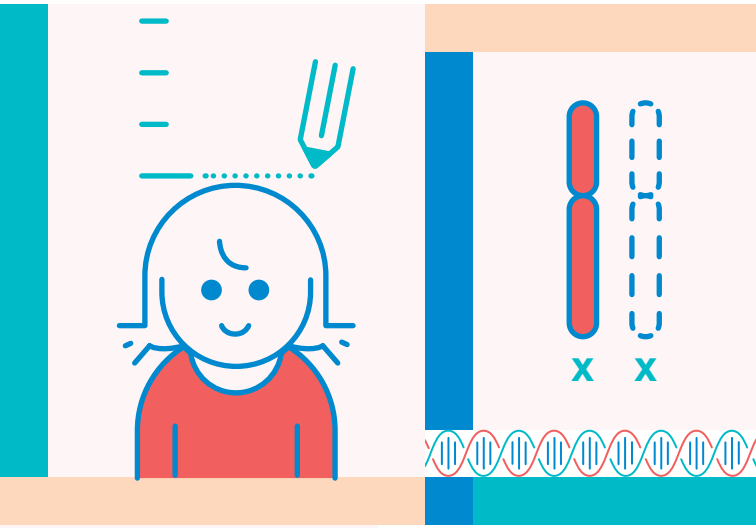
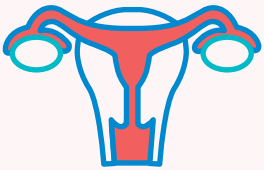
SingHealth DukeNUS
ACADEMIC MEDICAL CENTRE

Genomic
Medicine Centre

Turner Syndrome





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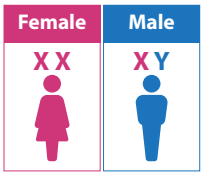
What is Turner syndrome?

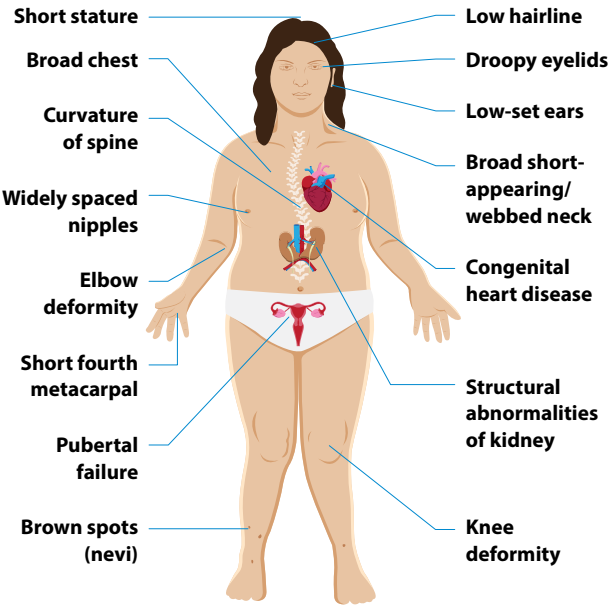
Turner syndrome (TS) is a chromosomal condition that describes girls and women with common features, physical traits and medical conditions that are caused by the complete or partial absence of one X chromosome.

Typically, females have 46 chromosomes which include two X chromosomes, while males have one X and one Y chromosome.



What are the features of TS?

The features of TS can vary widely and it is important to note that not all the features described will be seen in your child. Some girls and women with TS or mosaic TS have minimal or no symptoms.



Growth. Short stature (occurs in 95-100%)

Physical features. Lymphoedema (puffy hands and feet), broad chest and widely spaced nipples, droopy eyelids, low hairline, low-set ears, webbing of the skin of the neck, short fourth toe and short fingers, and arms that turn out slightly at the elbows

Cardiovascular (heart and blood vessels). Congenital heart disease (occurs in 23-50%) with heart defects such as aortic coarctation and bicuspid aortic valve, and hypertension (multifactorial)

Endocrine. Pubertal failure and infertility due to non-functioning ovaries, autoimmune hypothyroidism and osteoporosis

Kidney. Structural abnormalities of the kidney (e.g., horseshoe kidney) (occurs in 24-42%)

Ears. Chronic or recurrent middle ear infections, conductive and/or sensorineural hearing loss

Skin. Pigmented nevi (small brown moles on the skin)

Learning. The majority of individuals with TS are of average intelligence. Some may have impairments in visuospatial abilities, emotional immaturity, or specific learning disorders. Psychological and behavioural problems such as attention-deficit/hyperactivity disorder (ADHD) may also be present.

Susceptibility to certain medical conditions. Includes diabetes mellitus type 1, coeliac disease and inflammatory bowel disease

How common is TS?

It is estimated that TS occurs in approximately **one in 2,000 live female births.**

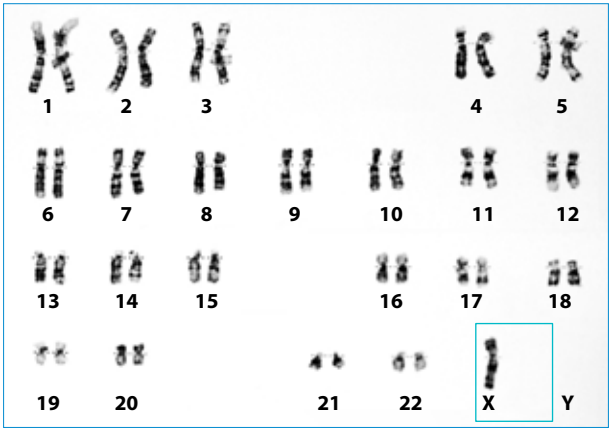
Are there different types of TS?

Girls who are missing one complete X chromosome have what is called monosomy X, often referred to as *'classic TS'.*

Others are missing only a part of the second X chromosome, or have some structural rearrangements of the chromosome.

Girls who have a missing or rearranged chromosome in some but not all cells of the body have a *mosaic karyotype.*

Karyotype showing classic TS



Can TS be cured?

While it is not possible to cure TS, much can be done to make sure your child has the best possible outcome.

How is TS managed?

The multidisciplinary medical team looking after your child will be able to address specific medical concerns and routine TS-related healthcare issues.

Treatment for those with TS should be individualised. Doctors, families and patients should decide on treatment options together.

Ongoing evaluation of the heart, liver, thyroid and hearing is needed from birth and throughout adult life.

Growth hormone therapy

Growth hormone (GH) therapy is recommended for short stature to optimise final height potential.

GH can be started at 4-5 years of age, and involves an injection given daily via the subcutaneous route. GH therapy is generally continued until a child reaches her final height.

Hormone replacement therapy

For girls with pubertal failure, puberty can be initiated with the use of oestrogen replacement therapy. Oestrogen therapy is also important for the prevention of osteoporosis.

Oestrogen can be given in the form of a tablet, skin patch or injection. Progesterone is added after oestrogen to help to produce withdrawal bleeds

(like periods). Once hormone treatment has begun, it is usually continued throughout life.

Below are some related medical issues and how they can be managed:

Medical issue	Treatment
Hypertension	Regular blood pressure monitoring
Learning difficulties	Early intervention therapies
Infertility	While a small number of women with TS will be able to get pregnant naturally, the vast majority will be infertile. Assisted conception using various forms of in-vitro fertilisation (IVF) may be successful.
Lymphoedema	Physiotherapy and support stockings

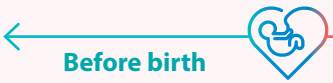
The social and psychological impact of TS cannot be underestimated and should be a priority in the care of any girl or woman with the condition. It is highly beneficial for them to connect with others who share similar experiences and concerns.

What causes TS?

TS occurs when all or part of one of the X chromosomes is absent or altered before or soon after the time of conception. It is not connected to or passed on from either parent, and there is nothing a person can do to increase or decrease the likelihood of their child having TS.

How is TS diagnosed?

The diagnosis of TS is made by looking at the complete set of chromosomes of the individual. This is called a karyotype. A karyotype shows the number and visual appearance of the chromosomes found in the cells of a person.



Before birth

The diagnosis can be made by taking a sample of amniotic fluid, or other foetal tissue, to look at the fetal karyotype.

After birth

The diagnosis is confirmed by taking a sample of blood or other tissue to obtain a karyotype.

How likely will I have another child with TS?

The risk of having another child with TS is low.

TS is a lifelong condition. Should you require financial assistance or emotional support, please approach your doctor for referral to a medical social worker.

Support Group

Club Rainbow Singapore

Club Rainbow Singapore supports and empowers children with chronic illnesses and their families by providing relevant compassionate services in their journey.
Tel: 6377 1789
Email: contact@clubrainbow.org
www.clubrainbow.org

Acknowledgements

Genetics Service, KK Women's and Children's Hospital

Dr Rashida Farhad Vasanwala, Endocrinology Service, KK Women's and Children's Hospital

Information contributed by:

SingHealth DukeNUS ACADEMIC MEDICAL CENTRE

Genomic Medicine Centre

KK Women's and Children's Hospital
6294 4050
www.kkh.com.sg

Singapore General Hospital
6321 4377
www.sgh.com.sg

Changi General Hospital
6850 3333
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