SOME INHERITED DISEASES IN THE HUMAN BODY: TURNER AND WILLIAMS SYNDROME

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Annotation: This article provides information on some of the inherited diseases that occur in the human body: Turner and Williams syndrome.

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Williams syndrome is a genetic condition present from birth that occurs because a small piece of chromosome 7 does not form properly after conception. Williams syndrome can't be cured, but treatment can help manage the symptoms, especially if started early.

Williams syndrome can cause delays in a child's development and learning. They can have problems with:

- speech, with the first word coming as late as 3 years of age
- motor skills such as walking and toilet training
- doing activities such as drawing or puzzles

However, children with Williams syndrome often have well-developed skills in language and music, as well as outgoing, social personalities.

They might have a distinctive appearance — a broad forehead, a small, upturned nose, a wide mouth with full lips, a small chin and problems with their teeth. They might also have weak muscles. They might also have weak muscles, be shorter than other family members or have dental problems.

Children might show certain behaviours, including:

- anxiety and phobias
- attention deficit hyperactivity disorder (ADHD)

- being very friendly and trusting strangers
- not having inhibitions, difficulty reading social cues
- having lots of tantrums

People with Williams syndrome are likely to develop health problems in later life, including problems with their heart, thyroid gland, diabetes, vision or hearing, or too much calcium in their blood.

Williams syndrome is diagnosed by observing and examining the child and also by using genetic testing. A doctor may notice the distinctive physical features, heart problems and developmental delay, and then use genetic testing to confirm the diagnosis.

Living with Williams syndrome

There is no cure for Williams syndrome and treatment focuses on managing each child's symptoms.

Early intervention is important in order to get the right support. If you have a child with the condition, this will usually include your child seeing a mix of health professionals such as paediatricians, speech pathologists, physiotherapists and occupational therapists.

Williams syndrome and adult life

People with Williams syndrome can live active and fulfilling adult lives. Some people will find paid work, and some will be able to live semi-independently.

Resources and support

Williams Syndrome Australia has a range of information and useful resources.

In Australia, some states have specific support groups for people with Williams syndrome, their families and carers.

The Raising Children Network can help you understand how to use the health and disability service system.

You might be able to get financial support through the NDIS.

Visit our genetic disorders guide to learn more about genes, types of genetic disorders and where to go for help and more information.

Turner syndrome is a genetic disorder that affects approximately 25 to 50 in every 100,000 girls born each year. It is also known as 45,X, monosomy X and Ullrich-Turner syndrome. It does not affect men or boys.

Normally, a female has 2 X chromosomes in each cell. Turner syndrome occurs when all or part of one X chromosome is missing. This is also known as monosomy. It can affect development before and after birth.

Turner syndrome symptoms

Girls and women who have Turner syndrome are affected in different ways. The main features of Turner syndrome are being short stature and having infertility (due to underdeveloped ovaries).

Girls with Turner syndrome might also have some or all of the following problems:

- difficulty feeding in infancy
- problems with hearing or eyesight
- problems with coordination
- puffiness of the hands and feet
- slower sexual development
- problems with the heart, liver or kidneys

Turner syndrome diagnosis

Turner syndrome might be suspected due to symptoms, but the diagnosis needs to be confirmed by genetic testing. It is possible to test for Turner syndrome before a

baby is born if the ultrasound or other prenatal tests show signs of Turner syndrome.

Sometimes a girl can have Turner syndrome and not have the condition diagnosed until childhood or puberty.

Living with Turner syndrome

Someone who has been diagnosed with Turner syndrome will need to have medical checks to make sure they are staying healthy and will need hormones to replace those that normally come from the ovaries.

If a woman with Turner syndrome wants to have children, she will probably need some help with her fertility.

Turner syndrome increases the risk of some other medical conditions such as:

- coeliac disease or gluten intolerance
- bowel diseases
- diabetes
- low thyroid hormone
- osteoporosis
- hearing loss

Doctors may want to check for these conditions.

Someone with Turner syndrome may have some mild learning difficulties, and may require some extra help.

However, most women and girls with Turner syndrome have normal intelligence and a normal lifespan.

Learn more here about the development and quality assurance of healthdirect content.

References

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