



TURNER SYNDROME—AN OVERAL VIEW

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Article Received on 17/04/2022

Article Revised on 07/05/2022

Article Accepted on 27/05/2022

ABSTRACT

Turner syndrome, a condition that affects only females, results when one of the X chromosomes (sex chromosomes) is missing or partially missing. Turner syndrome can cause a variety of medical and developmental problems, including short height, failure of the ovaries to develop and heart defects. Turner syndrome may be diagnosed before birth (prenatally), during infancy or in early childhood. Occasionally, in females with mild signs and symptoms of Turner syndrome, the diagnosis is delayed until the teen or young adult years. Girls and women with Turner syndrome need ongoing medical care from a variety of specialists. Regular checkups and appropriate care can help most girls and women lead healthy, independent lives. Turner syndrome (TS), sometimes referred to as congenital ovarian hypoplasia syndrome, is a genetic disorder. It is the most common sex chromosomal abnormality affecting girls and women. More specifically, it's a problem with one of the two X chromosomes -- the thread-like structures inside cells that are made of DNA. We get our DNA from our parents and it is the DNA that contains the specific instructions that make each living creature unique.

KEYWORDS: X chromosomes, ovarian hypoplasia, Regular checkups, DNA.

What is turner syndrome

Turner syndrome is a congenital condition, meaning it's something a person is born with. Each of us is born with two chromosomes. If you're female, you were born with two X chromosomes. If you're a male, you are born with one X and one Y chromosome. Turner Syndrome occurs when one of the X chromosomes is missing, either partially or completely. Turner syndrome often causes short stature, typically noticeable by age 5. It usually doesn't affect intelligence but can lead to developmental delays especially with calculations and memory. Heart problems are common, too. While TS can somewhat shorten life expectancy, screening for and treating known related conditions helps protect health.

Turner syndrome is a **genetic disorder, but it's usually not inherited**, except in rare cases. An inherited genetic condition means that a parent (or both parents) passed down a mutated, or changed, gene. In Turner syndrome, the chromosome change happens randomly before birth.

What are the types of turner syndrome?

The **type of Turner syndrome (TS)** a person has depends on the problem with the X chromosome: **Monosomy X:** Each cell has only one X chromosome instead of two. About 45% of people with TS have this type. It comes from the mother's egg or the father's sperm randomly forming without an X chromosome. After fertilization, the baby's cells also contain this

defect. **Mosaic Turner syndrome:** Also called 45, X mosaicism, this type makes up about 30% of Turner syndrome cases. Some of the baby's cells have a pair of **X chromosomes changes**, while other cells only have one. It happens randomly during cell division early in pregnancy. Worldwide, about 1 in every 2,500 female babies is born with Turner syndrome. It's the most common sex chromosomal abnormality found in women. Y chromosome material. In a small percentage of Turner syndrome cases, some cells have one copy of the X chromosome and other cells have one copy of the X chromosome and some Y chromosome material. These individuals develop biologically as female, but the presence of Y chromosome material increases the risk of developing a type of cancer called gonadoblastoma.

Inherited turner syndrome: In rare cases, babies may have inherited TS, meaning their parent (or parents) were born with it and passed it on. This type usually happens because of a missing part of the X chromosome.

Symptoms of turner syndrome?

The main symptom of Turner syndrome is short stature. Almost all females with TS:

- 1) Grow more slowly than their peers during childhood and adolescence.
- 2) Have delayed puberty and lack of growth spurts, resulting in an average adult height of 4 feet, 8

inches. (If diagnosed early, growth hormone can help the person reach near-normal height).

- 3) Another symptom is not experiencing typical sexual development. Most females with TS:
- 4) Don't experience breast development.
- 5) May not have menstrual periods.
- 6) Have small ovaries that may only function for a few years or not at all.
- 7) Typically, don't go through puberty, unless they get hormone therapy in late childhood and early teens.
- 8) Don't make enough sex hormones.
- 9) Besides short stature, females with Turner syndrome often have certain physical traits:
- 10) Broad chest.
- 11) Cubitus valgus, where the arms point out slightly at the elbows.
- 12) Dental problems.
- 13) Eye problems, such as a lazy eye or drooping eyelids.
- 14) Scoliosis, when the spine curves sideways.
- 15) Low hairline at the back of the neck.
- 16) Many skin moles.
- 17) Missing knuckle in a particular finger or toe, making the digit shorter.
- 18) Narrow fingernails and toenails.
- 19) Small lower jaw.
- 20) Swelling of the hands and feet.
- 21) Unusually short, wide neck or webbed neck (extra skin folds).

What **other medical problems** might people with Turner syndrome have?

Cardiovascular problems

People with Turner syndrome (TS) may have heart and blood vessel problems, some of which can be life-threatening. Up to 50% of people with TS have a problem with the structure of their hearts. Cardiovascular problems can include:

- Bicuspid aortic valve, with two valve leaflets instead of three.
- Coarctation of the aorta, when part of the aorta is too narrow.
- Elongation of the aortic arch, a section of the body's main artery.
- Hypertension (high blood pressure).
- Bone problems

Should exercise and get enough calcium and vitamin D for healthy bones. Bone problems include:

- Increased risk for fractures and osteoporosis (weak bones), particularly in women who haven't received estrogen treatment.
- Scoliosis, in about 20% of people.
- Other medical conditions

People with turner syndrome may also have:

- Autoimmune disorders: People with TS may develop hypothyroidism (not enough thyroid hormone). They may also develop celiac disease

(digestive disorder) and inflammatory bowel disease (digestive tract irritation).

- Hearing and ear problems: An incorrectly formed ear and frequent middle ear infections are common. Sensorineural hearing loss develops in more than 50% of adults.
- Kidney defects: Structural problems in the kidney-urinary system happen in about 30% to 40% of people with TS. Problems with urine flow can lead to kidney infections.
- Metabolic syndrome: People with TS are at high risk for metabolic syndrome, a group of problems that occur together. It includes central obesity (more excess weight around the waist area), insulin resistance (pre-diabetes), high blood pressure, high cholesterol, high triglycerides and Type 2 diabetes.
- Mental health challenges: They may have low self-esteem and anxiety or depression. They may feel this way because of their physical concerns, health problems and infertility.
- Vision problems: Both farsightedness and nearsightedness can occur along with red-green color blindness, but nearsightedness is the most common eye problem.
- Do people with Turner syndrome have developmental delays?
- People with Turner syndrome typically have normal intelligence. But they may have problems with visual motor and visual spatial skills. They can have difficulty seeing how objects relate to each other in space. For example, driving may be difficult.

People with turner syndrome may also have trouble with:

- Clumsiness.
- Executive functions, including problems with managing and planning, memory, attention, and cognitive flexibility -- or changing the way they think about something.
- Nonverbal problem solving, like math.
- Understanding social cues, like facial expressions.

Effects of the missing or changed chromosome

The missing or changed X chromosome of Turner syndrome causes problems during fetal development and other developmental problems after birth — for example, short stature, ovarian insufficiency and heart defects. Physical characteristics and health complications that arise from these chromosomal issues vary greatly.

Complications

Turner syndrome can affect the proper development of several body systems, but this varies greatly among individuals with the syndrome. Complications that can occur include:

- Heart problems. Many infants with Turner syndrome are born with heart defects or even slight abnormalities in heart structure that increase their risk of serious complications. Heart defects often include problems with the aorta, the large blood

vessel that branches off the heart and delivers oxygen-rich blood to the body.

- High blood pressure. Turner syndrome can increase the risk of high blood pressure — a condition that increases the risk of developing diseases of the heart and blood vessels.
- Hearing loss. Hearing loss is common with Turner syndrome. In some cases, this is due to the gradual loss of nerve function. An increased risk of frequent middle ear infections can also result in hearing loss.
- Vision problems. An increased risk of weak muscle control of eye movements (strabismus), nearsightedness and other vision problems can occur with Turner syndrome.
- Kidney problems. Turner syndrome may be associated with malformations of the kidneys. Although these abnormalities generally don't cause medical problems, they may increase the risk of urinary tract infections.
- Autoimmune disorders. Turner syndrome can increase the risk of an underactive thyroid (hypothyroidism) due to the autoimmune disorder Hashimoto's thyroiditis. There is also an increased risk of diabetes. Sometimes Turner syndrome is associated with gluten intolerance (celiac disease) or inflammatory bowel disease.
- Skeletal problems. Problems with the growth and development of bones increase the risk of abnormal curvature of the spine (scoliosis) and forward rounding of the upper back (kyphosis). Turner syndrome can also increase the risk of developing weak, brittle bones (osteoporosis).
- Learning disabilities. Girls and women with Turner syndrome usually have normal intelligence. However, there is increased risk of learning disabilities, particularly with learning that involves spatial concepts, math, memory and attention.
- Mental health issues. Girls and women with Turner syndrome may have challenges functioning in social situations, may experience anxiety and depression, and may have an increased risk of attention-deficit/hyperactivity disorder (ADHD).
- Infertility. Most females with Turner syndrome are infertile. However, a very small number may become pregnant spontaneously, and some can become pregnant with fertility treatment.
- Pregnancy complications. Because women with Turner syndrome are at increased risk of complications during pregnancy, such as high blood pressure and aortic dissection, they should be evaluated by a heart specialist (cardiologist) and a high-risk pregnancy doctor (maternal-fetal medicine specialist) before pregnancy.

Treatment

- 1) Oestrogen and progesterone replacement therapy
- 2) Oestrogen and progesterone replacement therapy may also be recommended. Oestrogen and progesterone are the female hormones responsible

for sexual development. Oestrogen also helps prevent brittle bones (osteoporosis).

- 3) In girls with Turner syndrome, the ovaries (female reproductive organs) do not work properly. As a result, the girl may not go through puberty and is likely to be unable to have a baby without help (be infertile).
- 4) Women with Turner syndrome usually need regular sex hormone treatment until they're about 50. After this time, the body usually stops producing Oestrogen and monthly periods stop. This is called the menopause.
- 5) Oestrogen replacement therapy is usually started around the time of normal puberty. In girls, this is around 11 years old. It may be recommended that Oestrogen replacement is started earlier in gradually increasing doses. Treatment will be tailored to the specific needs of each girl.
- 6) Oestrogen triggers the changes that usually happen during puberty, such as breast development. It's important for the health of the womb and bones (protecting against osteoporosis).
- 7) It can be given as a gel, tablet or patch. Low doses are used to start with and gradually increased to adult levels with time and age, to mimic normal puberty.
- 8) Progesterone replacement therapy is usually started after Oestrogen therapy and will cause monthly periods to start. It also may be given alone or combined in a tablet or patch with Oestrogen
- 9) Most women with Turner syndrome are unable to have children (infertile). A minority will be able to conceive naturally, so girls and women with Turner syndrome should have access to sexual health and contraception advice.
- 10) Assisted conception techniques, such as egg donation and in vitro fertilization (IVF), may be recommended for women with Turner syndrome who want to have children.
- 11) If a woman has Turner syndrome and becomes pregnant, she'll need regular heart checks because the heart and blood vessels will be put under extra strain during pregnancy. It's vital her womb is healthy and has developed fully during puberty for the pregnancy to be successful.
- 12) Some girls and women with Turner syndrome may develop psychological problems, such as low self-esteem or depression.
- 13) Doctors sometimes attribute these problems to physical appearance or infertility, but they're more commonly related to difficulties understanding other people's social behavior and how to respond appropriately. Associated problems often arise at home, in school and in the workplace.
- 14) Psychological therapy, such as counselling or cognitive behavioral therapy (CBT), may be recommended.

CONCLUSION

Turner's syndrome is a chromosomal disorder, a result of the partial or total absence of the second sexual chromosome. A relatively frequent alteration (1:2500 newborn girls), it presents a phenotypical variability which may cause a sub-diagnosis of its cases. It is important to know the clinical and genetic aspects which will lead to a timely detection of cases, and thus a proper management of associated co-morbidities to this genetic condition.

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