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Hereditary Genetics: Current Research

Commentary

A Short Description of Prader-Willi Syndrome

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DESCRIPTION

PRAHdur VILe or Prader-Willi Syndrome is a rare hereditary disorder that causes a variety of physical, mental and behavioral problems. An important feature of Prader-Willi Syndrome is a constant feeling of hunger. This usually starts around the age of two. People with Prader-Willi Syndrome always want to eat because they don't feel full (Polyphagia) and usually have difficulty controlling their weight. Most of the complications of Prader-Willi syndrome are due to obesity. Optimal management by a team approach, various professionals work with you to manage the symptoms of this complex condition, reduce the risk of complications and improve the quality of life of loved ones with Prader-Willi Syndrome.

Symptoms

Signs and symptoms of Prader-Willi Syndrome vary from person to person. Symptoms can change slowly from childhood to adulthood.

Infants

The signs and symptoms that may appear from birth are:

Reduced muscle tone: The main sign of infancy is a decrease in muscle tone (floppy muscle syndrome). Babies may rest straight instead of restraining the elbows and knees, feel tired when held, or feel like a stuffed animal.

Outstanding facial features: Children can be born with almond-shaped eyes, temples that narrow their heads, a downward-facing mouth, and a thin upper lip.

Poor suction reflex: Infants may have reduced suction reflex due to reduced muscle tone. Poor breastfeeding can make breastfeeding difficult and can lead to failure to thrive.

Generally poorly responsive: Babies may appear to have abnormal fatigue, poor response to stimuli, difficulty waking up and crying.

Underdeveloped genitals: Men can have a small penis and scrotum. The testicles may be small or may not descend from the abdomen to the scrotum (cryptorchidism). In females, the clitoris and labia may be small.

Early childhood to adult

Other features of Prader-Willi Syndrome occur in early childhood and last a lifetime and require careful management. These properties include:

Craving and weight gain: A typical sign of Prader-Willi Syndrome is a constant craving for food, leading to rapid weight gain from around the age of two. Constant hunger means that you eat often and consume most of it. Abnormal foraging behavior can occur, such as storing food or eating frozen foods or garbage.

Underdeveloped genitals: A condition known as hypogonadism occurs when the reproductive organs (male testicles and female ovaries) produce little or no sex hormones. This leads to genital underdevelopment, delayed puberty, and in almost all cases infertility. Without treatment, women may not begin menstruating or at all until they are 30 years old. Also, men have less hair on their faces and may not have a complete deepening of their voice.

Poor growth and physical development: Underproduction of growth hormone can lead to shorter stature, lower muscle mass and higher body fat percentage in adults. Other endocrine problems include thyroid hormone underproduction (hypothyroidism) and central adrenal insufficiency, which prevent the body from responding properly to stress and infections.

Cognitive impairment: Mild to moderate intellectual disability, such as thinking, thinking, and problem-solving problems, is a common feature of disability. Even people without significant intellectual disabilities have some learning disabilities.

Delayed development of athletic performance: Toddlers with Prader-Willi Syndrome are slower than other children and often reach milestones in physical activity such as sitting and walking.

Language issues: Speech is often delayed. Inadequate verbal clarity can be a problem that lasts into adulthood.

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Operational issues: Children and adults can be stubborn, angry, dominating, or manipulative. They can develop tantrum, especially when they are refused to eat, and cannot tolerate daily changes. They can also develop obsessive-compulsive and repetitive behaviors. Other mental disorders such as anxiety and excoriation can develop.

Insomnia: Children and adults with PraderWilli syndrome can suffer from sleep disorders such as impaired normal sleep cycles and sleep apnea. These disorders can cause excessive daytime sleepiness and exacerbate behavioral problems.

Other signs and symptoms: These include small limbs, spinal curvature (scoliosis), hip problems, decreased salivation, myopia and other visual impairments, thermoregulatory problems, high tolerance to pain, or lack of hair or eye pigmentation. (Dye loss) is included and the skin is pale.