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Turner Syndrome

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ABSTRACT
Turner syndrome is a rare chromosomal disorder that affects females. The disorder is characterized by partial or complete loss (monosomy) of one of the second sex chromosomes. TS is associated with certain physical and medical features including estrogen deficiency, short stature and increased risk for several diseases with cardiac conditions being among the most serious. The cognitive-behavioral phenotype associated with TS includes strengths in verbal domains with impairments in visual-spatial, executive function and emotion processing. Genetic analyses have identified the short stature homeobox (SHOX) gene as being a candidate gene for short stature and other skeletal abnormalities.

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I. Introduction

Turner syndrome is a rare chromosomal disorder that affects females. The disorder is characterized by partial or complete loss (monosomy) of one of the second sex chromosomes.[1]It is the most common sex chromosomal abnormality affecting girls and women.Turnersyndrome is named after Henry Turner who, in 1938, was one of the first doctors to report on the disorder in the medical literature[2]. It affects 1 in every 2000 to 2500 live births[3].TS is associated with certain physical and medical features including estrogen deficiency, short stature and increased risk for several diseases with cardiac conditions being among the most serious.[4]

MEANING:
Turner syndrome is a genetic condition caused by an abnormality on one of the sex chromosomes. It's also called monosomy X, gonadal dysgenesis.[5]

CAUSES:
1. Turner syndrome results when one normal X chromosome is present in a female's cells and the other sex chromosome is missing or structurally altered. The missing genetic material affects development before and after birth.
2. Turner syndrome can also occur if one of the sex chromosomes is partially missing or rearranged rather than completely absent. Some women with Turner syndrome have a chromosomal change in only some of their cells, which is known as mosaicism. Women with Turner syndrome caused by X chromosome mosaicism are said to have mosaic Turner syndrome.
3. Researchers have not determined which genes on the X chromosome are associated with most of the features of Turner syndrome. They have, however, identified one gene called SHOX that is important for bone development and growth.[6]

TYPES :
1. Classic Turner Syndrome (or non-mosaic Turner Syndrome) – all cells lack one X chromosome.
2. Mosaic Turner Syndrome – cells vary in chromosomal content, with some cells having both X chromosomes and others having an incomplete amount of X chromosomes (either with an absent X chromosome; with one full X chromosome and one incomplete X chromosome; or, in few cases, one full X chromosome and one incomplete Y chromosome)[7] .

SYMPTOMS:
1. Short stature. Most girls with Turner syndrome will be short for their age.
2. Typical facial features. This may include characteristic changes to the eyes, ears, mouth and neck – for example:

- Down slanting eyes

- Droopy eyelids.
- Prominent earlobes
- Crowding of the teeth
- A short webbed-like neck.

3.Bone features. For example:
- A broad chest with widely spaced nipples
- A wide carrying angle (this is the angle between the forearm and the hip when the arm is held by the side)
- Short fingers
- Nail changes
- Skin features : About 7 in 10 girls with Turner syndrome have a lot of moles and obvious blood vessels over the skin.
- Susceptibility to certain medical conditions. People with Turner syndrome are more likely to develop certain conditions such as: Diabetes mellitus
- Coeliac disease.
- An overactive thyroid gland (hyperthyroidism)
- inflammatory bowel diseases

4.Organ abnormalities: For example, about half will have an abnormality with the heart or major blood vessels. Up to 6 in 10 will have an abnormality of the kidneys or urinary tract.
5.Ovarian abnormalities. The ovaries are a pair of glands that lie on either side of the womb (uterus). Each ovary is about the size of a large marble. The ovaries normally make eggs (ova) and various female hormones - in particular, oestrogen. Almost all girls with Turner syndrome have ovaries that do not work properly. As a result, they:May not go through puberty.May have breasts which do not develop properly.May not ever get a period, and are likely to be infertile. However, up to 3 in 10 girls will have some changes of puberty and 1 in 200 may be able to get pregnant naturally[8] .
6. They may even suffer from learning disabilities.

COMPLICATION:
- Cardiovascular abnormalities: Heart valve disease is a prevalent abnormality, and patients with TS have a significantly higher incidence of aortic bicuspid

deformity. Patients with TS have a risk of dying mainly from an aortic dissection aneurysm[9] .
- Autoimmune diseases: TS causes a variety of autoimmune diseases such as thyroiditis, colitis, celiac disease, type 1 diabetes, and psoriasis, though the most common is Autoimmune thyroiditis. More commonly they suffer from hashimoto thyroiditis[10] .
- Skeletal abnormalities: Fractures are considered to be one of the major complications of TS.Landin-Wilhelmsen et al. found that osteoporosis and fractures are related to age in patients with TS; of 70 patients with TS, 16% had suffered a fracture and 50% were over the age of 45[11].

TREATMENT:
Although there is no cure for Turner syndrome, some treatments can help minimize its symptoms. These include
1. Human growth hormone. If given in early childhood, hormone injections can often increase adult height by a few inches.
2. Estrogenreplacement therapy (ERT). ERT can help start the secondary sexual development that normally begins at puberty (around age 12). This includes breast development and the development of wider hips. Healthcare providers may prescribe a combination of estrogen and progesterone to girls who haven't started menstruating by age 15. ERT also provides protection against bone loss.
3. Regular health checks and access to a wide variety of specialists are important to care for the various health problems that can result from Turner syndrome.These include ear infections, high blood pressure, and thyroid problems.[12]

TIPS FOR PARENTS DEALING WITH TURNER SYNDROME :
Girls with Turner syndrome might have specific medical problems and different physical characteristics. Parents or Caregivers could help children develop daily living skills and cope with new or challenging situations by doing the following.
1. Explain to the child about Turner syndrome so she would be able to explain the same to friends and relatives. Treat them according to their age and not size, especially while giving responsibility to them.
2. Arrange home so that it's comfortable for her (provide sturdy footstools in the bathroom and kitchen and easy access to clothing, closets, personal care items, and other necessities). Check her classroom

environment. Ask for help from teachers to provide the right accommodations so that she can reach water fountains, classroom materials, and supplies.

3. Help her cope with new situations and encourage her to ask her friends for help.Compliment her often on her strengths and coping skills.
4. Be patient, positive, and open to discussions about her limitations and fears.
5. Give medications on time.
6. Encourage participation in activities in which height isn't an issue, such as skating, diving, horseback riding, babysitting, or volunteer work. Volunteering, in particular, can be a big confidence booster
7. If she's depressed or has self-esteem problems, consider counselling with a mental health professional.
8. Don't ignore your instincts if you think she's sad or withdrawn.
9. Take care of yourself. You won't be able to provide good care for your daughter if you neglect your own needs.[13]

II. CONCLUSION:

Turner syndrome is a genetic condition where a female inherits one X chromosome instead of two. People with Turner syndrome have unique healthcare needs. An experienced healthcare team can make sure child gets the screening and support they need and help them live an independent and productive life. [14]

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