Fact Sheet :

Prader-Willi Syndrome

What is Prader-Willi Syndrome?

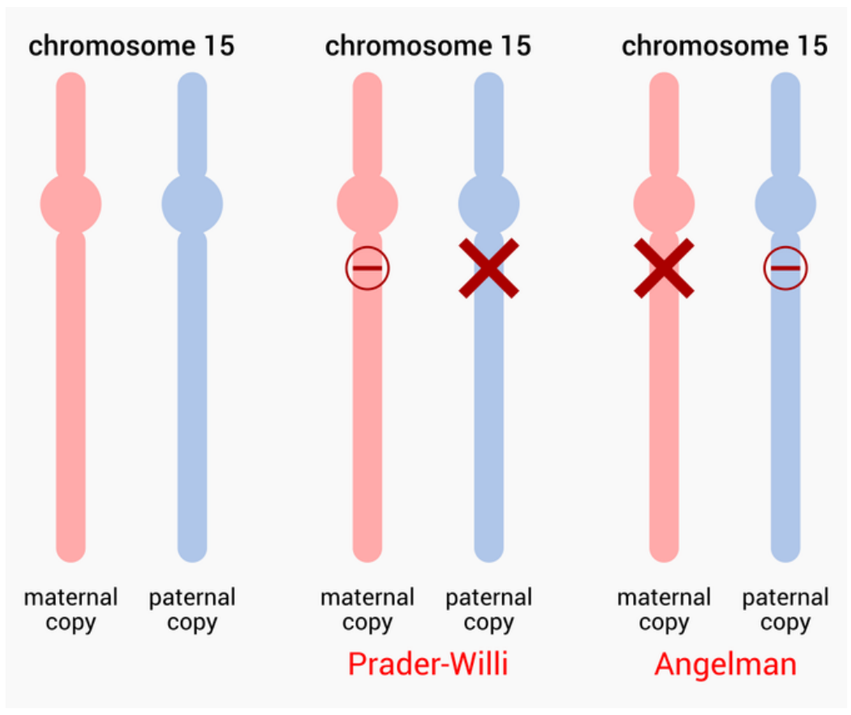
Prader-Willi syndrome is a rare genetic condition causing a wide range of physical symptoms, learning difficulties and behavioural challenges. It's usually noticed shortly after birth. Around 70% of cases of Prader-Willi syndrome are the result of missing genetic information from the copy of chromosome 15 inherited from the father. This is referred to as "paternal deletion". It is thought to affect part of the brain called the hypothalamus, which produces hormones and regulates growth and appetite.

It is thought that around one in 10,000 to 20,000 children are born with the syndrome, with females slightly more likely to have the condition than males and all ethnic backgrounds. It's extremely rare for parents to have more than 1 child with Prader-Willi syndrome.

The condition is named after Swiss physicians Andrea Prader and Heinrich Willi who, together with Alexis Labhart, described it in detail in 1956.

Symptoms of PWS

- excessive appetite & overeating, which can easily lead to dangerous weight gain
- restricted growth (children are much shorter than average)
- floppiness caused by weak muscles (hypotonia)
- learning difficulties
- lack of sexual development
- behavioural challenges, such as emotional outbursts and physical aggression
- Facial features (narrow temple distance & nasal bridge, almond-shaped eyes, mild squint, thin upper lip with downturned mouth)



PWS vs Angelman

PWS is similar to Angelman Syndrome as they both have missing genetic information on chromosome15, but differ in that the missing in formation comes from the father in PWS and the mother in Angelman's Syndrome

DID YOU KNOW?

Mayim Bialik, from the hit TV show 'The Big Bang Theory', earned her PhD in Neuroscience, specialising in Obsessive-Compulsive Disorder in adolescents with Prader-Willi Syndrome.

Diagnosis, treatment & prognosis

Prader-Willi syndrome can usually be confirmed by carrying out genetic testing. There is no cure for PWS but many patients will benefit from a supervised diet. Some symptoms can be treated with hormone therapy.

Someone with the syndrome can eat much more than other people and still feel hungry so it is essential that they follow a healthy, balanced eating plan, avoiding sweet treats and high-calorie items. Limiting food intake can be challenging for someone with PWS and may cause them to become frustrated when they want extra food, and their hunger can make them hide or steal food.

Most adults with Prader-Willi syndrome are not able to live fully independent lives, such as living in their own home and having a full-time job as environments and situations are too demanding.

It's almost unknown for either men or women with PWS to have children as their reproductive organs do not develop normally. But sexual activity is usually possible, particularly if sex hormones are replaced.

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