Prader-Willie Syndrome

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Definition

Prader-Willie Syndrome (PWS) is a genetic disorder found in l per l0-l5,000 live births. It is caused by the father's genetic material missing on the l5th chromosome, or by duplication of the mother's genetic material. This can happen in all races and is not inherited in families.

Introduction

The diagnosis of Prader-Willi Syndrome (PWS) used to be based on how a person looked or acted. Now, the diagnosis can be made by genetic testing.

There are common features and characteristics of people with PWS. They are often short with almond-shaped eyes, have small hands and feet, have thick saliva and have small genitals. They may have scratches and scabs from picking at their skin.

Babies and people with PWS have poor muscle tone and are "floppy". They are poor feeders as babies, due to a weak suck. They often have a weak cry and don't move around very much. As they grow, they may crawl, sit, walk, and talk at much later times, due to their muscle weakness.

People with PSW can have weak eye muscles, causing them to be very near-sighted or to have crossed eyes called strabismus. They might have poor balance and fall easily.

People with PWS may have some learning disabilities or may have some mental retardation. I.Q. scores range from 40 – ll0, with 70 as average.

People with PWS are often gifted with art or making crafts and have good long-term memory. They often understand language more than they are able to express. Many are good readers. Thinking in abstract terms, for example, about time, math, or money, can be difficult. Short-term memory is also affected by PWS. At times, people with PWS simply may forget information they have learned or have been told. Information may need to be given many times.

People with PSW have hyperphagia, which is being unable to control their appetite. This happens because the part of the brain that controls appetite is not working well in people with PWS. A person with PWS never feels full and is always looking for more food. This usually starts around ages 2-5 when children become mobile and can get to food on their own. A person with PW also has much slower metabolism and can gain weight much more quickly than the general population. Some people with PWS feel driven to obtain as much food as possible. This can be very dangerous as people can become hugely obese, with weight up to 400 pounds, leading to heart problems, high blood pressure, and diabetes mellitus.

There are other behaviors that might be found in people with PWS, due to involvement of the whole central nervous system. They might be very inflexible. Requiring a set routine. They might say the same thing over and over again, called perseveration. They might repeat the same activity over and over again, called obsessive-compulsion. They might be stubborn or if stressed or angry, become aggressive.

People with PWS also sometimes collect and stash away things like food, paper, yarn, magazines, etc., called hoarding. There is also a tendency for people with PWS to injure themselves by skin picking, biting or pulling off nails, pulling out their hair, pushing pins or tacks through their skin, biting their lips, or other forms of self-injury. They often pick at these self-administered sores, causing open sores. These open sores can bruise easily. Which can wrongly lead to the suspicion of physical abuse.

Diagnosis

The diagnosis can be made by genetic testing.

Prevention and Treatment

PWS cannot be prevented at this time.

People with PWS need lifelong services and supports. This is a syndrome that has no cure. A person cannot be "taught" out of the health or behavioral problems caused by PWS.

The services for a person with PWS might include the following:

- **Food management** – external management of access to food. This involved environmental controls, such as locked kitchens and 24-hour supervision. If left alone, most persons with PWS will start looking for something to eat.
- **Living arrangements** – highly structured living arrangements, designed to meet the individual need of a person with PWS.
- **Money** – assistance is managing money and access to money. Money is food to a person with PWS.
- **Behavioral supports** – behavioral supports to assist them in managing their behaviors and emotions.
- **Nutritional plan** – nutritional and dietary plans for weight-reduction, if necessary, or maintenance of a healthy weight. Most persons with PWS need low-fat diets of around l,000 calories per day.
- **Health care** – a health care plan by a health provider familiar with the syndrome and regular medical monitoring.
- **Respite** – respite for families who have their sons or daughters at home.
- **Education** – family and professional education concerning PWS.
- **Family support** – support for the families of the person with PWS. This syndrome has an impact on every member of the family.

Emergency Situations – What can go wrong?

- **Breathing concerns** – due to poor muscle tone in the chest, people with PWS are at risk for breathing problems and lung infections. People with PWS can have short periods where they stop breathing during sleep, call sleep apnea.
- **No vomiting** – people with PSW usually do not vomit. If someone with PWS does begin vomiting, it may be the sign of a serious illness. Also, if a person’s stomach becomes large or bloated, they should be checked right away.
- **Not able to feel pain** – many people with PWS do not feel regular pain due to lack of pain signals. They might be very sick before starting to say that anything is wrong. If there is any change in behavior or condition, the person should be medically checked.
- **Medications** – people with PWS may not be able to handle normal dosages of medications. Medicines that cause drowsiness and those used to be rid of extra water in the body should be used with great caution.
- **Body temperature problems** – the part of the brain that adjusts our body temperature does not work well in people with PWS. A person who is quite sick with an infection may never get a fever. On the other hand, minor illness or receiving anesthesia has caused severe fever.
- **Hyperphagia/uncontrolled appetite** – uncontrolled appetite can lead to a life-threatening weight gain. People with PWS must be supervised 24 hours per day in all settings where food could be obtained.

Conclusion

Prader-Willi Syndrome is a genetic disorder whose diagnosis is made through genetic testing. There are many serious health concerns associated with this syndrome so early diagnosis and treatment is very important. There is no cure for this syndrome and those with PWS will require lifelong services and supports.

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