Williams Syndrome – Some Answers!

If you're looking for information on Williams Syndrome, the chances are you know someone who might have this complex condition. Even a definitive diagnosis raises so many different questions and concerns. We've put together this leaflet to provide answers to some of the questions that are often highest on the list, beginning with:

What is Williams Syndrome?

What does that mean?

What are the implications for the life and health of the person affected?

How can we help them?

How does this affect our family?

What can we expect in the future?

Please do explore our website (http://WilliamsSyndromeAdult.com) and the links it contains.
Sign up for our newsletter by entering your name and email address – it's great to keep in touch.
When it comes to Williams Syndrome or living with/caring for someone who has Williams Syndrome, what would you like to know?
Here's hoping you keep in touch and, when you're ready, add your experiences to the resources available for all families and carers providing support for people with Williams Syndrome.

What is Williams Syndrome?

Williams Syndrome is a genetic condition that affects between one in every 8,000 or 25,000 births, depending on which part of the world you live in or which website you get your information from.

http://opac.yale.edu/news/article.aspx?id=7015

Either way, it's pretty rare and most people won't personally know someone who has Williams Syndrome or WS. The fact that you're searching for this information suggests that you probably do know such a person!

The syndrome was first described in 1961 by a Dr Williams based on a number of characteristics in people affected; basically four children all with aortic stenosis, learning difficulties and particular facial features.

The fact that this is a "syndrome" means that there are several features and that not all affected people show all of the symptoms or to the same degree. I guess this also emphasises that people with Williams Syndrome are also unique individuals who have some things in common.

Some of the things you might notice are that children often show delayed development, feeding problems and are slow to develop speech.

As they get a little older you might notice that they have a fascination for music, listening to it, singing along or playing instruments, hearing music can affect them deeply. Their speech also develops fluently after a slow start and children with WS are often described as over-friendly and are very comfortable with the presence of adults, even strangers.

Children may show signs of hyperactivity, they – and WS adults – are often easily distracted, and may be spooked by loud noises such as sirens or thunder. You'll often notice that they are quite happy talking to themselves!

Most people with WS have some degree of learning difficulty, particularly as applied to math or numbers, and whether as children or adults, they often have a characteristic "look" about them. They are often shorter in height than any siblings and their faces show common features including slightly prominent eyes, a smaller upturned nose, a longer than average gap between nose and upper lip and small or widely spaced teeth are often visible through an open mouth.

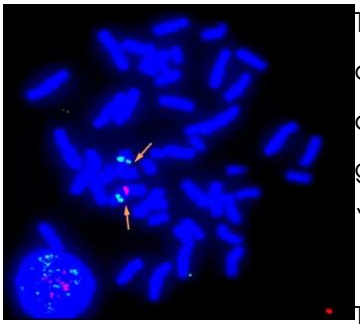
In the mid 1990s researchers were able to confirm that the root cause of WS was a small deletion on chromosome 7, that means, in people with WS, a piece of genetic material that contains about 25 different genes, is missing from one of their chromosomes.

What Does That Mean?

Williams Syndrome – a genetic condition – a chromosomal deletion?!! What??

All our genes are inherited in our chromosomes and they contain the biological instructions about how to make our bodies. We get one set of chromosomes from our Mom, another from our Dad. That means we have two copies of most genes in each cell of our bodies.

In a person affected by WS, somewhere along the line, possibly in the formation of the egg or the sperm cell or very early after conception, a random event deletes a small piece from one copy of chromosome 7. The size of the missing piece is different in different people but it often contains 20 to 30 genes and the affected person then has only one copy of these genes rather than the normal two copies.



The absence of these genes on chromosome 7 causes the many different effects we see in a WS person. One gene that is central to this deletion and is absent in a majority of people affected by WS is the gene for a protein called elastin. If you thought that sounds a bit like "elastic" you'd be right.

This protein elastin occurs in the types of muscle that line our blood vessels and the fact people have only one copy not two, may contribute to the fact that many people with WS have conditions which involve narrowing of the blood vessels such as aortic stenosis or renal artery stenosis. Don't worry if you don't know what that means just yet, it's something a doctor can and should check for and a great place to start is by making a simple blood pressure measurement as many people with WS have high blood pressure.

Other genes that are deleted in Williams Syndrome seem to be involved with speech and language, motor co-ordination, and other learning functions. Although the details are not fully understood, the genetic deletion affects many different physical and cognitive or emotional systems leading to the learning disabilities, poor visuo-spatial skills, poor co-ordination and amazingly, their fascination with music in various forms.

I can begin to explain more about what this means, how it happens and how a "simple deletion" can have such a wide range of profound effects but that will come later. Look out for my special science report on the website (http://WilliamsSyndromeAdult.com).

What Are The Implications For The Life And Health Of The Person Affected?

This is a big question and an important one but, remember, any individual may not have all of the symptoms and different people will be affected to differing degrees. This is not a rule book!

Also remember, I am not a medic so it's really important that you also get more specialist information but this might just get you started with some good questions of your own.

The big "life" thing – well, different individuals have very different potential. The majority of WS people will live healthy lives once the medical things (see later) have been appropriately managed. There's been less research into long term care or life expectancy as most effort has been invested in the care and health of children. As adults, Williams people are much less likely to live independent lives and throughout their schooling will need some degree of support. They are less likely to be employed though some do work in more sheltered and supportive employment while the majority of WS adults live with their parents, carers or in supported households.

As for health - there are many medical conditions that may affect someone with WS and these can come into play at different times of their life, some more acute in childhood, others developing with age, hence the need for ongoing monitoring.

Very young children can have problems feeding (colic, reflux, vomiting) and they may "fail to thrive". Their development is often delayed in many ways, they are slow to speak and can be slow to learn to walk. Once mobile, they are usually highly active and into everything.

It's often said that children with Williams Syndrome have a particular appearance, often referred to as "elfin faces": slightly raised or puffy eyes, a flat bridge to a small upturned nose, a longer than average gap between nose and upper lip and a small pointed chin.

Teeth, when they erupt, can be unevenly spaced, some may be missing and they can have weak or damaged enamel. Their teeth are usually very visible through that infectious, wide-mouthed, Williams Syndrome smile.

Once they learn to talk, there's no stopping them. They chatter! Their behaviour os often described as over-friendly as they are extremely trusting, even of strangers.

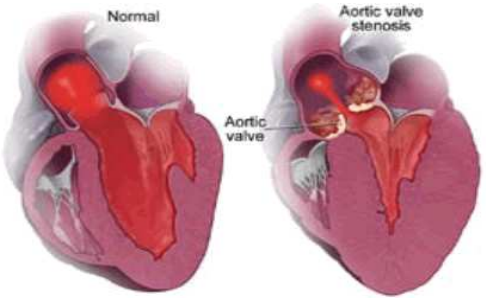
They may be frightened of loud or sudden noises, one sign of hyperacusis. A majority are often fascinated by music of any sort, it can make them very happy or intensely sad. I remember my own sister crying her heart out every time she heard the Christmas Carol "Away in A Manger", while "Lily The Pink" will send her into fits of giggles, every single time!

Children and adults may show poor attention to many things, they really do forget what they went into the room for, can just zone out and stop listening half way through a conversation. Yet it's not an inability to focus. They can be intensely engaged with other activities or objects, certainly music, often babies, baby animals or even mechanical objects – cars, ships, planes – are an intense focus of interest to them.

Many people with WS are very skilled at "reading people". On the one hand they are so friendly and trusting that they are vulnerable to being taken advantage of. Being extremely sensitive to other people's emotions, they can sense the slightest disdain or displeasure and are extremely keen to be liked and loved.

Many WS people also have issues with urination and/or constipation, conditions which again need monitoring and management.

Some affected children also show hypercalcaemia, that is, too much calcium which can lead to deposits of calcium in the kidneys. This can be monitored though it is sometimes good to avoid giving children with WS calcium supplements.



The elastin deficiency may contribute to problems with blood vessels (aortic stenosis or renal artery stenosis) which can be too narrow or to heart murmurs.

These should be actively monitored throughout life.

Learning deficits may also appear in a majority of children. Some can enter mainstream schools with some degree of support but most will need more specialised educational support or schooling. Their verbal skills and fluency develop rapidly. They speak well and fluently, though often their use of complex vocabulary seems slightly odd. They can be very precocious or persistent in their questioning. They will often be able to sing – and remember many dozens or hundreds of songs, picking up tunes and lyrics almost effortlessly.

These evident verbal skills are often starkly in contrast with poorer numeracy skills. Even simple arithmetic can be very difficult and many WS children have great difficulty counting money or telling the time. Basic concepts such as more or less can totally baffle them. This will often persist into adulthood which often limits their potential for independent living but again, skills can be developed with appropriate support.

Many but not all people with WS develop diabetes as they get older and also specific forms of deafness. Again these should be monitored. Adults will often report feeling unspecified joint pains or abdominal pain which may, or may not be associated with diverticulitis which does seem to develop earlier than usual in some WS adults.

It's also important to be aware that many adults with WS are also affected by one or more of a wide range of psychiatric conditions. They often show heightened anxiety or may develop phobias. The key here is to know that these conditions too can be monitored, managed and treated successfully.

How can we help them?

People born with WS will need extra medical, physical and emotional support. The amount of support and exactly what's needed will depend on the needs of each individual person and the ways in which WS expresses itself. You, their families and medical practitioners will need to ensure that their medical needs are monitored and met throughout life. I can't tell you how to do this. You need to use your own intuition and work in partnership with the appropriate medical specialists.

What I can tell you is how I have helped my sister and what I've read about or learned from other people affected by WS.

I've observed an unusual combination of fearlessness in social settings with vulnerability and insecurity. "Am I good enough?", "Do they like me?", "Do you love me?" are questions I often heard either explicitly or implicitly. I guess, particularly as children are growing up they are so often told "Don't do that!", "Stop that!", Why can't you...?!". My sister, although brought up in a loving and caring family was told she was naughty or clumsy far more often than she was praised for doing something well. Many adults with WS also show these scars and are very self critical, particularly when it comes to an appraisal of their numeracy skills.

I've discovered that routines and systems help. Having regular times, set ways of doing things can give people a secure structure to operate in. Doing a regular thing in a regular way can certainly help their learning, from bathing routines to getting dressed or a procedure for leaving the house.

Don't ask too much at first. "Put your shoes on, then get your coat and gloves" can be just too complicated until it becomes part of their routine. How can you make any task into a system?

In general, there's a lot of evidence to show that we learn things better and more effectively when we are doing things that we enjoy. The thing that most Williams people seem to enjoy most is music. I've found that learning to do things in association with music can be a way to help learning, maintain interest and encourage persistence.

Williams people are well known for poor motor skills and poor co-ordination. Even something as simple as catching ball can be difficult and persistent failure can be totally disheartening. Associating ball games with music helped. Keeping things short, knowing when to stop and praising every little positive step can help. Remembering that everything takes a lot, and I mean A LOT longer than you ever thought possible is also essential, so be patient and then some!

There are many specific things that I've learned will help and have used to develop the skills and confidence of my adult sister. I can only guess that they would be even more effective if used with a

younger adult or child who might be more flexible and receptive to new ways of learning and doing things. I'm compiling these into a book right now so keep coming back to see if I've finished!

How does this affect our family?

Sharing your life, at whatever level, with another person will surely change your life. Learning to live with or care for someone with Williams Syndrome will definitely have an effect. If I recall the arrival of my sister, she was small and vulnerable and demanded lots of attention from everyone.
I remember lots of crying and my parents had many sleepless nights.

Once my sister became mobile, crawling and later walking, she was literally, "in to everything". She seemed to have boundless energy, couldn't be left alone for a moment. We shared a bedroom and, when she should have been in bed sleeping she would be "up to no good" spreading talcum powder into the carpet, taking books from shelves, clothes from cupboards, basically exploring her environment in a way that wasn't appreciated by her big sister.

On going to school she washed her hands after going to the toilet unsupervised but left the water running and flooded the place, at least twice before lessons were learned and she gained a constant, adult companion! Be prepared to be constantly attentive for years to come.

There were many times of frustration and probably at least as many times of pure joy. Simple pleasures and delight shared by all around. There's infectious laughter in abundance, hugs and grins given to all. Moments of complete stunned silence as the precocious Williams child asks a totally inappropriate question to an unrelated adult – again! I guess as a child, a sibling, I was embarrassed more easily than I could laugh at these situations sometimes.

You might find yourself frustrated by their inability to remember the simplest things... leaving the room and forgetting why isn't restricted to "senior moments" for a Williams person, it seems to happen all the time. In contrast they can also become obsessive about some things and you wish and you pray that there was some way of distracting them or making them forget.

Most people will embrace their Williams friends but there will also be times of tears, frustration or plain embarrassment. Personally, my experience of living with a Williams Syndrome sister is overwhelmingly positive – but it's not always easy or without painful challenges.

What can we expect in the future?

The answer here is honestly, "It depends". It does depend on the degree to which each individual with Williams Syndrome is affected. It will also be influenced by how much help, encouragement and support they receive.

It has been reported that Williams children are musically gifted and they certainly seem to be both fascinated and deeply moved by music. The giftedness aspect though might also depend on how much this natural interest is actively developed into skills...

Williams children who are actively taught self-help skills early will stand a greater chance of being more independent as adults. It can be painful and frustrating to continually encourage the development of skills in numeracy – practically counting money or calculating how much change to expect from a transaction – but persistence pays off!

Few adults with Williams Syndrome live independently but most live happily. Some will marry but a majority won't. Most will live healthy lives while some will die young from complications arising from medical conditions – a really good reason to establish systems for regular medical monitoring.

At the moment, treatment of genetic conditions like Williams Syndrome isn't a possibility. But we can work together to give all people affected richer and happier life experiences.

What Else Would You Like To Know?

The resources in this leaflet are frequently updated and additional resources are being created – more about the science, more about the associated medical conditions, more about the possible symptoms and much more about practical ways of helping to develop the skills, talents and potential for independent living skills in people who are affected by Williams Syndrome...

What else would you like to know? Please do share your questions on our website:
http://WilliamsSyndromeAdult.com/Your-Questions/

These resources will be shared through the website, discussed on the blog and distributed in a newsletter. Make sure that you're signed-up to receive these updates as they are released. To sign up for the Newsletter for free visit:
http://WilliamsSyndromeAdult.com/

