genetic abnormality called uniparental disomy involves the inheritance of two imprinted maternal alleles and causes Prader-Willi syndrome in up to 25% of cases.

Deletion of Chromosome 15q (Paternal Deletion)

Cutting off Chromosome of (15) Quinceanera-dress and Missing Father

The affected genes are located on the long arm of chromosome 15, specifically at the locus 15q11-13. Recall that "q" refers to the long arm of a chromosome, while "p" represents the short arm. A mutation on the paternal 15q allele, whether through a deletion, inversion, etc., will lead to Prader-Willi syndrome.

Signs And Symptoms

Hyperphagia

Hiker-fajita

A classic symptom of this syndrome is hyperphagia, or increased appetite and impaired satiety. These individuals will seek out food at all costs and binge eat constantly. This is theorized to be caused by increased serum levels of ghrelin and other peptide hormones that trigger hunger.

Truncal Obesity

Trunk Fat

Dysfunction of the HPA (hypothalamic-pituitary-adrenal) axis leads to growth hormone deficiency, and patients typically have short stature with accompanying truncal obesity. In conjunction with their uncontrollable eating habits, morbid obesity is a common complication of this syndrome.

Hypogonadism

Hippo-gonads

Gonadal development depends on pituitary hormones like follicle-stimulating hormone (FSH) and luteinizing hormone (LH). These pituitary hormones are dependent on the hypothalamic hormone GnRH, or gonadotropin-releasing hormone. Since this entire axis is disrupted in Prader-Willi patients, gonadal development is impaired, and complications like cryptorchidism, lack of secondary sexual characteristics, and osteoporosis can occur.

Undescended Testicles (Cryptorchidism)

Testicles Not Descending

About two-thirds of males affected by Prader-Willi syndrome exhibit cryptorchidism. Treatment involves either beta-hCG administration or surgical orchiopexy. However, these patients often remain sterile.

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Intellectual Disability (Mental Retardation)

Tar Covered Book

Patients with Prader-Willi syndrome most often fall within the borderline low, mild, or moderate intellectual disability categories. Their intelligence deficit is not as severe as with Angelman syndrome, but these patients will usually still require lifelong assistance.

Neonatal Hypotonia (Floppy Baby)

Neon Floppy Baby

Prader-Willi infants exhibit poor reflexes and tone, leading to the description of “floppy baby.” These newborns also present with eating and breathing issues during the neonatal period. Choking episodes are common, and parents should be educated on how to handle these emergencies.

Almond Shaped Eyes

Almond Eyes

A characteristic also used to describe the eyes of Down syndrome patients, almond-shaped eyes are associated with Prader-Willi patients.

Narrow Bifrontal Diameter

Narrow Bifocals

This term describes a “narrowed face,” as measured from temple to temple, that is typical with Prader-Willi patients as well as with other syndromic disorders.

Thin Upper Lip

Thin Upper Lip

A characteristic also used to describe the lips of Fetal Alcohol Syndrome patients, a thin upper lip is associated with Prader-Willi patients.

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