Williams syndrome

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.

Williams syndrome is a rare genetic disorder. Williams syndrome is characterised by distinctive physical features and behaviours, including a distinctive facial appearance, mild intellectual disability and an overly sociable personality.

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

Treatment 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 include:

- distinctive facial features – include a long upper lip, small chin, generous lips, chubby face, upturned nose and flattened nose bridge. The overall picture is usually quite recognisable to the experienced eye. Some may have a squint
- weight and growth problems – the newborn may have a low birth weight and gain weight slowly. Adults are usually shorter than average
- feeding difficulties – the baby 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 – include smaller than normal teeth, oddly shaped teeth, widely spaced teeth and a misaligned bite
- cardiovascular defects – 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 – include groin and umbilical (belly button) hernias
- kidney problems – include variations in shape and function
- muscle and joint problems – include poor muscle tone, weak muscles, overly loose joints, muscle contractures and poor physical coordination.
Intellectual and personality traits may include:

- 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.

**Absent genetic material with Williams syndrome**

In Williams syndrome, a section of genetic material on chromosome 7 is deleted, due to a random error when either the sperm or the egg was forming. The absent material includes the gene that controls 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 distinctive physical features of Williams syndrome. Other genes are also deleted, which is likely to cause other features of Williams syndrome.

Williams syndrome is inherited in an autosomal dominant manner, although most people with Williams syndrome do not reproduce. In most people with Williams syndrome, the condition occurs as a result of a new genetic change occurring at the time of conception. The 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 relatively new type of 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 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 defects and any other physical problems that could present complications
- surgery to correct heart and blood vessel defects, if necessary
- dental and orthodontic treatment
- physical therapy from a physiotherapist
- occupational therapy.

**Where to get help**

- Your **GP (doctor)**
- Victorian Clinical Genetics Services (VCGS), Royal Children’s Hospital Tel. (03) 8341 6201

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