

A CASE OF PRIMARY AMENORRHEA ASSOCIATED WITH X-AUTOSOMAL
TRANSLOCATION [46,X,t(Xq-;5q+)]

To the Editor: A recent article by Sarto et al. [1] brings to mind an early paper on this subject by Mann et al. [2]. This was the case of an 18-year-old school girl with primary amenorrhea and a translocation involving chromosomes of groups B and C. She was sex-chromatin positive and autoradiography disclosed that a normal X chromosome was consistently late replicating.

Recently one of us (J. H.) has reinvestigated this case utilizing Giemsa banding and fluorescent microscopic techniques. The B-group chromosome was identified as a no. 5 and the C-group chromosome involved in the translocation proved to be the X. Colposcopy demonstrated that the patient had streak gonads and an infantile uterus.

To our knowledge, this was the first X-autosomal translocation to be reported in man. The original title of the article reflected the belief that this was indeed an X-autosomal translocation, but we were forced to change the title in order to get the article accepted.

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REFERENCES

1. SARTO GE, THERMAN E, PATAU K: X inactivation in man: a woman with t(Xq-; 12q+). *Am J Hum Genet* 25:262-270, 1973
2. MANN JD, VALDMANIS A, CAPPs SC, PUIITE RH: A case of primary amenorrhea with a translocation involving chromosomes of groups B and C. *Am J Hum Genet* 17:377-383, 1965