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Short Communication

UNUSUAL STAINING PROPERTY OF HUMAN Y-CHROMOSOME

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Several structural aberrations of the human Y chromosomes such as isochromosome, dicentrics, rings, inversions and deletions have been found in phenotypically abnormal as well as normal males. Employing advent banding techniques, some aberrations have now been proven as variant (polymorphism or heteromorphism) Y chromosomes (Verma *et al.*, 1982). With the QFQ technique, (ISCN, 1978), the distal two-thirds of the long arm of the human Y chromosome is brightly fluorescent. We report a case of a physician whose Y chromosome is unusual by QFQ technique and more so by CBG and RFA techniques.

This male was selected from a project to study the acrocentric heteromorphism in an East Indian population. He is clinically normal with a negative medical history. He is neither exposed to ionizing radiation or any chemical mutagens or drugs. He has two clinically normal daughters. His family members are in India and were not available for cytogenetic evaluation but they are clinically normal.

Chromosome preparations were made from cultured peripheral blood. First, the QFQ technique was performed (Verma and Dosik, 1976). An unusual Y chromosome was noted that led to further analysis by the CBG and RFA techniques (Sumner, 1972; Verma and Lubs, 1975) at high resolution. A total of 75 cells were examined by multiple banding techniques. The morphology of all autosomes and X-chromosome was normal. No breaks, gaps, or exchanges were seen.

The first impression was that this individual had a very long Y chromosome. Therefore, the length of the Y chromosome was measured using Y/F index (Verma *et al.*, 1982). It was 1.56 ± 0.11 which is the longest Y among 70 normal males examined from India. Secondly, the long arm of the Y chromosome was segmented with 'brilliant' and 'dull' fluorescent. At least four brilliant segments can be observed at high resolution by the QFQ technique (Fig. 1). This impression led to believe that these four brilliant segments should stain very darkly by the CBG technique. After employing the CBG technique, a more unusual situation was noted. In fact, there were two cells where Y chromosome appeared metacentric. Although

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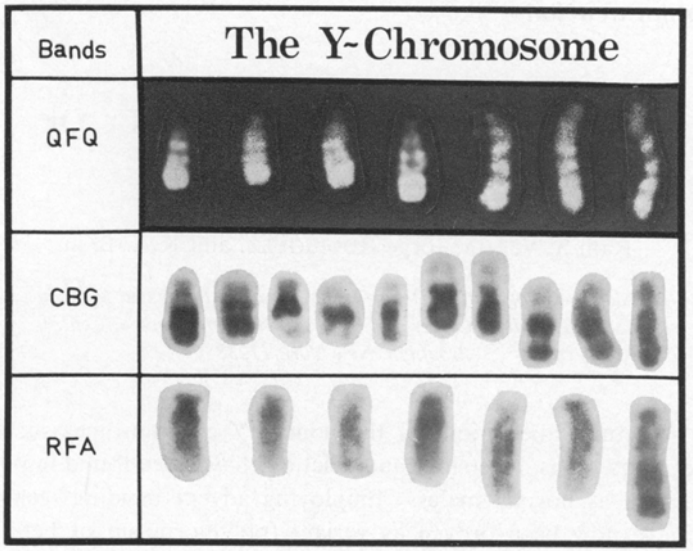

Fig. 1. The Y chromosome as stained by the QFQ, CBG and RFA techniques. Two cells with metacentric Y chromosome seen only by CBG technique. RFA chromosomes were photographed in color but printed in black and white.

segmentation was noted by the CBG technique, no four clear bands can be demonstrated. By RFA technique, the complete long arm was red in color with variable gradation. In general, by the RFA technique, the proximal segment of Yq is yellow green while distal segment is red.

To our knowledge, such a highly unusual Y chromosome with variable morphology has not been seen before. The unusualness is accounted for the fluorescent segment which is presumably genetically inert and therefore, has no clinical significance. There are, so far, two genes that have been localized on the Y chromosome (McKusick, 1980). After critical examination, we found that short arm (Yp) is stable in this physician. This was done by culturing his lymphocytes periodically during two years.

The present case demonstrates that the fluorescent segment does not play any role in phenotypic features in male individuals. However, more work is needed to assign different functions to different regions of the human Y chromosome before any definite genotype-phenotype relations can be established.

REFERENCES

ISCN. 1978. An international system for human cytogenetic nomenclature. In *Birth Defects; Original Article Series*, Vol. 14, No. 8, The National Foundation, New York.

McKusick, V. 1980. The anatomy of the human genome. *J. Hered.* **71**: 370–391.

Sumner, A.T. 1972. A simple technique for demonstrating centric heterochromatin. *Exp. Cell Res.* **75**: 304–306.

Verma, R.S., Rodriguez, J., and Dosik, H. 1982. The clinical significance of pericentric inversion of the human Y chromosome: A rare "third" type of heteromorphism. *J. Hered.* **73**: 236–238.

Verma, R.S. and Fosik, H. 1976. An improved method of photographing human fluorescent chromosomes. *J. Microsc.* **108**: 339–341.

Verma, R.S. and Lubs, H.A. 1975. A simple R-banding technique. *Am. J. Hum. Genet.* **27**: 110–117.

Verma, R.S., Evans-McCalla, M., and Dosik, H. 1982. Human chromosomal heteromorphisms in American blacks. VI. Higher incidence of longer Y owing to non-fluorescent (nf) segment. *J. Med. Genet.* **17**: 297–301.