Frequently Asked Questions

1. **If no members of my family are affected by PWS, can I still have a child with the condition?**
   Yes. The majority of cases of PWS occur in families where there has been no history of the condition previously reported. The genetic change involved occurs randomly and the cause of it unknown.

2. **Since PWS is a genetic condition, is it possible to have more than one child with the condition?**
   It is extremely unlikely for this to happen as the vast majority of cases are not inherited but rather occur by chance.

3. **What causes the preoccupation with food and the compulsion to eat?**
   They are caused by a problem in a part of the brain (hypothalamus) which normally controls feelings of hunger and fullness. Hence, people with PWS do not feel full after a meal and have an urge to eat constantly.

4. **Will diet plans that are commercially available be able to reverse obesity?**
   No. Unfortunately no diet plans has worked consistently for people with PWS. Hence, it is important that eating and exercise habits be regularly monitored and the environment carefully considered to prevent easy access to food.

5. **Do people with PWS have a shortened lifespan?**
   Yes. The primary cause of death is related to obesity, hence by taking steps to prevent it, a normal lifespan can be achieved.

6. **How do the mental and behavioural problems affect the life of a person with PWS?**
   Early intervention to address these issues may help in enabling the individual to achieve many things in life such as completing school and being employed. However, constant support from family and friends is crucial to achieve these goals. Psychotropic medications may also be helpful in managing difficult and unpleasant behaviours.

References

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MALAYSIAN RARE DISORDERS SOCIETY

Rare Disorders Series:

Prader-Willi Syndrome

What is Prader-Willi Syndrome (PWS)?

PWS is a genetic disorder that results in a number of physical, mental and behavioural problems.

A primary characteristic of PWS is the patient failing to thrive in the first two years of life, followed by compulsion to eat, which may lead to extreme obesity in childhood.

PWS is caused by the lack of, or defect on critical genes, located on a specific section of chromosome 15 that is inherited from the father.

All genes (with the exception of male sex chromosomes) come in pairs; one coming from the father and another from the mother. Genes contain 'recipes' to make proteins.

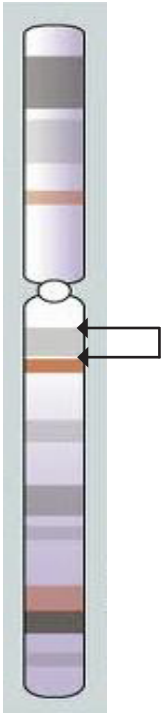
Some genes are only active when they are paternally inherited. This is the case of the genes associated with PWS. Hence, when there is a change in these genes, the required proteins cannot be made, which results in PWS.

More than 95% of all PWS cases are not inherited. These happen by chance and are not due to anything that either parent did or did not do before or during pregnancy.

Chances of PWS recurrence in a family are very low. However, it is advised that couples who have a child with PWS seek genetic counselling to ascertain the risk for future pregnancies.

PWS is considered a spectrum disorder. This means that symptoms and severity will vary from patient to patient.

Patients are advised to consult a geneticist to obtain further information on disease progression.



The region on chromosome 15 where the genes which cause PWS are located. A change in certain genes at this region of a paternal chromosome will result in PWS.

Signs and symptoms

- Poor muscle tone at infancy (hypotonia)
- Poor weight gain in first year of life due to poor sucking
- Delay in achieving developmental milestones
- Rapid weight gain between ages one to six
- Behavioural problems such as temper tantrums and violent outbursts
- Obsession with food and foraging habit
- Under developed sexual organs
- Delayed puberty

Other ways PWS may affect the patient

- Short stature
- Infertility
- Severe obesity
- Difficulty in sleeping and breathing
- Delayed motor and speech skills

Testing

Genetic counselling by qualified personnel is advised if there is a family history of the condition and before any genetic testing is done.

Special genetic tests to identify PWS can be performed on individuals who exhibit symptoms.

Due to the complexities involved in the genetics of PWS, more than one test may be required before a confirmed diagnosis can be made.

Testing involves the drawing of blood samples from the patient and his/her parents.

Treatment

There is currently no known cure for PWS.

Treatments and therapies are given to manage symptoms and to maximise the skills and potential of individuals with PWS.

A balanced, low calorie diet as well as regular exercise is recommended to manage weight related issues.

Growth hormone injections may be required to increase growth and influence metabolism

Multi-disciplinary management is required to treat obesity, diabetes mellitus and obstructive sleep apnoea syndrome.

Early intervention programs which incorporate speech, occupational and developmental therapies are important to maximise the child's potential and independence.