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A unique Y/Y translocation in an infertile male

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Abstract

A monocentric and submetacentric Y/Y translocation without evidence of mosaicism was observed in a male with tall stature and azoospermia. Chromosome measurements of the abnormal Y of this patient and the normal Y of his father were undertaken to determine more precisely the chromosome break points in this translocation. The measurements failed to show a definite loss of chromatin from the original short arm of the proband's Y chromosome. The azoospermia and infertility in our patient, in contrast to his father, suggest that infertility may be associated with this specific Y/Y translocation in human males.

Many chromosome translocations are well documented and their phenotypic effects known, while other chromosome translocations remain inadequately studied because of their rarity. The Y/Y translocation is one of those infrequently observed translocations in man for which little information is available relevant to its phenotypic effect. To our knowledge, only one case has been previously reported (Wahlstrom et al., 1976). In that report the individual was found to be a mosaic, with the majority of fibroblasts showing a 45,X chromosome complement lacking the abnormal Y chromosome. Here, we report a patient with a similar aberrant Y chromosome without evidence of mosaicism.

A 27-year-old white male was referred to us for evaluation of infertility. He had been married for 2 years to a 25-year-old woman who had had one child by a previous marriage. The couple had practiced normal, unprotected intercourse for 1 year without establishing a conception.

The patient gave a history of using LSD and marijuana on a regular basis at the age of 17 years, but he had not used drugs for several years. His past medical and family history were unremarkable except for gonorrheal urethritis, which was successfully treated in 1971. He had been exposed to no excessive radiation or industrial toxins.

Physical examination revealed a tall, slender, and well-developed white male with normal mentality and a height of 6 ft, 6 in. He had a normal male hair distribution and body musculature. No abnormality was found on examination of the head and neck, nor was there evidence of cardiovascular disease or abnormalities. The chest revealed no gynecomastia, and the abdomen was normal. The testicles were

somewhat small, measuring 3.5 cm in length by 2.5 cm in width, and were normally positioned in the scrotum. Rectal examination showed good anal tone and a normal prostate. The neurological examination was unremarkable.

Semen analyses performed on three separate occasions over a period of several months showed azoospermia. The hemogram and urinalysis were normal, as were blood chemistry studies. The blood follicle stimulating hormone (FSH) level was 100 mU/ml, with a normal male value of 4–20 mU/ml; the lutenizing hormone (LH) level was 10.5 mU/ml, with a normal male value of 4–20 mU/ml; and the serum testosterone level was 625 ng/dl, with a normal male value of 300–800 ng/dl.

The testicular histology was consistent with maturation arrest to the primary spermatocyte level. The seminiferous tubules showed normal numbers of Sertoli cells, spermatogonia, and primary spermatocytes, but no spermatids or spermatozoa were seen in the lumen.

Cytogenetic investigations were performed on three separate occasions from peripheral blood lymphocytes, buccal cells, and skin fibroblasts in an attempt to detect possible intertissue mosaicism. Visual and photographic analysis of 139 metaphase plates derived from peripheral blood lymphocytes and 210 metaphase plates obtained from skin fibroblast cultures revealed a consistent 46,X,t(Y;Y)(qter→p11::q11→qter) chromosome complement (fig. 1). This was confirmed by GTG-, QFQ-, and CBG-banding procedures. The buccal cells were 40% Y-chromatin positive, with a normal male control of 41%. The Y-chromatin was larger and more oblong in shape in this patient's cells than in normal male controls. There was no evidence of X-chromatin in the buccal cells analyzed.

Peripheral blood was also obtained from the patient's father for chromosome analysis. His chromosomes were analyzed utilizing GTG- and QFQ-banding procedures, and no evidence of an abnormal Y chromosome was obtained.

Vernier caliper measurements from karyotypes of our patient and his father were undertaken to assist in determining the origin of the Y/Y translocation. Our measurements showed no significant difference in the length of the short arms of the proband's original Y chromosome in the Y/Y translocation and the father's normal Y chromosome. Therefore, a marked deletion of the short arm of the Y chromosome was not detected by the use of the chromosome measurements described. However, we cannot rule out the possibility that some genetic information was lost as a result of a small deletion of the terminal portion of the short arm.

It is difficult to determine the precise cytological origin of this chromosome. It does appear that most, if not all, of the Yp material is present in this chromosome, as well as two copies of Yq material, based on unbanded and banded (QFQ, CBG, and GTG) studies and chromosome measurements of the proband and his father. There was a difference in length of the two arms and the lightly fluorescent material of the aberrant Y chromosome, producing a sub-metacentric structure. With QFQ- and GTG-banding it was found that there was a difference in banding pattern between the long and short arms of the aberrant Y chromosome. This difference resided in the area nearest the centromere.

All human chromosomes have C bands, but the centromeric band may be difficult to identify in the Y chromosome. Therefore, the chromosomes were stained with the G-II procedure to identify the centromeric area and a single centromere was seen. Apparently, the submetacentric Y chromosome has one centromere.

The exact origin of the aberrant Y chromosome in the paternal germ line is unknown. The following hypothesis was considered: Nondisjunction occurred during paternal meiosis, giving rise to two Y chromosomes. Two translocation chromosomes were then formed by a reciprocal translocation. The smaller translocation chromosome was lost, whereas the larger translocation chromosome represented the aberrant Y chromosome of our patient. This appears to be the simplest explanation for the origin of the submetacentric aberrant Y chromosome of our patient without evidence of mosaicism.

It is conceivable that a pericentric inversion or deletion of an isochromosome may have occurred to form the different arm lengths, but this would require at least two separate physical rearrangements. These explanations appear less likely and are not supported by the morphological and banding characteristics of the Y chromosome of our patient and of his father. Furthermore, a patient with an isochromosome for Yq generally presents with some stigmata of Turner's syndrome, which our patient did not.

If the origin of the Y chromosome in our patient arose following a nondisjunctional event and reciprocal translocation, then the terminal portion of the short arm of the Y chromosome of our patient may be deleted. If this is true, it would provide additional evidence that the gene(s) necessary for spermatogenesis is located on the terminal portion of the short arm of the Y chromosome in man.

References

- Wahlstrom J; Akesson HO, and Eriksson B: A man with presumptive Y/Y translocation, observed in a forensic psychiatric department. Clin. Genet 10: 82-88 (1976). [PubMed: 954229]

Fig. 1.
The Y chromosome of the patient stained with Giemsa (a); the GTG procedure to show G-bands (b); the CBG procedure to show C-bands (c); and the G-11 procedure (d).