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TURNER SYNDROME, WHICH OCCURS DURING PREGNANCY

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Annotation: this article provides information on turner syndrome, which occurs during pregnancy. You can find out what treatment measures are used in this case and the reasons for its origin in this article. The article is written in the case of using internet resources.

Key words: turner syndrome, X chromosome.

Turner syndrome results from a deletion or the non-functioning of one X chromosome in females. About half of the population with Turner syndrome have monosomy X (45,XO). The other 50% of the population has a mosaic chromosomal component (45,X with mosaicism).

Some types of anomalies in the X chromosome that can lead to a nonfunctioning X chromosome are as follows:

- Isochromosome Xq, where there are two copies of the long arm of the chromosome that are connected head to head.
- Ring chromosome, where a part of the ends of short and long arms of the X chromosome is missing
- Xp or Xq deletion, where the deletion of part of the short arm of the X chromosome takes place

Some patients with Turner syndrome can have a Y chromosome mosaicism. Although not a cause of Turner syndrome, the SHOX (short stature homeobox-containing gene on the X-chromosome) is associated with the short stature found in Turner syndrome. Turner syndrome is usually not inherited but is a random event during reproduction.

As per the literature, Turner syndrome is seen in about 1 in 2000 to 1 in 2500 live female births.However, the true prevalence remains unknown as many patients with a mild phenotype may remain undiagnosed or are diagnosed late in adulthood.The occurrence of Turner syndrome is almost the same in different ethnicities and different countries. With increased awareness of prenatal ultrasound scans, the prevalence of Turner syndrome at birth is decreasing; this is because some mothers carrying fetuses with Turner syndrome choose to terminate the pregnancy.

Most instances of Turner syndrome are not inherited. When monosomy X is the cause, the chromosomal abnormality is a random event during the formation of reproductive cells in the person’s parent. An error in cell division is called nondisjunction and can result in reproductive cells with an abnormal number of chromosomes. For example, a sex chromosome can become lost from an egg or a sperm cell due to nondisjunction. If an atypical reproductive cell contributes to the genetic makeup of a child, each cell will possess a single X chromosome, and the other sex chromosome will be missing.

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Mosaic Turner syndrome is likewise not an inherited condition. It occurs due to a random event during the cell division stage in the early fetal development of the affected individual. As a result, some of a person's cells have the usual two sex chromosomes, while other cells contain only one copy of the X chromosome. Other sex chromosome abnormalities are possible in females with X chromosome mosaicism. Rarely, Turner syndrome can result from a partial deletion of the X chromosome, and this can pass from one generation to the next.

Turner syndrome can be identified prenatally with abnormal ultrasound findings of increased nuchal translucency, nuchal cystic hygroma, coarctation of the aorta/left-sided cardiac anomalies, brachycephaly, horseshoe kidney, polyhydramnios, oligohydramnios or non-immune fetal hydrops. In the female newborn, Turner syndrome can present with congenital lymphedema of the hands and feet, webbed neck, nail dysplasia, narrow and high-arched palate, and short fourth metacarpals or metatarsals. As they grow up, the girls develop short stature, “shield” chest with widely spaced nipples, webbed neck, low hairline at the base of the neck, cubitus valgus, and Madelung deformity of the forearm and the wrist.

Patients with Turner syndrome usually have normal intelligence but may have specific neurocognitive deficits, e.g., problems with visuospatial organization. This situation can lead to an increased risk of learning disabilities, especially involving calculations, memory, and attention. In adolescence, females will often present with delayed puberty or primary amenorrhea, secondary to premature ovarian failure. “Streak gonads” are a characteristic of Turner syndrome. These are the ovaries, mainly consisting of connective tissue and no follicles or only a few atretic follicles.

Patients with Turner syndrome also have an increased risk of cardiovascular malformations, which in turn leads to increased mortality risk in these individuals. Some of the cardiac malformations are aortic valve abnormalities (primarily bicuspid aortic valve), elongated transverse aortic arch, pulmonary venous anomalies. Aortic dissection further increases the risk of death in these patients. Hearing loss is common due to either recurrent otitis media causing conductive hearing loss, or due to the defect in the outer hair cells on the cochlea causing sensorineural hearing loss. Renal anomalies are common in Turner syndrome and include collecting system malformations, positional abnormalities, and horseshoe kidneys.

Ocular abnormalities can present with Turner syndrome, such as nearsightedness or farsightedness, strabismus, amblyopia, epicanthic folds, ptosis, hypertelorism, and red-green color blindness.Turner syndrome increases the risk of autoimmune disorders, including hypothyroidism, celiac disease, and inflammatory bowel disease. Due to the presence of dysgenic gonads, females with Turner syndrome are at an increased risk of developing gonadoblastoma.

Turner syndrome may be prenatally diagnosed by chorionic-villus sampling or amniocentesis. Turner syndrome should be suspected when a prenatal ultrasound shows fetal hydrops, cystic hygroma, or cardiac defects. The diagnosis requires confirmation after birth with karyotype testing. Occasionally, the karyotype can be normal if it is mosaicism, and if there is a strong suspicion, a FISH study is an option in addition to the karyotype. Genetic testing with karyotype

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analysis is necessary to confirm the diagnosis in individuals with characteristic clinical features described above. The first step is a karyotype analysis with peripheral blood mononuclear cells.

In adolescence, the patients can present with either delayed onset of puberty or amenorrhea. Elevated levels of follicle-stimulating hormone (FSH) are suggestive of Turner syndrome, and the anti-Mullerian hormone (AMH) may be a more sensitive marker for predicting ovarian failure. If the initial karyotype is normal in a patient with clinically suspected Turner syndrome, a second karyotype should be performed using a different tissue like skin, buccal mucosa cells, or bladder epithelial cells.

Following a diagnosis of Turner syndrome, management includes evaluation for other associated abnormalities like cardiac anomalies, renal anomalies, and learning disabilities. Screening should be a part of the baseline evaluation, and patients should undergo periodic screening thereafter. At initial diagnosis, patients should get renal ultrasonography and cardiovascular evaluation, including echocardiography in infants and children, and MRI in older girls and women.

Some of the screening laboratory tests include:

At four years of age and above: serum TSH to screen for autoimmune thyroiditis and tissue transglutaminase with total IgA to screen for celiac disease.

At ten years of age and above: fasting blood glucose, glycated hemoglobin, ALT, AST, serum creatinine, and urinalysis to screen for diabetes mellitus, fatty liver, and kidney disease.

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