



Fact Sheet

From ReproductiveFacts.org



The Patient Education Website of the American Society for Reproductive Medicine

What do I need to know about Turner syndrome and having children?

What is it?

Turner syndrome (TS) is an infrequent genetic disorder in females. About 1 in every 2,000 female babies born is affected.

What causes TS?

TS is not related to the genetic makeup of a mother or father, but is a chance happening and cannot be passed on from either parent. TS occurs when all or part of one of the two X chromosomes in the developing embryo is lost.

How is TS diagnosed?

TS is diagnosed by chromosome analysis, also called karyotype testing. A karyotype is usually performed on blood. An affected baby can be diagnosed before birth by testing cells collected by amniocentesis, or placental cells collected by chorionic villus sampling.

Karyotyping provides both a count of the number of chromosomes (bundle of genetic material) as well as information on appearance of each chromosome (normal versus abnormal). Some females with TS have only a single complete "X" chromosome in their cells instead of the usual 2 complete X chromosomes.

Other women have a single X chromosome in many of the cells of their body and two or more X chromosomes in the remaining cells. Having different cells with different chromosomes within the same person is called mosaicism.

Are there any physical traits of Turner syndrome?

There are physical features that are common in girls/women with TS. Not all girls/women will have all the characteristics. They include:

- Short stature (typically under 5 feet tall)
- Low hairline
- Receding lower jaw
- Short and webbed neck
- Scoliosis
- Puffy hands and feet
- Increased carrying angle of elbows
- Narrow and high arched palate
- Broad chest
- Flat feet

Are there any health problems associated with TS?

The number, type, and severity of health problems associated with TS are variable; some women are minimally affected while others develop serious health concerns. Examples of health concerns associated with TS are:

- Absent/delayed puberty
- Premature ovarian insufficiency ("early menopause")

- Fertility problems due to lack of eggs
- Hearing problems
- Heart and blood vessel defects
- Ear infections
- Kidney problems
- Thyroid problems
- Diabetes
- Potential for learning difficulties

Can TS be cured or treated?

TS is a genetic disorder and cannot be cured. However, treatment strategies are available to reduce the health burden related to the diagnosis. Examples of some treatments available include:

- Growth hormone to increase height
- Estrogen to help develop secondary sexual characteristics, such as breast development, in girls with delayed puberty
- Estrogen to improve bone mass and strength
- Medicine and surgery to correct heart and blood vessel defects

How does TS affect the ability to have children?

Infertility is common in girls and women with TS due to rapid loss of eggs within the ovaries. Spontaneous pregnancies are rare. Pregnancy is possible using donor eggs but with increased risks.

Is it safe for women with TS to become pregnant?

Women with TS are at a particularly high risk for developing heart vessel problems during pregnancy, specifically rupture (bursting) of the aorta (a large vessel leading from the heart). A woman with TS is 100 times more likely to die during pregnancy than all other pregnant women. This risk does not end with the pregnancy; heart vessel changes from being pregnant can cause early death for the mother after the pregnancy is over.

A detailed cardiology (heart) exam with echocardiogram or magnetic resonance imaging (MRI) is warranted.

What else should a woman with TS know before considering pregnancy?

Before considering trying to get pregnant with donor eggs, women with TS should have a thorough cardiology exam and consult with specialists in maternal-fetal medicine and cardiology. Women with TS are advised to consider using a gestational carrier (a woman who carries a pregnancy for another).

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