

Chromosomes

Name

Institution Affiliation

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Turner syndrome is due to a complete or incomplete lack of the second sex chromosome. Around half have lost their second chromosome entirely. The other half possesses variations such as mosaicism with one abnormal cell line and a single normal cell line (Zhong & Layman, 2012). This condition is found in nearly one in every 2,000 live female births. Still, approximately 99% of embryos are lost prenatally (De Marqui, 2015).

Symptoms of Turner Syndrome

At birth, the infant exhibits a short fourth metacarpal, gonadal dysgenesis, webbed neck, and nail dysplasia. In childhood, girls display delayed puberty and a short stature. Moreover, there is substantial cognitive impairment in the visual and mathematical areas. At adulthood, women exhibit infertility. Ninety-eight percent of women with Turner Syndrome have a failure of the ovaries (De Marqui, 2015). Moreover, there is a high risk for osteoporosis, hypertension, and hyperthyroidism.

Turner Syndrome: Cellular Processes

During the fusion of the sperm and the egg, the DNA from each split their chromosomes and afterward fuse back together with the new egg and sperm. The chromosome becomes incomplete when one of the chromosomes fails to replicate to fuse with the other cell.

The possibility of treating aneuploidies in utero. Not only can aneuploidy and structural fetal anomalies be detected through a sonography in the first trimester, but the detection provides opportunities for improved treatment options. Properly conducted ultrasound in the first trimester can be utilized in identifying aneuploidy.

Personal opinion of treating aneuploidies in utero. I am convinced that it should be done if there is technology to handle the treatment in utero. A third of girls displaying Turner

Syndrome have kidney malformations. Although these incidences do not lead to medical problems, they put them at a higher risk of acquiring urinary tract infections and high blood pressure (Zhong & Layman, 2012). The treatment of aneuploidies in utero may assist these girls to live a normal life.

References

- De Marqui, A. B. (2015). Turner syndrome and genetic polymorphism: a systematic review. *Revista Paulista de Pediatria (English Edition)*, 33(3), 363-370.
doi:10.1016/j.rppede.2015.06.001
- Zhong, Q., & Layman, L. C. (2012). Genetic considerations in the patient with Turner syndrome—45,X with or without mosaicism. *Fertility and Sterility*, 98(4), 775-779.
doi:10.1016/j.fertnstert.2012.08.021