

Physical Characteristics of Prader-Willi Syndrome

An individual with Prader-Willi syndrome may have many of the following physical characteristics or only a select few.

There are some distinctive facial features associated with Prader-Willi syndrome that are noticeable in babies soon after birth. These include almond-shaped eyes, narrow bridge of nose, narrowing of forehead at the temples and thin upper lip and upturned mouth.

Babies born with Prader-Willi syndrome often have a lower than average birth weight.

Due to weak muscles babies may appear floppy shortly after birth, this is referred to as hypotonia. A baby suffering from hypotonia may also have poor reflexes, a weak cry, and be unable to suck properly which causes them to be underweight in their first year. Hypotonia generally improves with age, with most adults with Prader-Willi syndrome only showing decreased muscle tone and bulk.

Hypoplasia refers to the incomplete development of the sexual reproductive systems (ovaries and testes). In baby boys, the penis may be abnormally small and one or both testicles may be undescended (remain in the abdomen). Genitals in girls may also be underdeveloped.

Around one in three individuals with Prader-Willi syndrome will have hypopigmentation which presents as unusually fair eyes, skin and hair relative to others in the individual's family. Hypopigmentation is more common with the [deletion genotype](#) because individuals lose one copy of the 'pigmentation gene' that is located on chromosome 15.

Strabismus (crossed eyes) occurs in around 60-70% of individuals with Prader-Willi syndrome.

Due to the decreased levels of human growth hormone (HGH), children with Prader-Willi syndrome may be shorter than peers of the same age. Artificial human growth hormones can be prescribed in order to replace the lack of hormone and encourage growth.

Obesity is common in Prader-Willi syndrome, as [hyperphagia \(excessive eating\)](#) is a well-known symptom; along with poor muscle tone which makes physical activity more difficult resulting in less calories burnt.