GTPIM106

Genetics of Turners Syndrome

Patient Information Material

What is Turner's Syndrome?

Turner's syndrome (TS) is a medical problem that affects one in 2500 girls. Most girls are of short stature with impaired sexual development and infertility. Other features may include, shield chest, widely spaced nipples, and webbed neck, bone deformities, small uterus/ovaries, cardiac defects, hypothyroidism and type 2 diabetes mellitus. Also, roughly about 10% of aborted fetuses have Turner Syndrome.

Why the condition is generally not detected in childhood?

It is a multi-system disorder that is not always diagnosed in the newborn period. Although there are certain early childhood symptoms like vitiligo, psoriasis, webbed neck, lymphodema, an awareness of the clinical condition throughout childhood, adolescence, and adulthood is necessary. Usually, the condition is diagnosed in teenage girls when they come with the chief complaint of primary amenorrhea (no menstrual periods).

What steps are undertaken for diagnosing TS?

Clinical assessment of Turner syndrome is based on the above mentioned clinical features. However, an ultrasound scan may be used as a screening test to detect the absence of a uterus and ovaries; this is followed by a confirmatory test i.e. Chromosomal Analysis or Karyotyping. The test is done on blood sample and requires culturing the cells in the lab. The most common abnormality seen is the absence of one X chromosome.

What results in Turner Syndrome (TS)?

The egg and sperm cells undergo cell division, where the 46 chromosomes are divided in half and the egg and the sperm cells end up with 23 chromosomes each. When a sperm with 23 chromosomes fertilizes an egg with 23 chromosomes, the embryo is complete with 46 chromosomes with a pair of sex chromosomes (XX in female and XY in male). Sometimes, an error during fertilization can cause a missing sex chromosome (X). This cell fails to contribute an X chromosome resulting in TS also known as "monosomy X". The loss of X can be in all cells (Standard type) or in a percentage of cell population (Mosaic type) or due to structural defect of X chromosome (Deletion).

How is Turner Syndrome Treated?

Turner syndrome is a condition that is caused by a chromosomal abnormality, thus no specific cure. However, physicians can help correct growth problems by administering hormones. Research is going on for new therapies.

Can a woman with TS have children?

Most of the women with TS cannot have children. However, in cases where uterine development is normal, pregnancy can result by in vitro fertilization (IVF) procedure that is used to create an embryo from donor egg followed by implant into the uterus (womb) of the woman with TS. Operative care and post delivery mother and child care are important.

Can TS be diagnosed before birth?

Chromosomal abnormalities such as Turner syndrome can often be diagnosed before birth by analyzing cells in the amniotic fluid or from the placenta, this is called Prenatal Diagnosis. Fetal ultrasound with cystic hygroma findings suggest Turner syndrome and require a prenatal diagnostic procedure. Ultrasound guided procedures like amniocentesis, chorionic villus biopsy and cordocentesis will allow testing of fetal samples during pregnancy.

Is Turner syndrome hereditary?

No. Although Turner Syndrome is caused by a genetic or chromosomal defect, it is not hereditary in nature. Most of the time it is an accidental event and recurrence of such events for future pregnancies is very low unless parents are carriers of any rare chromosomal rearrangements.

What happens in a genetic counseling session?

Counseling done by a qualified genetic counselor focuses on gathering factual information, explanation of causes of Turner Syndrome, it's types, prognosis, genetic testing, and current research.

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