

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

This form of PWS occurs in about 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 a paternal deletion: the child is missing some genes from its father.

- **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?**

### **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.

### **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.

### **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.

### **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.

### **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.

### **Growth**

Babies with PWS are often slow to gain weight initially and they may sometimes need tube feeding. Children with PWS may need treatment with growth hormone. The need for growth hormone therapy should be assessed in both children and adults.