Turner’s syndrome

Summary

- Turner’s syndrome is a random genetic disorder that affects women.
- Usually, a woman has two X chromosomes. However, in women with Turner’s syndrome, one of these chromosomes is absent or abnormal.
- With appropriate medical treatment and support, a girl or woman with Turner’s syndrome can lead a normal, healthy and productive life.
- Treatment aims to correct any physical defects and help bring about puberty.

Turner’s syndrome is a random genetic disorder that affects females. The main characteristics include short stature and infertility. Usually, a female has two X chromosomes. However, in females with Turner’s syndrome, one of these chromosomes is missing or abnormal. The missing genes cause the abnormalities and features found in women with Turner’s syndrome. Other names for Turner’s syndrome include monosomy X, 45X and Ullrich-Turner syndrome.

The effects and severity of the condition vary widely, depending on the degree of chromosomal abnormality. Turner’s syndrome affects approximately one in 2,000 female babies born. However, the occurrence of this abnormality before birth may mean it is more common than generally thought. It has been estimated that only one per cent of fetuses with this abnormality survive to term and as many as 10 per cent of miscarriages have this chromosomal abnormality.

Diagnosis of Turner’s syndrome

The condition may be diagnosed at various life stages including:

- before birth (prenatally) – usually if an amniocentesis has been performed or abnormalities are seen during an ultrasound
- at birth – due to certain physical features
- in childhood – when the young girl doesn’t grow at a similar rate to her peers
- during the teenage years – when puberty fails to arrive
- in adulthood – during investigations for infertility.

Turner’s syndrome is diagnosed using a number of tests including:

- amniocentesis and chorionic villus sampling (before birth)
- clinical history
- physical examination
- psychological and educational assessment
- blood tests and chromosome analysis
- genetic tests.

Symptoms of Turner’s syndrome

The most significant features of Turner’s syndrome include:

- short stature – average adult height is 143 cm (4’ 8”)
- infertility – due to underdeveloped ovaries
- congenital heart defects – in about 50 per cent of affected women
- spatial awareness issues – problems with tasks such as maths
• absence of menstruation (amenorrhoea)
• hearing problems.

Less significant features may include:
• sunken, wide chest with broadly spaced nipples
• extra skin (‘webbing’) on the neck
• puffy hands and feet
• inability to straighten the elbow joints
• pigmented moles
• soft upturned nails
• low hairline.

The cause of Turner’s syndrome

Genes are the blueprint for our bodies, governing factors such as growth, development and functioning. Humans have 46 paired chromosomes, with two sex chromosomes that decide gender and 44 chromosomes that dictate other factors. Our estimated 30,000 genes are beaded along these tightly bundled strands.

Usually, a female has two X chromosomes. However, in females with Turner’s syndrome, one of these chromosomes is absent or abnormal. For example, one of the ‘arms’ of the affected chromosome might be missing, or the affected chromosome could have an unusual shape. The missing genes cause the range of anomalies and symptoms associated with this condition. The direct link between the missing genes and the particular problems is not yet well understood.

Complications of Turner’s syndrome

Some of the medical complications that may need to be considered include:

• Congenital heart defect – the various structures of the heart may fail to develop normally in utero. While some of these correct themselves, others need surgery.
• Hearing problems – women with Turner’s syndrome may have some deafness caused by childhood ear infections. They may also develop nerve deafness caused by degeneration in the hearing nerves.
• Middle ear infection (otitis media) – girls with Turner’s syndrome are more vulnerable to ear infections because their ear tubes are narrower than normal.
• High blood pressure (hypertension) – this occurs more commonly in teenage and adult women with Turner’s syndrome. It may be caused by a narrowing (coarctation) of the aorta (a major artery), which can be surgically repaired. However, often a reason for the increased pressure can’t be found.
• Kidneys – an ultrasound may show some structural abnormalities in the kidneys, but these differences don’t usually affect how well the kidneys work.
• Thyroid function and diabetes – there is a higher rate of type II diabetes and thyroid gland disorders in women with Turner’s syndrome.

Premature menopause and Turner’s syndrome

Since the X chromosomes dictate female physical characteristics, missing genes interfere with sexual development. Infertility is caused by the failure of the ovaries to grow properly – they then undergo a premature menopause. There may be some eggs present at birth, but these degenerate soon after. Only around five to 10 per cent of girls with Turner’s syndrome menstruate naturally and the rest need hormone replacement therapy.

Turner’s syndrome is a lifelong condition. However, many treatment options are available to help affected girls and women reach their potential in all aspects of life. Treatment aims to correct any physical defects and help bring about puberty. Options include:

• surgery to correct any heart defects
• growth hormone therapy to increase height
• hormone replacement therapy to trigger menstruation and the development of secondary sexual characteristics such as breasts

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- regular monitoring to check hormone levels
- regular follow-up and management of medical conditions
- treatment for the management of complications such as high blood pressure
- assisted reproduction.

**Where to get help**

- Your doctor
- Turner Syndrome Association of Australia Tel. (07) 3298 6635
- Royal Children’s Hospital, Department of Endocrinology Tel. (03) 9345 5951
- Genetic Support Network Vic Tel. (03) 8341 6315
- Monash Medical Centre Tel. (03) 9594 6666
- Royal Women’s Hospital Tel. (03) 8345 2000

**Things to remember**

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