A (Y,12) TRANSLOCATION PRESENTING AS AZOOSPERMIA AND EARLY MATURATION ARREST – REVIEW of (Y,AUTOSOME) TRANSLOCATIONS

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INTRODUCTION

Translocations between Y chromosome and autosomes (Y/A), excluding between Yq12 and short arm of acrocentric chromosomes, are relatively rare and associated with infertility in 80% patients.[1] Less than 5 cases with a (Y,12) translocation have been reported in the literature, including this case.

PATIENTS

A 36 year-old man was admitted to our Department of Infertility because of history of 10 year primary infertility. Her wife was healthy and 29 years old. He had been operated for bilateral varicoceles 6 years ago. He was phenotypically and clinically normal, including testicular size, epididymis and vas deferens. Semen analysis showed azoospermia. FSH, LH, Prolactin and Testosterone levels were within normal range, except low T/E2 ratio (9.2). Cytogenetic analysis found a reciprocal translocation between Yq12 and 12p11.2. The karyotype was 46,XY, t(Y,12)(q12,p11.2) (**Figure 1.**). No microdeletions within AZFa/b/c regions. Single seminiferous tubule – biopsy and TESE were performed for eventual sperm retrieval, however, no spermatids or spermatozoa were detected. Bilateral testicular biopsy showed complete maturation arrest at the secondary spermatocyte stage.

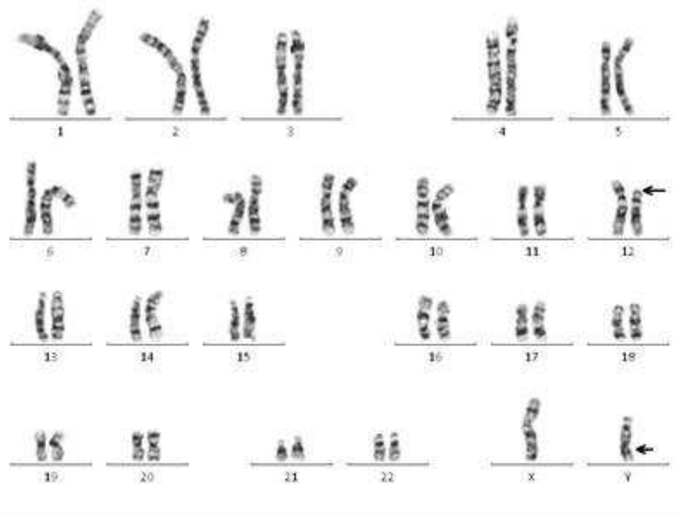

Figure 1. Patient' karyotype showed a translocation between a long arm of Y chromosome and a short arm of chromosome 12. Genetic breakage occurred at Yq12 and 12p11.2.

Chromosomal translocation occurs 4-10 times more frequently in infertile than fertile men.[2] (Y-autosome) translocations are rare and involve all autosomes except chromosomes 11 and 20.[1,3] (Y;12) translocations reported in the literature include 46,XY-12,+der(12)t(Y;12);[1] 46,XY,t(Y;12)(q12;q24.33);[4] 46,XY, t(Y,12)(q12,p13.3);[3] and our case 46XY,t(Y,12)(q12,p11.2).

The most common type is translocation of the heterochromatic region of Y chromosome on the short arm of acrocentric chromosomes (13, 14, 15, 21, 22).[1,5] The heterochromatic region of Y chromosome (Yq12) contains no genes involving in the spermatogenesis and no genetic information, and therefore, does not affect the patient phenotype and fertility.[3] Except this type of translocation, any (Y, autosome) translocation certainly leads to phenotype abnormalities and or infertility (80% of patients).[1] Translocation between sex chromosome and autosomes has greater effects on fertility than those between autosomes.[6]

Pathogenic mechanisms of infertility in these reciprocal translocations are due to disorders of X and Y chromosome pairing and or breakpoints at the AZF regions of Yq11.[4]

Maturation arrest at the spermatocyte or spermatid is the typical histology of the rearrangement between Y chromosome and autosomes.[7] Recently, meiotic defects resulting to maturation arrest have been described in detail in Li and his colleagues' study.[8]

CONCLUSION

We report a balanced reciprocal translocation between Y chromosome (Yq12) and autosome number 12 (12p11.2) causing azoospermia due to early maturation arrest. More studies are needed to better understand clinical manifestations, underlying mechanisms and effects on patient's fertility.

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