

WHAT IS IT?

Williams Syndrome occurs at random.

It is a congenital syndrome due to an error on chromosome 7 (at the elastin gene focus).

There is a typical facial appearance and variable learning difficulties. There may be problems of calcium metabolism within the first two years of life and other problems may occur in the heart, kidneys, eyes, teeth, and with the hearing.

HOW IS A DIAGNOSIS MADE?

Diagnosis is not easy as the effects can vary considerably, but the different clues can be added up to produce a near-certain diagnosis.

The cause of Williams Syndrome is a microdeletion of part of chromosome 7 which includes the elastin gene, and a blood test (called the FISH technique) can establish if the elastin gene is in fact missing.

WHAT ARE THE CLUES?

Facial Features - Williams children share a certain facial similarity, referred to as 'elfin' features. They include

- a wide mouth, with large, slack bottom lip
- upturned nose with flattened bridge
- slightly 'bulgy' cheeks
- irregular teeth widely spaced
- sometimes a squint.

Early Problems - may include any or all of the following symptoms:

- low birth-weight, often after being 'late for dates'
- slow weight gain (sometimes weight loss)
- below average growth
- very slow feeding
- restless sleeping
- irritability
- possible hernia
- possible excessive vomiting leading to dehydration and constipation
- possible raised calcium-level.

Heart Problems - all WS individuals appear to have a slight narrowing of the aorta above the valve. In many cases this is insignificant, but occasionally it can lead to more serious heart defects.

Later Problems may include

- hyperactivity in early years
- extreme uninhibited behaviour
- excessive talking in an inappropriate and 'adult' manner
- over-friendliness with strangers
- compulsion to talk to adults, while being unable to make friends with peers
- high verbal ability leading to artificial expectations of matching mental ability
- obsessional interest in certain things, e.g. cars, trains, vacuum cleaners, wheels, etc.
- fears of height, open stairs, uneven surfaces
- very short concentration span and easily distracted, adding to learning difficulties
- emotional immaturity exhibited as over-reaction to events, and exaggerated displays of fear, excitement, sadness, happiness, etc.

Hypersensitivity to Noise - this is the clue most common to WS children: about 90% show great distress on hearing sudden loud noises, such as guns firing, balloons bursting, Christmas crackers, fireworks, etc.

TREATMENT

There is no 'cure' for Williams Syndrome. When hypercalcaemia is present, a low calcium diet can stabilise the situation - but any damage done before birth cannot be reversed. Early diagnosis will lead to a better understanding of the problems which may arise, resulting in a happier life for the child, and relief and support for the parents. Continuing education can help to realise the child's full potential.

WHAT IS BEING DONE?

The Foundation was formed as a Registered Charity in 1980, with the aims of promoting research, and providing help and support for families with affected children. The Foundation incorporates Williams Syndrome and Infantile Hypercalcaemia. The incidence of Williams Syndrome is approximately 1 in 25,000. The Foundation hears of over 75 cases a year - and this figure is rising as awareness grows

By 2004, over 1,300 cases were known in the U.K., and similar organisations are established in the USA, New Zealand, Canada, Australia and most countries in Europe. The foundation is currently funding research projects costing £300,000.

HELP AND SUPPORT FOR FAMILIES

Research helps future generations. How can existing children benefit now?

It's not too late to help.

The Foundation finds many ways to help WS children and their families. There are Regional Contacts around the country - all parents of WS children - who keep in touch with local families, visit new contacts, arrange meetings and organise fund-raising events



The Foundation organises annual national meetings, as well as local gatherings for families.

The Foundation pays for Group Holiday weeks for unaccompanied adults, and also families (self-catering) every year. In 2004 these cost over £25,000, and expenditure increases every year. This is one of the best ways in which the Foundation can help families.

The Foundation acts as an information and advisory service and keeps parents in touch with the Foundation and with each other through regular newsletters and magazines. It also keeps a register of all known WS cases, and provides access to professionals to promote research.

HOW CAN YOU HELP?

The WS Foundation is run for parents by parents. There are no paid fund-raisers, and funds go directly to research and welfare, apart from minimal administrative expenses. The Foundation is supported by a panel of eminent medical / professional advisers. The Foundation is still relatively unknown, but very cost-effective, so any contribution to our aims would be most gratefully received. Gift Aid, Direct Debit forms and further information can be obtained from the Registered Office.