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## Williams syndrome

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### Summary

- Williams syndrome is a rare genetic disorder that is caused by the deletion of genetic material on chromosome 7.
  - Typical characteristics include distinctive facial features, mild intellectual disability and an overly sociable personality.
  - Williams syndrome may be undiagnosed, which means that many people with the disorder fail to get the support and treatment they need until later in life.
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Williams syndrome is a rare genetic condition. It is characterised by distinctive physical features and behaviours, including a distinctive facial appearance, mild intellectual disability and an overly sociable personality.

Williams syndrome is caused by a missing segment (a 'deletion') of genetic material on chromosome 7. These deletions occur randomly. About one in every 20,000 babies is born with Williams syndrome. Males and females are equally affected, and the condition is found across all races and countries. There is currently no cure.

Treatment for Williams syndrome aims to support the person and manage their symptoms. The outlook depends on the degree of severity. In some cases, a person with Williams syndrome can complete school, work and look after themselves. However, others may need lifelong care.

### Symptoms of Williams syndrome

Most people with Williams syndrome share a wide range of physical, social and cognitive (thinking and acquiring knowledge) traits. These characteristic features may occur to a greater or lesser degree.

Some common physical traits of Williams syndrome include:

- distinctive facial features – including a long upper lip, small chin, generous lips, chubby face, upturned nose and flattened nose bridge. Some people with Williams syndrome may have a squint
- weight and growth problems – a newborn baby with William syndrome may have a low birth weight and gain weight slowly. Adults with William syndrome are usually shorter than average
- feeding difficulties – babies may have problems with feeding, including swallowing difficulties and excessively slow eating
- hyperacusis – extreme sensitivity to sound, which may include being startled by ordinary noise levels
- dental problems – including smaller than normal teeth, oddly shaped teeth, widely spaced teeth and a misaligned bite
- cardiovascular problems – common heart problems include narrowed aorta (aortic stenosis) and narrowed pulmonary artery (pulmonary stenosis)
- hypercalcaemia – higher than normal levels of calcium circulating in the blood
- colic – it is thought that the colic or irritability frequently experienced by babies with Williams syndrome may be caused by hypercalcaemia
- hernias – groin and umbilical (belly button) hernias
- kidney problems – including variations in shape and function
- muscle and joint problems – including poor muscle tone, weak muscles, overly loose joints, muscle contractures and poor physical coordination.

Intellectual and personality traits may include:

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- intellectual disabilities – such as developmental delays, below average IQ, learning difficulties, poor spatial skills and attention deficit hyperactivity disorder (ADHD)
- overly sociable personality – typically, the person with Williams syndrome is inappropriately outgoing, with no fear of strangers or social interaction.

### **What causes Williams syndrome?**

Williams syndrome is caused by a partial deletion of up to 28 genes on chromosome 7. This means that a section of genetic material on chromosome 7 is missing. It is believed that some of these genes are involved in the production of elastin.

Elastin is a protein that provides elasticity and suppleness to various structures, including skin and blood vessel walls. This lack of elastin is most likely the cause of some of the distinctive physical features of Williams syndrome.

Most cases of Williams syndrome are caused by random events that occur during the formation of the egg or the sperm in an unaffected individual.

Williams syndrome can also, rarely, be inherited from a parent who has the condition. In the rare event the condition is inherited, it is inherited in an autosomal dominant manner. This means that just one copy of the altered chromosome 7 is enough to cause the condition. Due to this inheritance pattern, an individual with Williams syndrome has a 50 per cent chance of passing the condition onto any future children.

Williams syndrome is not linked to anything the parents did or didn't do during pregnancy.

### **Diagnosis of Williams syndrome**

Williams syndrome often goes undiagnosed. This means many people fail to get the support and treatment they need until later in life.

Diagnostic tests include:

- medical history
- physical examination to check for typical features
- blood test to check for elevated calcium levels (although high calcium levels are not always involved)
- chromosome microarray testing – a chromosome test that detects Williams syndrome and many other small chromosome changes
- fluorescent in situ hybridisation (FISH) chromosome test to check for the deletion of the elastin gene on chromosome 7. FISH testing has mostly been replaced by chromosome microarray.

### **Treatment for Williams syndrome**

There is currently no cure for Williams syndrome. Treatment aims to support the person and manage associated symptoms.

There may be many physical, developmental, intellectual and social problems present in Williams syndrome, which may require a team of health professionals.

Treatment options can include:

- speech therapy
- social 'training' – for example, how to greet people appropriately instead of automatically kissing and hugging
- regular medical monitoring of heart and blood vessel issues and any other physical problems that could present complications
- surgery to correct heart and blood vessel issues, if necessary
- dental and orthodontic treatment
- physical therapy from a physiotherapist
- occupational therapy.

### **Genetic counselling and Williams syndrome**

If you or a family member have been diagnosed with a Williams syndrome, or if it runs in your family, it can be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand Williams syndrome and what causes it, how it is inherited, and what a diagnosis means for you or your child's health and development.

Genetic counsellors are trained provide information and support that is sensitive to your family circumstances, culture and beliefs.

If Williams syndrome runs in your family, a genetic counsellor can explain what genetic testing options are available to you and other family members. You may choose to visit a genetic counsellor if you are planning a family, to find out your risk of passing the condition on to your child, or to arrange for prenatal tests.

**The Genetic Support Network of Victoria (GSNV)** is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by Williams syndrome.

### Where to get help

- Your **GP (doctor)**
- **Victorian Clinical Genetics Services (VCGS)**, Royal Children's Hospital Tel. **1300 118 247**
- **Williams Syndrome Association of SA** Inc Tel. **(08) 8258 3867** or **(08) 8285 3776**
- **Better Start for Children with a Disability** Tel. **1800 242 636**
- **Genetic Support Network of Victoria (GSNV)** Tel. **(03) 8341 6315**

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