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Short Communication

# An Overview of Turner Syndrome

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## DESCRIPTION

Turner syndrome, a subtype of Down syndrome, is a genetic disease that affects female development. The most common sign of Turner syndrome, which develops around the age of five, is short height. Premature ovarian failure (ovarian hypo function) is also rather prevalent. The ovaries develop correctly at first, but egg cells (oocytes) die prematurely and the majority of ovarian tissue degenerates before the baby is born. Many of the affected girls do not reach puberty without hormone therapy, and the majority of them are unable to conceive (infertile). Only a small percentage of Turner syndrome females reach adulthood with normal ovarian function. About 30% of females with Turner syndrome have extra folds of skin on the neck (webbed neck), a low hairline at the back of the neck, puffiness or hand and foot swelling (lymphedema), skeletal deformities, or kidney issues. A cardiac abnormality, such as a constriction of the big artery exiting the heart (coarctation of the aorta) or abnormalities of the valve that joins the aorta to the heart, affects one-third to one-half of people with Turner syndrome (the aortic valve). These heart abnormalities can lead to life-threatening complications [1-4].

Turner syndrome affects the majority of girls and women of average ability. Although these traits differ among afflicted individuals, developmental delays, nonverbal learning difficulties, and behavioral issues are conceivable. This illness affects roughly one out of every 2,500 young girls around the world, but it is far more common in pregnancies that do not make it to term (miscarriages and stillbirths).

The X chromosomal defect identifies the type of Turner syndrome (TS) an individual seems to have: Monosomy X is a condition in which each cell has only one X chromosome rather than two. About 45 percent of TS sufferers have this type. It develops when the mother's egg or the father's sperm develop at random without an X chromosome.

Inherited Turner syndrome, in rare cases, children may inherit Turner Syndrome (TS), which means that their parent (or parents) were born with the disorder and passed it on to them. A missing part of the X chromosome is frequently the cause of this sort.

The X chromosome, one of the two sex chromosomes, is linked to Turner syndrome. Each cell in the human body has two sex chromosomes: females have two X chromosomes and men have one X chromosome and one Y chromosome. Turner syndrome is caused by the presence of one normal X chromosome in a female's cells and the absence or structural alteration of the other sex chromosome both during birth, the lack of genetic material has an effect on development. About half of people with Turner syndrome have monosomy X, which means that each cell in their body only has one copy of the X chromosome instead of the usual two. Turner syndrome can also develop when one of the sex chromosomes is missing or altered but not fully gone. Mosaicism is a chromosomal alteration that occurs in only some of the cells of some women with Turner syndrome. Mosaic Turner syndrome is a kind of Turner syndrome characterized by X chromosomal mosaicism in women. Researchers are still trying to figure out which genes on the X chromosome are linked to the majority of the symptoms of Turner syndrome. However, they have discovered a gene called SHOX, which is vital for bone development and growth. Turner syndrome is caused by the loss of one copy of this gene, which results in short height and skeletal deformities in women.

## CONCLUSION

Turner syndrome is not inherited in the majority of cases. The chromosomal anomaly develops as a random occurrence during the production of reproductive cells (eggs and sperm) in the affected person's parent when this condition is caused by monosomy X. Nondisjunction, a type of cell division defect, can result in reproductive cells with an incorrect number of chromosomes. Nondisjunction can cause an egg or sperm cell to lose a sex chromosome, for example. If one of these atypical reproductive cells contributes to a child's genetic makeup, the child will have only one X chromosome in each cell and the other sex chromosome will be missing. It happens at random during cell division in early foetal development in an affected individual. As a result, certain cells in an affected person's body have two sex chromosomes, while others only have one copy of the X chromosome. Females with X chromosomal mosaicism may also have other sex chromosome abnormalities. Turner syndrome, which is caused by a partial deletion of the X

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chromosome, is rarely passed down from generation to generation.

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