MEDIBEATS
The Medical Bulletin

In Clinical Signs

Turner Syndrome / Bonnevie-Ullrich Syndrome / Ullrich Turner Syndrome

Definition :

- Turner syndrome (TS) is a neurogenetic disorder characterized by partial or complete monosomy-X.

History :

- Turner syndrome also referred to as congenital ovarian hypoplasia syndrome was first described by Henri Turner, an Oklahoma physician in 1938.
- It is the most common sex chromosomal abnormality found in females.

Prevalence :

1/2500–5000 at birth.

Etiology :

- Turner syndrome is due to complete or partial chromosome X monosomy in all or some of the body cells. The missing chromosome is usually the paternal one. No apparent relationship with advancing maternal age has been described.
- 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 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.

Clinical Features :

- It is the most common cause of 1° amenorrhea.
- C - Cardiac defects (Bicuspid aortic valve, coarctation of aorta, aortic dissection).
- L – Lymphedema of hands and feet.
- O – Ovaries (streak), decrease fertility, increase risk of cancer.
- W – Webbed neck.
- N – Nipples (widely shaped/ shield chest), nail dysplasia, narrow and high-arched palate.

- S – Short stature (SHOX gene defect), short 4th metacarpals or metatarsals.
- Low hairline at the base of the neck, cubitus valgus, and Madelung deformity of the forearm and the wrist.
- Hearing loss.
- Renal anomalies include collecting system malformations, positional abnormalities and horseshoe kidneys.
- Ocular abnormalities such as myopia or hypermetropia, 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.
- Increase risk of Metabolic Syndrome.

Diagnosis :

- Prenatal chorionic villus sampling and/or amniocentesis with karyotyping.
- Postnatal karyotyping (if prenatal karyotyping not done).
- 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.
- Elevated levels of follicle-stimulating hormone (FSH) are suggestive of Turner syndrome.
- 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.

Prenatal USG findings :

- Increased nuchal translucency
- Nonimmune hydrops fetalis (NIHF)
- Cystic hygroma
- Hypoplastic left heart syndrome & coarctation of aorta
- Renal anomalies - horse-shoe kidney
- A short femur

Treatment :

- The management of patients with Turner syndrome is multidisciplinary.
- Treatment is basically symptomatic and complete recovery is not possible.
- The primary therapies for affected individuals are Growth hormone therapy & Estrogen therapy.
- For child bearing – IVF with donor oocyte.

Prognosis :

- Most fetuses die in utero, often in the first or early second trimester.
- An additional group dies later due to generalized hydrops.
- Only a minority of fetuses reaches term of gestation.
- After birth, these females show short stature, pterygia, borderline mental retardation and later amenorrhea from nonfunctioning streak gonads (primary hypogonadism).
- Survival is generally good, except for cases affected by hypoplastic left heart syndrome, which represents a life-threatening condition.
-

Dr. C. Naveen Kumar
Kangayam