Saint Mary's Hospital
Manchester Centre for Genomic Medicine

Information for Patients

Prader-Willi Syndrome (PWS)

What is Prader-Willi Syndrome?

Prader-Willi Syndrome (PWS) is a genetic condition that was first described in 1956 by three Swiss doctors – Dr's Prader, Labhart and Willi because they noticed that there were similarities in a few children they had seen with developmental delay and feeding problems.

Children with PWS are born with floppy muscles and have severe feeding difficulties and poor weight gain in the first year of life. However, by the time they are three years old, they develop a huge appetite. This can lead to rapid weight gain and obesity, even in early childhood. Children with PWS may also have learning difficulties and behavioural problems.

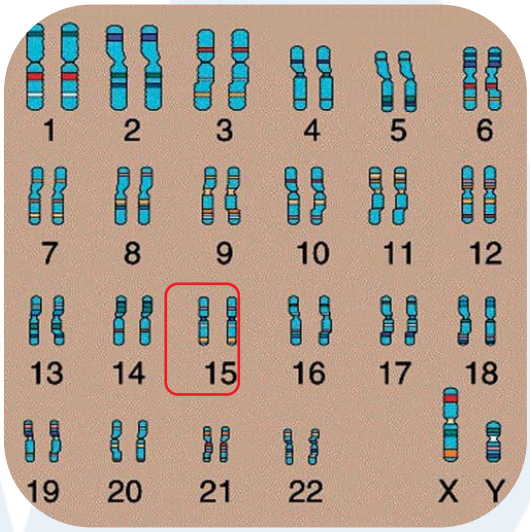
Who gets PWS?

Approximately 1 in every 15,000 births is affected by Prader-Willi Syndrome. Every year in the UK, there are about 35 children born with PWS. It occurs in both boys and girls and people of any ethnic background can be affected by it.

What causes PWS?

Prader-Willi Syndrome is due to missing or inactive genes on one of an individual's two chromosome 15s - the one normally contributed by the father.

The chromosomes are the packages of genes found in nearly every cell of the body. We have 46 chromosomes (see diagram). The chromosomes come in pairs because we inherit one set from each parent. The genes that cause Prader-Willi Syndrome are on chromosome 15.



Is PWS inherited?

Most cases of PWS are not inherited - they are caused by a genetic error that occurs at or near the time of conception for unknown reasons. In a very small percentage of cases (1% or less), a genetic change that does not affect the parent is passed onto the child and in these families more than one child may be affected.

All families in whom there is a person affected with Prader-Willi Syndrome should have the chance to talk to a geneticist or genetic counsellor.

Three different causes of PWS

Prader-Willi Syndrome occurs when a baby has failed to inherit some active genes from a specific section of his/her father's chromosome 15. There are three different ways that this can happen:

1. A small section of chromosome 15 is missing - a paternal deletion.

This is the most common form of PWS. At the time of conception, something happens that causes a small part of the chromosome 15 which is inherited from the child's father to disappear. This is called a chromosome deletion. It is unlikely to happen again in another pregnancy.

2. The baby has two copies of his/her mother's chromosome 15 - maternal uniparental disomy (UPD).

This form of PWS occurs in approximately one in four cases. It happens when a baby is born with two copies of his/her mother's chromosome 15 and no copy of his/her father's chromosome 15. The effect is the same as that of a paternal deletion: the child is missing some genes from its father.

3. The baby's PWS genes are 'switched off' - an 'imprinting' mutation.

Rarely (in about one in twenty cases), the PWS genes on the father's chromosome are present, but they don't work properly and they seem to be 'switched off'. This is caused by a change (a mutation) in the gene on chromosome 15 that turns the PWS genes on and off. The process of turning these genes on and off is called imprinting. This rare type of mutation can be inherited or can start for the first time in an affected child.

What kind of problems do people with PWS experience?

- **Growth**

Babies with PWS are often slow to gain weight initially and they may sometimes need tube feeding. Children with PWS benefit from treatment with growth hormones. The need for growth hormone therapy should be assessed by a paediatric endocrinologist.

- **Feeding and speech problems**

Weak and floppy muscles may cause feeding problems in young babies with PWS. Speech development is often delayed. The need for speech therapy should be assessed in infancy. In rare cases, speech is severely affected.

- **Appetite and weight management**

Children with Prader-Willi Syndrome have a huge appetite. They can become obsessed by food. The combination of eating too much and lack of physical activity can lead to rapid weight gain and obesity. Parents may find it difficult to prevent these children from eating too much. Weight control often requires strict restrictions on the child's access to food. This may mean locking the kitchen and food storage areas.

- **Physical skills**

Children with PWS usually learn to sit, walk and crawl later than other children. They can continue to have problems with strength, co-ordination and balance. Physical and occupational therapies help children to develop these skills.

- **Behaviour**

Infants and young children with PWS are typically happy and do not have serious behaviour problems. Older children and adults, however, do have behaviour problems. They find it particularly difficult to cope with changes in their daily routines.

Behavioural symptoms usually start at about the same time as over-eating problems. Daily routines and a firm and structured environment seem to work best for behaviour management.

- **Education and learning**

Children with PWS usually have learning problems. Like all children, they have strengths and weaknesses. They usually need special help at school; either within a mainstream school or in a special needs school.

- **Sexual development**

Sex hormone levels (testosterone and oestrogen) are usually low in Prader-Willi Syndrome. Both sexes have a good response to treatment for hormone deficiencies, although side-effects have been reported. Puberty usually starts late. A few women with Prader-Willi Syndrome have had children, but no men with PWS have fathered children to date.

For more information

If you need more advice about any aspect of Prader-Willi Syndrome, you are welcome to contact:

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Saint Mary's Hospital
Oxford Road
Manchester
M13 9WL

Telephone: (0161) 276 6506 (Reception)
Facsimile: (0161) 276 6145

The department is staffed Monday–Friday, 9.00 am to 5.00 pm.

Website: www.mangen.co.uk

If you would like further support and advice you can contact:

Prader-Willi Syndrome Association (UK)
PWSA UK, Suite 4.4
Litchurch Plaza
Litchurch Lane
Derby
DE24 8AA

Telephone: 01332 365 676
Facsimile: 01332 360 401
E-mail: admin@pwsa.co.uk
Website: www.pwsa.cp.uk

Please let us know if you would like this leaflet in another format (e.g. large print, Braille, audio).

We would like to acknowledge our Clinical Genetics colleagues at Guy's and St Thomas' Hospital NHS Trust who originally designed and wrote this leaflet.

Seen in clinic by (doctor): _____

And (Genetic Counsellor): _____

Telephone number: _____

Family reference number: _____