A CASE OF TURNER SYNDROME IN AN ADOLESCENT WOMAN WITH RARE VARIANT OF KARYOTYPE

Salmerah G. Panambulan-Bantuas, MD; Marie Janice S. Alcantara-Boquerin, MD, FPOGS, FPSRM, FPSGE

Department of Obstetrics and Gynecology, Davao Regional Medical Center

INTRODUCTION

Turner syndrome (TS) is one of the most common chromosomal disorders and likely the most common genetic disorder of females. It affects 1 in 2,000 to 2,500 female live-births. However, TS may present with variants of karyotype and clinical features which warrants further and thorough investigation due to the complications that may occur with the condition

The degree which patients are affected is determined by the specific chromosomal abnormality and it may have only a few features which may be associated with the syndrome (Chen et al, 2016).

According to studies, majority of the patients with TS have 45, X karyotype, the classic type, occurring in 50% of cases and some will have mosaicism occurring in 30% of cases. The isochromosome or structural abnormality accounts for the remaining 20%. This case report aims to present a rare variant karyotype of TS, its work-up and management.

CASE PRESENTATION

A 20-year-old, nulligravid, single consulted at the out-patient department due to primary amenorrhea. On history, the birth event, perinatal and neonatal periods were unremarkable. There were no significant medical history and any prior surgeries. The patient has not attained menarche yet. Her growth development was at par with age. However, during her adolescent period, delayed cognitive function was noted which led her to have poor academic performance and often times conflict with her peers.

The patient is phenotypically female, with normal BMI. Physical examination revealed a broad chest with wide spaced nipples, upper extremitie in cubitus valgus, and Tanner stage 1 for breast and genitalia (Figure 1).

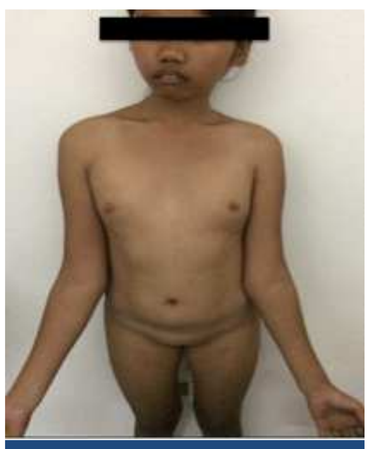

Figure 1

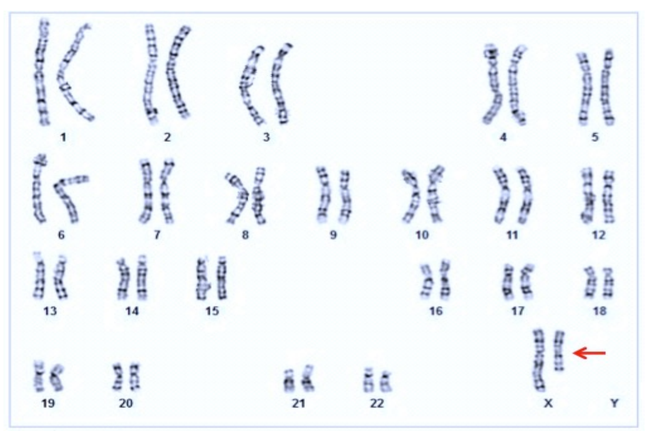

Figure 2

Transrectal ultrasound showed an infantile uterus, with 0.8x0.7cm left ovary and no appreciable right ovary. Karyotyping requested showed a normal one X chromosome and unbalanced translocation between short arm of another X chromosome (Xp) and long arm of chromosome 20 (Figure 2). Hormonal work-up showed a hypergonadrophic hypogonadism compatible with that of a postmenopausal woman. Additional imaging studies such as KUB Ultrasound and 2D Echo were all normal. Laboratory work up such as liver enzymes, 75g OGTT and Thyroid panel tests were in normal limit.

DISCUSSION

Turner syndrome is a disorder in females characterized by the absence of all or part of a normal second sex chromosome leading to a constellation of physical findings that often include congenital lymphedema, short stature and gonadal dysgenesis (Sybert and McCauley, 2004).

In the index patient, a rare variant of TS is presented that with a ischoromosome variation with a specific karyotype of (46X, der (X;20) (p10; q10), +20. The patient did not present with the classic physical findings of a TS patient. She had no overt physical abnormalities in childhood and initially had a good functional status hence causing the delay of seeking medical attention.

Almost all patients with TS exhibit short stature, however, in rare cases such as in our patient, it was not appreciated. It was mentioned on the study of Sybert and McCauley (2004) that deletions distal to Xq21 may have no effect on stature. Another common finding in TS patients is absence of sexual characteristics. This is the most common feature of TS in which 90% has gonadal failure which may attributed to the loss of terminal long arm material of X chromosome. The patient presented with primary amenorrhea due to ovarian dysfunction. Intelligence is usually normal in TS patients usually with good verbal and reading skills. However, they may develop learning difficulties in visual-spatial relationships. Our patient, on the other hand, had delayed cognitive function, with nonverbal learning disabilities, impaired peer relationships and immaturity.

On further work-ups, there were no other notable major complications of TS such as cardiac and kidney problems, endocrine disorder (hypothyriodism and diabetes mellitus), dermatologic and skeletal development.

CONCLUSION

Our case was an isochromosome variant of TS who presented with primary amenorrhea, non-typical physical abnormalities of the usual TS patient and delayed cognitive funstion. Further screening showed hypergonadotrophic hypogonadism which may lead to infertility. In the patient, the cognitive disabilities were the main dilemma, in which family support is highly recommended. Knowledge of the variants of TS is necessary for better prognostication and treatment.

REFERENCES

1. Luleci, G., Hosgor, O., Karauzam, S., Uner, M. A rare case of Turner's syndrome presenting with Mullerian agenesis. Journal of Human Reproductive Sciences. December 2013. Hereditas 113: 217-220.
2. Ramos, H., and Ang, F. A variant of Turner's syndrome presenting with Secondary Amenorrhea. Philippine Journal of Internal Medicine. September 2010. Volume 48, 41-43.
3. Muntaj, S., Ganaie, F., Purva, S., Radhika, S., Tilak, P., Karyotypic Variables in Turner syndrome: A Case Series, 2015. International Journal of Scientific Study.July 2015. Vol 4, Issue 4
4. Sybert, V. and McCauley, E. Turner's Syndrome. The New England Journal of Medicine. September 2004. 351: 1227-1238.
5. Chen, L., Sun, H., Chen, T., Xie, R., Wang, F., Chen, X., Tian, J. A rare case of Turner syndrome with a special karyotype: a case report and review of literature. International Journal of Clinical and Experimental Medicine 2016; 9 (11)