For more information

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

CLINICAL GENETICS DEPARTMENTS:

Northern Scotland (main base Aberdeen)
Tel: 01224 552120 / Fax: 01224 559390
(Aberdeenshire, Moray, Highland, Western & Northern Isles)

Tayside (main base Dundee)
Tel: 01382 632035 / Fax: 01382 496382
(Perth & Kinross, Angus, North East Fife)

South East Scotland (main base Edinburgh)
Tel:0131 537 1116 / Fax: 0131 537 1153
(Borders, Lothian, South West Fife)

West of Scotland (main base Glasgow)
Tel: 0141 354 9200 / Fax: 0141 232 7986
(Glasgow, Argyll & Bute, Ayrshire, Dumfries & Galloway, Stirling, Lanarkshire, Falkirk)

If you would like further information about Williams Syndrome, you are welcome to contact:

Williams Syndrome Foundation
161 High Street
Tonbridge
Kent TN9 1BX
tel: 01732 365152 fax: 01732 360178
web: www.williams-syndrome.org.uk

Seen in clinic by .....

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Supported by ScotGEN

Williams Syndrome

CLINICAL GENETICS CLINICAL GENETICS

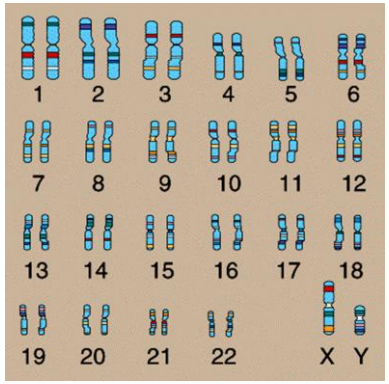
Information Leaflet

What is Williams syndrome?

Williams Syndrome is a rare genetic condition affecting about 1 in 20,000 people. It affects males and females equally and is caused by an alteration in one of the chromosomes.

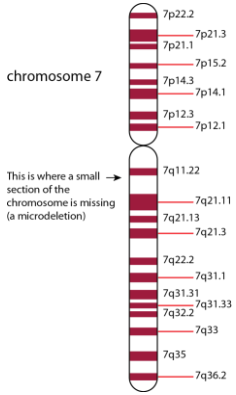
What are chromosomes?

The chromosomes are the packages of genes found in nearly every cell of the body. We have 46 chromosomes. The chromosomes come in pairs because we inherit one set from each parent. They are numbered according to their size.



How is Williams syndrome diagnosed?

By examining specially prepared chromosomes under a microscope, the tiny alteration can be detected. A part of one copy of chromosome 7 is missing in individuals with this condition. This is called a microdeletion.



What causes Williams syndrome?

An individual can be the first person in the family to have the chromosome alteration (sporadic). The microdeletion can be caused during formation of the egg or the sperm. We do not know why this happens.

An affected person has a 50% (1 in 2) chance of passing on the deleted chromosome 7 to their own children. In other words, they have a 50% or 1 in 2 chance of having a child with Williams Syndrome. This chance applies every time they have a child.

What are the features of Williams syndrome?

Physical features

People with Williams Syndrome often have an open mouth with full lips, a button-shaped nose with a flat bridge and their cheeks appear full. The iris or coloured part of the eye sometimes has a star-like pattern to it. The nails can be small.

Growth

Babies with Williams Syndrome can sometimes have a low birth weight. They can also be slow to gain weight in infancy and childhood and are usually of below-average height.

Heart

Some children with Williams Syndrome are born with a heart defect. Most commonly, there is a narrowing of the aorta. This is the main artery supplying blood to the body from the heart. Usually it is mild, but occasionally can be severe enough to require specialist treatment.

Behaviour and intelligence

People with Williams Syndrome are often described as friendly and chatty. They have a dislike of loud noises. They also have mild to moderate learning difficulties.

Other Features

Feeding problems and vomiting can occur in infancy and childhood and the children can be poor sleepers. Some people have a squint or poor vision and require glasses. The calcium level in the blood is sometimes raised during the first few years of life.

Can Williams syndrome be treated?

Feeding problems and vomiting can occur in infancy and childhood and the children can be poor sleepers. Some people have a squint or poor vision and require glasses. The calcium level in the blood is sometimes raised during the first few years of life.

Can unaffected family members pass it on?

Unaffected family members have no greater chance of having a child with Williams Syndrome than the general population. Parents with an affected child have a very small chance of having another child with the condition (less than 1%).

Can subsequent pregnancies be tested for Williams syndrome?

The chromosomes of a baby can be looked at during pregnancy using a method called CVS (chorionic villous sampling - taking a sample of the developing placenta) or amniocentesis (sampling the fluid surrounding the pregnancy). These procedures carry a small risk and the pros and cons should first be weighed up.

There are other leaflets that explain these tests in more detail. Ultrasound scans during pregnancy can also be used to look for heart defects in a baby.