Reproductive Facts

Patient fact sheet developed by the
American Society for Reproductive Medicine

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

What is Turner Syndrome (TS)?

Turner syndrome (TS) is a rare genetic disorder in females. About one in every 2,000 female babies born is affected.

What causes TS?

TS is not related to the genetic makeup of the mother or father.

TS is not related to the genetic makeup of the mother or father. It 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 a type of genetic testing called karyotype testing. Karyotype can either be performed on blood or on cells collected from the mouth (buccal swab). An affected baby can be diagnosed by testing cells collected by amniocentesis or placental cells collected by chorionic villus sampling. Karyotyping provides both a count of the number of chromosomes (genetic material) as well as information on appearance of each chromosome (normal versus abnormal). Females with TS have only a single complete "X" chromosome in their cells instead of two.

Are there any physical traits of Turner syndrome?

There are physical features that are common in girls with TS. Not all girls will have all of the characteristics. These characteristics include:

1. Short stature (typically under 5 feet tall)
2. Low-set ears
3. Receding lower jaw
4. Short and webbed neck
5. Scoliosis
6. Puffy hands and feet
7. Increased carrying angle of elbows
8. Narrow and high arched palate
9. Broad chest
10. Flat feet

Are there any health problems associated with TS?

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

1. Delayed puberty
2. Premature ovarian insufficiency ("early menopause")
3. Fertility problems due to lack of eggs
4. Hearing problems
5. Heart and blood vessel defects
6. Ear infections
7. Kidney problems
8. Thyroid problems
9. Cataracts
10. Diabetes
11. Risk for certain type of cancers, e.g. brain tumor
12. 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:

1. Growth hormone to increase height
2. Estrogen to help develop secondary sexual characteristics, such as breast development, in girls with delayed puberty
3. Estrogen to improve bone mass and strength
4. Medicine and surgery to correct heart and blood vessel defects
5. Infertility treatment

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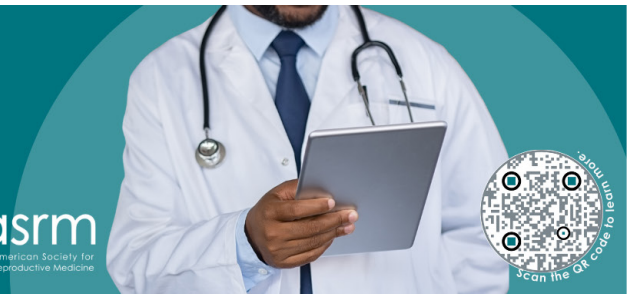

Does TS affect the ability to have children?

Infertility is common in girls with TS due to rapid loss of eggs within the ovaries. Spontaneous pregnancies are rare. However, pregnancy is possible with the use of donor eggs.

Is it safe for women with TS to become pregnant?

Women with TS are at a particularly high risk for developing heart problems during pregnancy. An evaluation of the heart with an echocardiogram or Magnetic Resonance Imaging (MRI) is warranted as risks of complications from heart problems are increased in pregnancy.

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

A woman with TS who is considering attempting pregnancy with donor eggs 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) to avoid possible life-threatening complications.

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