- **Estrogen replacement therapy**
  - Different girls will have different extent of ovarian insufficiency. The medical team will be monitoring the pubertal progression and ovarian function of the affected girl. If no spontaneous puberty is noted, estrogen replacement therapy (oral pills) will be initiated at around 12 years of age to stimulate the development of secondary sexual characteristics (breast development and menstrual periods etc.).
- **Reproductive technologies**
  - Advances in reproductive technologies can help these women to get pregnant. Donor eggs can be fertilized with husband's sperm to form embryos which then can be replaced back into the womb following the use of hormone treatment to prepare the womb lining.
  - If Turner syndrome is diagnosed early in girls who may have residual ovarian function, oocytes can be retrieved following ovarian stimulation and frozen for future use if needed.
  - For those who still maintain ovulatory function or those who have frozen eggs, they may attempt conception using their own eggs, and the option of preimplantation genetic testing can be discussed. This involves the formation of embryos using in vitro fertilization, and testing the chromosomal make-up of the embryos before replacing them back to the womb of the woman. Further details can be provided by the reproductive medicine specialists if needed.
  - In any of the above situations, specific pre-conceptional assessment and counselling is important in view of the higher associated risks to the mother and fetus during pregnancy. The pregnancy should be managed in a multi-disciplinary team specialized in managing high-risk pregnancies.
- **Learning issues**
  - Most girls with Turner syndrome attend mainstream schools. Some of them could have various degree of learning difficulties and may need extra assistance in learning. With early diagnosis, timely assessment and referral to developmental pediatricians for assessment and screening for cognitive issues could facilitate early intervention strategies.

Girls with Turner syndrome need ongoing medical care from a variety of specialists. Nevertheless, the condition is rather well-understood in the medical field and standardized surveillance protocol in taking care this group of girls is available worldwide. With regular follow-ups and appropriate care, most Turner girls can lead happy, healthy, independent lives!

Should I proceed to confirmatory testing? And when?

Options of confirmatory tests

	Timing	Pros :)	Cons :(
Chorionic villus sampling (CVS)	11-14 weeks of pregnancy	Can be done earliest	Procedure-related risk of miscarriage 0.1-0.2% Chance of confined placental mosaicism, result may not represent the fetus
Amniocen-tesis	16-20 weeks of pregnancy	More accurate than CVS	Procedure-related risk of miscarriage 0.1-0.2%
Post-natal blood test	After birth, can use cord blood or baby's blood	Most accurate No risk of miscarriage	Only know about the condition after delivery

Which confirmatory test should I choose?

Your medical team will discuss with you on your options based on the best available information. The decision will be affected by the following:

- Will the confirmatory test affect your immediate management during pregnancy?
- Are there any abnormal findings during the routine anomaly scan that will give further hints on the severity of the conditions?
- The medical issues and potential challenges ahead for girls with Turner Syndrome

Please contact your doctor for further discussion.

Turner Syndrome

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What is Turner syndrome?

Turner syndrome is a genetic condition in females. It affects around 1 in every 2500 girls.

This condition is the result of a chromosomal abnormality but it is usually not inherited. Instead, it occurs as a random event during reproduction.

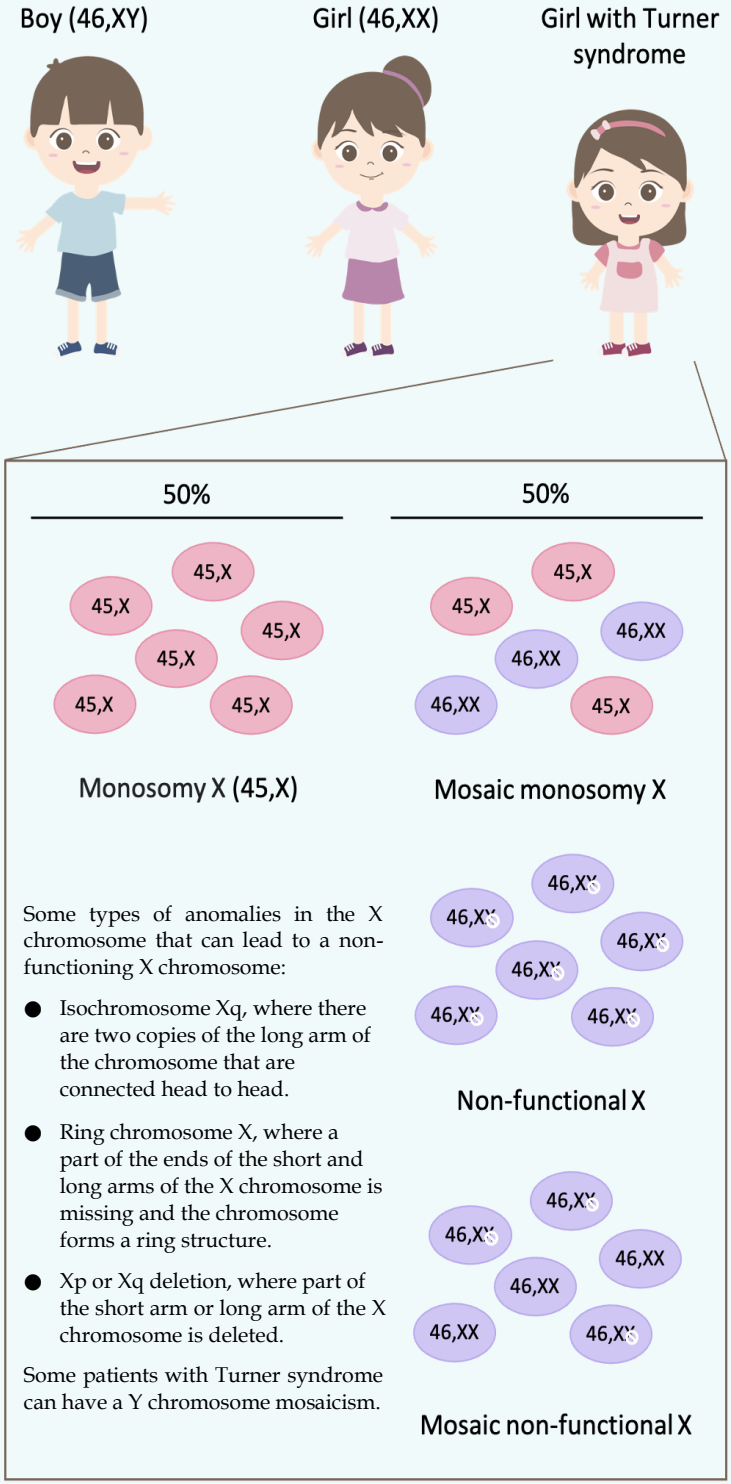
Usually, a person has 46 chromosomes in each cell, divided into 23 pairs, which includes two sex chromosomes. For girls, they typically have two X chromosomes (or known as 46,XX), but girls with Turner syndrome have only one X chromosome (monosomy X or 45,X) or the non-functioning of one X chromosome (for details please refer to the picture on the right). About half of the population with Turner syndrome have monosomy X (45,X). The other 50% of the population has a non-functioning X chromosome and/or mosaicism. Mosaicism refers to an individual having more than one chromosomal make-up in their body. This occurs when there is abnormal chromosome sorting at the time of cell division during early embryonic development.

How is Turner syndrome diagnosed?

Turner syndrome may be diagnosed before birth (antenatally), during infancy or in early childhood. Occasionally, in those with mild features of Turner syndrome, the diagnosis could be delayed until adolescence or early adulthood.

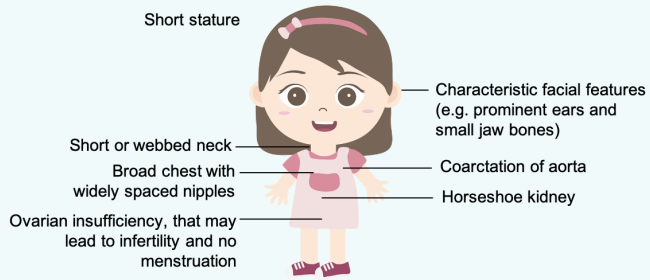
Turner syndrome would be suspected based on certain unique physical features. These can vary widely — some girls with Turner syndrome have more pronounced features, while some have only a few subtle features. Occasionally, it would be detected incidentally from other investigations, e.g. non-invasive prenatal testing (NIPT) for fetal aneuploidy screening.

To confirm the diagnosis of Turner syndrome, your medical team will order a test that looks at the chromosomes — a karyotype.



What is the implication and what issues can happen?

Turner syndrome can cause a variety of medical and developmental issues, including short stature and ovarian insufficiency (which may result in delayed puberty and infertility). Some of them also have heart and kidney defects. As for cognitive function, most girls with Turner syndrome have IQ scores in the normal range but some of them may have specific types of learning issues.



I was told that my baby may have Turner syndrome from NIPT; how accurate is it?

A positive result of monosomy X from NIPT indicates an approximately 40% chance that the fetus could actually have Turner syndrome. This value could be even lower in routine clinical practice due to certain maternal factors such as aging and mosaicism in the mother or placenta, which are not related to the genetic component of the fetus.

Can Turner Syndrome be cured or treated?

Turner syndrome is a chromosomal disorder and hence there is no direct cure for the condition. However, a number of treatment approaches can help:

- **Growth hormone treatment**
  - With growth hormone treatment, the final adult height could be improved — often reaching the low normal range if growth hormone treatment is started early enough. The medical team will be monitoring the growth of the affected child and will initiate timely treatment. Growth hormone treatment for Turner syndrome is approved worldwide, and in Hong Kong, the cost is covered by the Hospital Authority.