

## About PWS

### Main characteristics

Prader-Willi syndrome (PWS) was first described in 1956 by Swiss doctors, Prof. A Prader, Dr A Labhart and Dr H Willi, who recognised the condition as having unique and clearly definable features. These features are, as medically described:

- **Hypotonia:** weak muscle tone, and floppiness at birth.
- **Hypogonadism:** immature development of sexual organs and other sexual characteristics.
- **Obesity:** caused by excessive appetite and overeating (hyperphagia), and a decreased calorific requirement owing to low energy expenditure levels. (Obesity is not normally a feature of those whose food intake is strictly controlled.)
- **Central nervous system and endocrine gland dysfunction:** causing varying degrees of learning disability, short stature, hyperphagia, somnolence (excessive sleepiness), and poor emotional and social development.

### Other characteristics

Many people with PWS also exhibit characteristic facial and other physical features. These include: almond-shaped eyes, a narrow forehead (measured across), a down-turned mouth with a triangular-shaped upper lip, and small hands and feet.

People with PWS have poor large muscle strength, often coupled with poor coordination and balance. Muscle tone can be improved with appropriate therapy and exercise. Small muscle strength is usually better. Some of the physical features can be improved with **growth hormone treatment** which improves height, hand and foot length, face shape and muscle tone.

Most people with PWS have borderline or moderate learning disabilities. Some people have recorded IQs of 90 or above, whilst a minority have severe learning difficulties. The average IQ for people with PWS is around 60 - 70, although individuals may sometimes find it difficult to perform at their IQ level, as emotional and social skills are often less developed. Reading and writing skills are usually considerably better than number skills and abstract thinking. Individuals often excel at sedentary activities such as jigsaw puzzles, "Wordsearch", sewing, drawing and colouring.

### Cause

PWS is caused by an abnormality on chromosome 15 which occurs around the time of conception. The majority of cases (about 70%) are caused by a deletion (a missing small piece of genetic material) on the chromosome 15 inherited from the father, whilst about 25% are caused by inheriting two chromosome 15s from the mother, instead of one from the mother and one from the father (uniparental or maternal disomy). A small minority of people have a translocation or imprinting irregularity involving chromosome 15, which can be of hereditary origin. The recurrence risk of PWS in a family for those who have children with deletion or disomy types of PWS is extremely small. No blame attaches to either parent - currently PWS is thought to be a purely accidental occurrence and is not a result of any actions or inactions on the part of parents. See also: **Genetics**

[http://www.pwsa.co.uk/assets/files/About%20PWS/Genetics\\_text.pdf](http://www.pwsa.co.uk/assets/files/About%20PWS/Genetics_text.pdf)

## Atypical PWS

A few people have a PWS diagnosis, but do not have the typical physical features - in particular they may be taller than most people with PWS, even without growth hormone.

## Acquired PWS

PWS-like symptoms can be "acquired" by damage to the hypothalamus during a person's life, such as head injury or surgery, or from a dysfunction of the hypothalamus. In these cases, the person does not have any of the genetic abnormalities and few of the physical characteristics of PWS, but acquires some or all of the behavioural and appetite problems which are associated with the syndrome. Management techniques used with people with true PWS can also be helpful in acquired PWS.

## Incidence and Prevalence

An epidemiological study carried out in one health region in the UK in 2000 estimated birth incidence to be in the region of 1:22,000, with a lower bound (ie the lowest number estimated) for population prevalence of 1:52,000. Previous estimates have estimated a birth incidence between 1:10,000 and 1:25,000. To give an idea of how rare PWS is, the incidence of Down Syndrome is 1:715 births - about 30 times more likely to happen than PWS.

We estimate that there may be around 1500 - 2000 people with PWS in the UK of all ages; the PWSA UK is in touch with around 1,200 of these people and/or their families.

Much of the uncertainty with regard to the statistics for incidence and prevalence is due to the fact that PWS may still go undiagnosed in some people, especially older adults; also, there is currently no central register held by the government of people diagnosed with rare syndromes.

## Stages of Development: Appetite and weight gain

Seven stages of development with regard to appetite and weight gain have been described by Dr Jennifer Miller and associates. In an article in the [Journal of Medical Genetics \(2011\)](https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3285445/), <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3285445/> she describes these stages thus:

*"Phase 0 occurs in utero, with decreased fetal movements and growth restriction compared to unaffected siblings.*

*Phase 1 the infant is hypotonic and not obese, with sub-phase 1a characterized by difficulty feeding with or without failure to thrive (ages birth–15 months; median age at completion: 9 months). This phase is followed by sub-phase 1b when the infant grows steadily along a growth curve and weight is increasing at a normal rate (median age of onset: 9 months; age quartiles 5–15 months).*

*Phase 2 is associated with weight gain—in sub-phase 2a the weight increases without a significant change in appetite or caloric intake (median age of onset 2.08 years; age quartiles 20–31 months;), while in sub-phase 2b the weight gain is associated with a concomitant increased interest in food (median age of onset: 4.5 years; quartiles 3–5.25 years).*

*Phase 3 is characterized by hyperphagia, typically accompanied by food-seeking and lack of satiety (median age of onset: 8 years; quartiles 5–13 years). Some adults progress to:*

*Phase 4 which is when an individual who was previously in phase 3 no longer has an insatiable appetite and is able to feel full."*

## Infancy

Babies with PWS are very floppy at birth (hypotonia), and the ability to suck is weak or absent. Tube-feeding may be required for the first few days or weeks of life; breast-feeding is rarely initially successful. Babies show little interest in feeding during the first few months of their lives, have a very weak cry, and sleep for most of the time during the early weeks.

Male babies may have noticeably underdeveloped genital organs. Female babies may also have underdeveloped genitalia, but this is much harder to detect.

Developmental milestones such as sitting, standing, walking and talking are generally delayed, but most children with PWS are able to attain all these abilities by the time they are about 5 years old. Often, infants with PWS are very lovable and placid, and seem to draw admiration wherever they go.

## Twins

If twins are identical, both will have PWS. If they are non-identical, then one could have PWS and one not. Several families with non-identical twins are known to PWSA UK, where only one twin is affected, but the Association is also aware of a family where there are identical twins, both with PWS.

## Childhood

Some time between the ages of one and four years, most children with PWS begin to show a heightened interest in food (some do not show this interest until they are older than four) and in severe cases develop what appears to be an insatiable appetite, so that they may try to obtain food by any means possible. The degree to which this occurs varies considerably between individuals, but there is always a preoccupation with food in play and talk. This can be helped by good management, dietary control and educating the child about his or her diet.

However, if energy intake has not been carefully monitored, weight gain is rapid; hence, a lower calorie diet (energy controlled regime) is essential throughout the person's life. An appropriate exercise programme helps to keep weight levels down and also improves muscle strength. If weight is not controlled, fat accumulates in a characteristic way on the buttocks, stomach, lower trunk and thighs.

Children with PWS are generally placid and friendly, but may begin to exhibit stubborn or obsessive behaviour, and outbursts of temper if they cannot get their own way or are denied access to food.

## Adolescence

People with PWS do not usually reach full sexual development, and there have been only four known cases worldwide of a woman with PWS having a child. There are currently no known cases of a man with PWS fathering a child.

People with PWS are individual in their growth pattern, and there is no set way in which they all develop. A minority start to develop sexually at a young age, although this does not continue to full development. In the majority, puberty is delayed until the late teens. Men have a small penis, and undescended testes are common. Pubic and facial hair may be scanty, and voice change may not occur. In women, breast development is often small, and menstruation, if it occurs, may be irregular and/or scanty.

Teenagers with PWS who have not received growth hormone do not experience the growth spurt which usually occurs in the early teens and hence tend to be a few inches shorter than average: men average about 5'2" (155cm) and women average about 4'10" (145cm). However, those on growth hormone very often reach at least the same height as their parents. Sex hormones (both male and female) can help with sexual development, but this will depend to some extent on the individual's existing hormone levels. Behavioural and eating problems may become more challenging during the teens and early twenties.

## Life as an adult

As adults, people with PWS have varying abilities in attaining independence, although all will need some form of support or monitoring to help with controlling their food intake, and thus their weight. Despite the fact that many individuals have the intellectual and physical ability to work, they are usually ill-equipped on an emotional and social level to deal with the stresses and demands of the ordinary workplace. However, they can make a positive contribution to society in many ways and may be involved in voluntary work, craft work, or have a part-time job. Many people live with their families, but an increasing number are living in residential homes, or being supported to live in the community.

In the past, life expectancy was short because of health problems associated with massive obesity, but nowadays life expectancy is increasing because of better dietary management and better understanding of the problems associated with PWS. The oldest known person with PWS in the UK was a woman who died at the age of 74.

## The Treatment of PWS

Apart from various hormone treatments and some surgical intervention (eg, to bring down undescended testes), there is currently no "cure" for PWS. There have been many advances in the fields of genetics, and researchers are not far off identifying exactly which genes are involved in PWS. No drug so far has proved to be of lasting help with regard to suppressing appetite.

Generally speaking however, many of the adverse effects of the syndrome can be lessened by good dietary management, exercise programmes, good general health care, and by good general management of behaviour and education. The help of a dietitian, paediatrician, physiotherapist, occupational therapist, educational psychologist, and (if necessary) speech therapist should be sought as soon as a diagnosis is made.

*Remember - everyone with PWS is an individual, and different characteristics and symptoms can vary considerably. They can also vary over time.*

## Further information

Information for GPs <https://www.pwsa.co.uk/assets/files/Information-for-health-professionals/GPs.pdf>

Information for Emergency Departments and other hospital staff  
<https://www.pwsa.co.uk/home/emergency-medical-information>

Multidisciplinary paediatric health oversight [https://www.pwsa.co.uk/PWS\\_Multi-Disciplinary\\_FINAL\\_web\\_version.pdf](https://www.pwsa.co.uk/PWS_Multi-Disciplinary_FINAL_web_version.pdf)

Healthcare information for parents and carers <https://www.pwsa.co.uk/information-for-families/health>