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Original investigations

Y;autosome translocations and mosaicism in the aetiology of 45,X maleness: assignment of fertility factor to distal Yq11

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Summary. Three 45,X males have been studied with Y-DNA probes by Southern blotting and in situ hybridization. Southern blotting studies with a panel of mapped Y-DNA probes showed that in all three individuals contiguous portions of the Y chromosome including all of the short arm, the centromere, and part of the euchromatic portion of the long arm were present. The breakpoint was different in each case. The individual with the largest portion (intervals 1–6) is a fertile male belonging to a family in which the translocation is inherited in four generations. The second adult patient, who has intervals 1–5, is an azoospermic, sterile male. These phenotypic findings suggest the existence of a gene involved in spermatogenesis in interval 6 in distal Yq11. The third case, a boy with penoscrotal hypospadias, has intervals 1–4B. In situ hybridization with the pseudoautosomal probe pDP230 and the Y chromosome specific probe pDP105 showed that Y-derived DNA was translocated onto the short arm of a chromosome 15, 14, and 14, respectively. One of the patients was a mosaic for the 14p+ translocation chromosome. Our data and those reported by others suggest the following conclusions based on molecular studies in eight 45,X males: The predominant aetiological factor is Y;autosome translocation observed in seven of the eight cases. As the remaining case was a low-grade mosaic involving a normal Y chromosome, the maleness in all cases was due to the effect of the testis determining factor, *TDF*. There is preferential involvement of the short arm of an acrocentric chromosome (five out of seven translocations) but other autosomal regions can also be involved. The reason why one of the derivative translocation chromosomes becomes lost may be that it has no centromere.

Introduction

Maleness associated with a 45,X karyotype is a rare condition in which the individuals have male external genitalia and testes. Sexual development is often abnormal or delayed; most patients are azoospermic and many have other abnormalities. In the last 2 years the application of molecular techniques has shown that two alternative mechanisms may give rise to this condition. Firstly, translocation of Y chromosomal DNA onto an autosome has been observed in four patients (Maserati et al. 1986; Disteche et al. 1986; Gal et al. 1987; Magenis et al. 1987). The presence of Y DNA in the patient described by Gal et al. (1987) had been signalled in an earlier paper on the same patient (Schempp et al. 1985). Secondly, low-grade mosaicism involving an intact Y chromosome was observed in one patient (de la Chapelle et al. 1986). Eight additional non-mosaic 45,X male patients were described in the literature before molecular studies became possible (reviewed by Gal et al. 1987). Based on cytogenetic observations, several authors had suggested that the probands carried Y chromosome material translocated onto an autosome (Subrt and Blehova 1974; Vignetti et al. 1977; Koo et al. 1977; Turleau et al. 1980; Schempp et al. 1985).

It thus appears possible that most 45,X males are the result of Y;autosome translocations, as recently suggested (Fraccaro et al. 1987). Since one of the derivative translocation chromosomes is lost, the variable clinical phenotypes may depend on what Y chromosome material is lost, and on the origin and amount of autosomal material lost. To elucidate these questions we undertook a cytogenetic and molecular reinvestigation of three previously studied 45,X males. Our results confirm the earlier findings of Subrt and Blehova (1974) and Turleau et al. (1980) based on cytogenetic observations, that the two 45,X males have Y;autosome translocations involving the short arm of an acrocentric chromosome. In addition one case, previously reported not to have Y-DNA (de la Chapelle et al. 1986, case 2) also has a Y;autosome translocation but is a mosaic for it. Our data provide an explanation for the loss of one of the derivative chromosomes. Finally we report phenotypic findings indicating that a Y chromosomal gene related to fertility (Tiepolo and Zuffardi 1976) is located in interval 6 on Yq11.

Materials and methods

Patients

Case 1 is a phenotypically normal male belonging to a family described in detail by Subrt and Blehova (1974). He is the grandfather of a boy with multiple congenital abnormalities. An identical chromosome abnormality was found in the boy, his normal father, grandfather (our case 1) and great-grandfather. All four males have 45 chromosomes and no Y chromosome. Extra material on one 15p was originally interpreted as representing part of Yp.

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Table 1. Southern hybridization of Y-DNA probes on lymphocyte DNA

Interval[a]	DNA probe used				Presence (+) or absence (−) of Y-specific restriction fragments						
	Probe/locus	Enzyme	Stringency[b]	Reference	Case 1	Case 2	Case 3	Father of case 3	Mother of case 3	Normal males	Normal females
1	pDP132	*Taq*I	H	D.C. Page, unpublished	+	+	+	+	−	+	−
2	pDP61	*Taq*I	H	D.C. Page, unpublished[c]	+	+	+	+	−	+	−
3	50f2/A,B	*Eco*RI	M	Guellaen et al. 1984	+	+	+	+	−	+	−
3	pDP105/A	*Taq*I	M	D.C. Page, unpublished	+	+	+	+	−	+	−
4A	pDP34	*Taq*I	H	Page et al. 1984	+	+	+	+	−	+	−
4B	pDP97	*Eco*RI	H	D.C. Page, unpublished[d]	+	+	+	+	−	+	−
4B	50f2/D	*Eco*RI	M	Guellaen et al. 1984	+	+	+	+	−	+	−
5	12f	*Taq*I	H	Bishop et al. 1984	+	+	−	+	−	+	−
6	50f2/C,E	*Eco*RI	M	Guellaen et al. 1984	+	−	−	+	−	+	−
6	pDP105/B	*Taq*I	M	D.C. Page, unpublished	+	−	−	+	−	+	−
7	pY431-HinfA	*Taq*I	M	K. Smith, unpublished	−	−	−	+	−	+	−

[a] Vergnaud et al. 1986; Page 1986
[b] H, High stringency (hybridization at 47°C, wash at 65°C); M, medium stringency (hybridization at 42°C, wash at 55°C)
[c] Probe derived from plasmid 115 (Geldwerth et al. 1985)
[d] Probe derived from cosmid Y97 (Wolfe et al. 1985)

Case 2 is an azoospermic male described in detail by Turleau et al. (1980). The parents were cytogenetically normal. Based on extensive cytogenetic analysis Turleau et al. (1980) concluded that euchromatic Y chromosome material was translocated onto one chromosome 14p in the patient.

Case 3 is a 14-year-old boy who was born with penoscrotal hypospadias (de la Chapelle et al. 1986, case 2). His psychomotor development is slightly retarded. Lymphocytes and fibroblasts from this patient were extensively studied cytogenetically, and the karyotype was judged to be 45,X without evidence of translocation. Moreover, molecular studies involving Southern blotting with several single copy and two repetitive Y chromosomal DNA probes produced no evidence of Y chromosome derived DNA (de la Chapelle et al. 1986).

Southern blotting

DNA was extracted from blood lymphocytes, lymphoblastoid cells or cultured skin fibroblast cells by published methods (Kunkel et al. 1977). Restriction enzyme digestion, electrophoresis, transfer and hybridization of DNA were performed as previously described (Page and de la Chapelle 1984).

In situ hybridization

The metaphases for in situ hybridization experiments were obtained from phytohaemagglutinin-stimulated 3-day cultures of whole blood or from Epstein Barr virus transformed lymphoblastoid cell cultures. In situ hybridizations with ^{3}H-labelled probes were carried out as described previously (Andersson et al. 1986). Probes were labelled to specific activities of 1.5×10^7 cpm/μg (pDP230) and 9×10^6 cpm/μg (pDP105) and used at concentrations of 20–50 ng/ml. Slides were developed after 4–16 days of exposure and metaphases were stained with 0.25% Wright's stain and photographed. To obtain better banding of chromosomes the slides were destained by immersing them in a series consisting of 95% ethanol, then 95% ethanol, 1% HCl, and finally methanol. They were then treated with 0.03% trypsin in 0.012% EDTA-Hanks solution for 2–4 min (Popescu et al. 1985) and restained for G-bands with Wright's stain.

DNA probes

DNA probes detecting Y-specific restriction fragments used in Southern blotting experiments are listed in Table 1. Probe pDP230 (DXYS20) (Page et al. 1987b) detects homologous pseudoautosomal sequences on Yp and Xp in normal human males. Probe pDP105 (DYZ4) (D.C. Page, unpublished) defines multiple Y-specific loci, one on Yp (interval 3) and one on Yq (interval 6) in normal males.

Results

Cytogenetic studies

As described in detail previously, extra chromosome material occurred on one 15p in case 1 (Subrt and Blehova 1974) and on one 14p in patient 2 (Turleau et al. 1980). Re-investigation

of patient 3 yielded results as follows: All mitoses from recent phytohaemagglutinin-stimulated blood cultures and a recently established lymphoblastoid cell culture had an elongated short arm on one chromosome 14. The same chromosome was also seen when the slides from blood cultures done in 1975 and 1979 were re-investigated. It did not occur in mitoses from the father, mother or brother. In a fresh fibroblast culture the 14p+ chromosome was observed in 4 mitoses out of 29 (14%); while in the remaining 25 mitoses there were 2 chromosomes 14 without any p+ phenomenon. Fibroblast mitoses prepared in October 1982 were re-studied. These came from the same culture that was used to produce DNA for the Southern blotting experiments reported previously (de la Chapelle et al. 1986). Re-analysis of 38 mitoses indicated that 14p+ was present in 11 (29%) and absent in 27 mitoses. Examples of lymphocyte derived chromosomes belonging to the affected pair in the three patients are shown in Fig. 1.

Southern hybridization

Southern blotting experiments using lymphocyte or lymphoblast DNA from the 45,X males were done with a panel of probes detecting different intervals on the normal Y chromosome (Vergnaud et al. 1986; Page 1986). The results are shown in Table 1. All three individuals had single-copy Y-DNA from the euchromatic part of the chromosome but lacked the repetitive DNA, *DYZ2*, that characterizes the heterochromatic portion (interval 7). The amount of Y chromosomal material present was different in the three males. Individual 1 had the largest portion (intervals 1–6), patient 2 had intervals 1–5, and patient 3 intervals 1–4B. These findings are in good agreement with the cytogenetic observations (Fig. 1) that individual 1 appeared to have the largest, and patient 3 the smallest, addition of chromosomal material on the short arm of the affected autosome.

In patient 3 our previous study had failed to detect any Y-DNA (de la Chapelle et al. 1986). In the previous experiments only DNA from fibroblasts had been used, while the findings obtained recently and shown in Table 1 were on lymphoblastoid cell DNA. We therefore repeated the experiments using fibroblast DNA from harvests made in 1982 and from a new fibroblast culture started in 1986. In all these samples the probes from intervals 1–4B did hybridize with the patient's DNA but the hybridization signal was extremely weak, no more than one-tenth that found in lymphoblast DNA. This explains why the signal was not detected in our previous study.

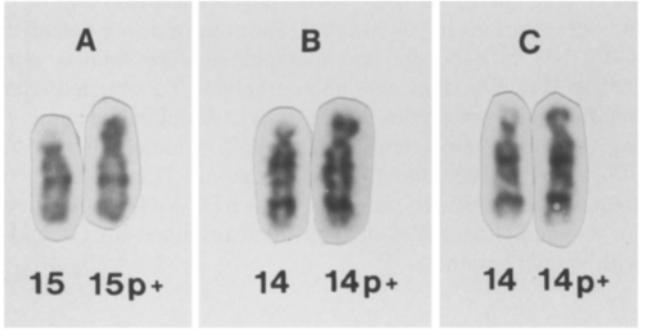


Fig. 1A–C. G-banded translocation chromosomes from the lymphocytes of the three 45,X males. **A** Case 1, **B** case 2, **C** case 3

In situ hybridization

The results obtained with the Y-specific probe pDP105 and the pseudoautosomal probe pDP230 in lymphocyte or lymphoblastoid metaphases are shown in Table 2 and Fig. 2. The short arms of the translocation chromosomes were heavily labelled with both probes in all three individuals. The pseudoautosomal probe pDP230 also showed a peak on Xp in all experiments. These findings suggest that the extra material in the acrocentric translocation chromosome is derived from the Y chromosome in all three 45,X males.

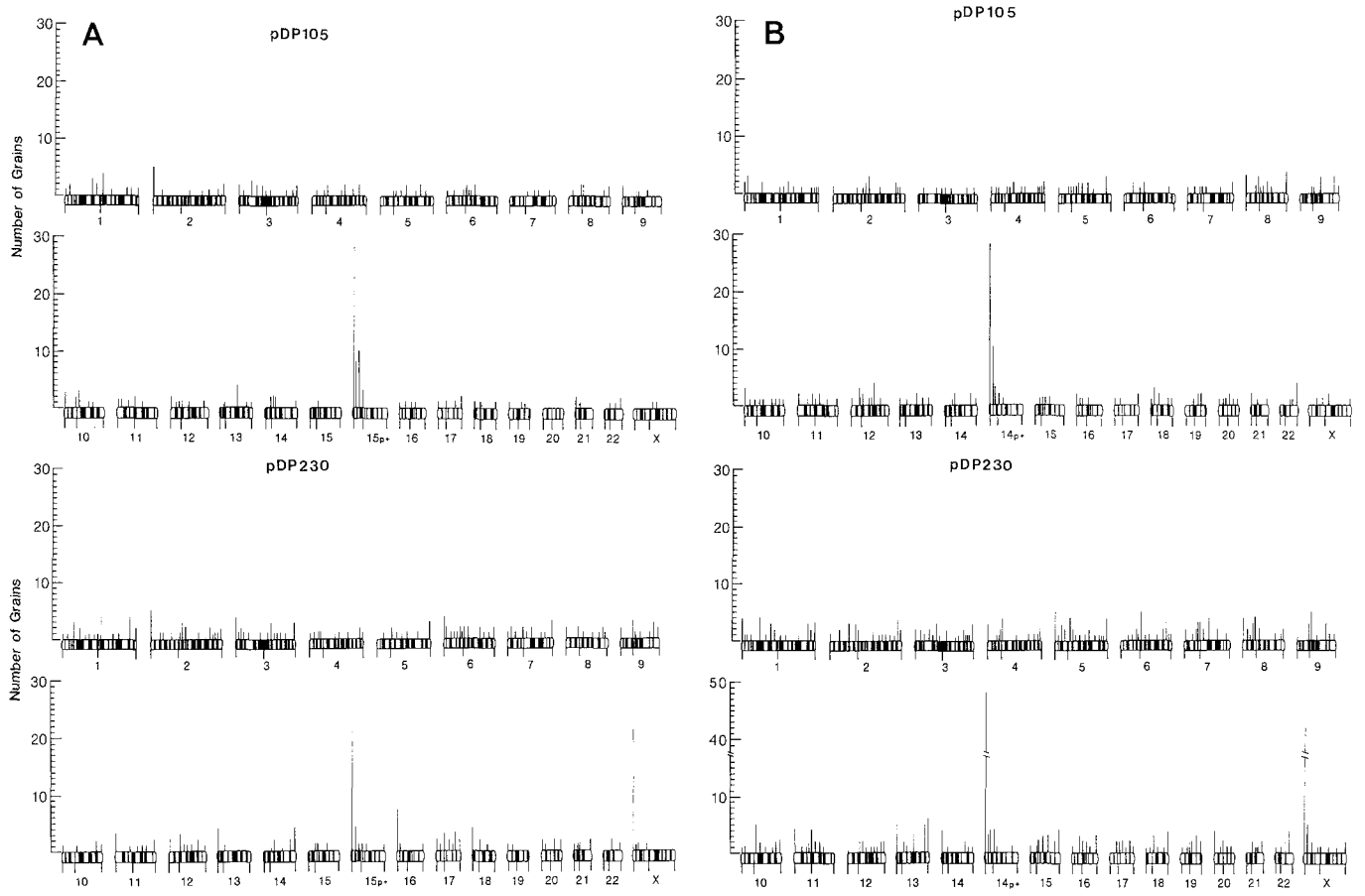
Discussion

Aetiology of maleness in 45,X males

In addition to the three individuals we describe, Y;autosome translocation has been demonstrated by DNA studies in four other cases (Maserati et al. 1986; Disteche et al. 1986; Gal et al. 1987; Magenis et al. 1987). In a few additional patients reviewed by Gal et al. (1987) and Fraccaro et al. (1987) Y;autosome translocation may be present even if unsuspected on cytogenetic grounds. Thus Y;autosome translocation is the most common cause of 45,X maleness. In comparison, similar studies in numerous XX males have not disclosed any case of Y;autosome translocation. Instead, X-Y interchange is the predominant cause of XX maleness (Guellaen et al. 1984; de la Chapelle et al. 1984; Page et al. 1985; 1987a; Affara et al. 1986; Vergnaud et al. 1986; Andersson et al. 1986; Petit et al. 1987; Magenis et al. 1987). However, in both conditions maleness is obviously caused by the presence of *TDF*, which resides in interval 1 on the tip of the short arm of the Y chromo-

Table 2. Grain counts on the metaphases from the three 45,X males after in situ hybridization with probes pDP105 and pDP230

Individual	Probe	No. of mitoses	No. of mitoses with grains on chromosomes		No. of grains on chromosomes		
			14p+(%)	15p+(%)	14p+(%)	15p+(%)	Total
1	pDP105	34		32 (94)		49 (24)	204
	pDP230	55		30 (55)		34 (12)	288
2	pDP105	40	36 (90)		44 (18)		243
	pDP230	58	46 (79)		61 (13)		469
3	pDP105	31	28 (90)		40 (16)		251
	pDP230	49	39 (80)		48 (12)		395



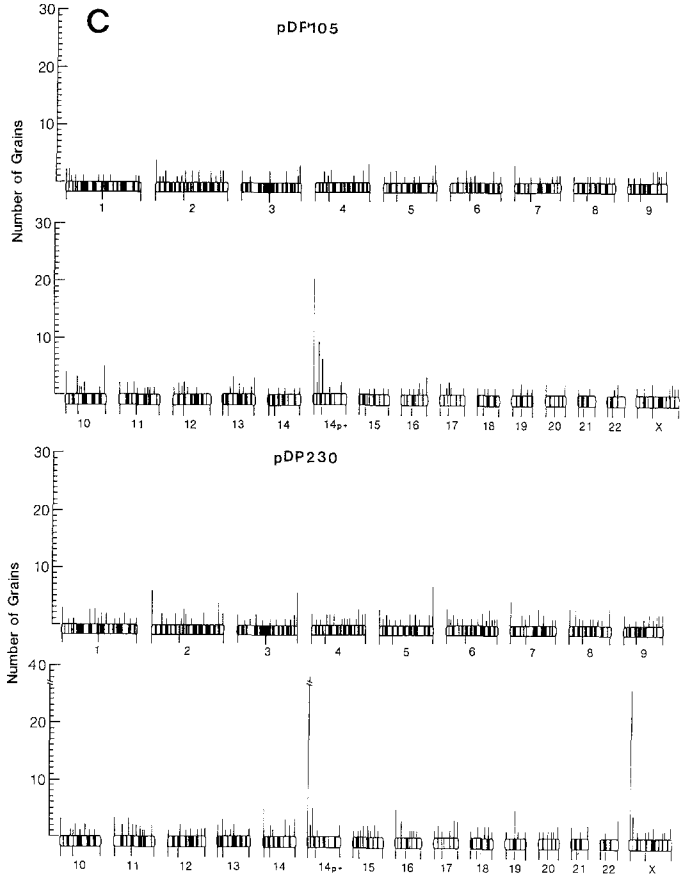


Fig. 2A–C. Histograms showing the localization of grains after in situ hybridization with probes pDP105 and pDP230. **A** Case 1, **B** case 2, **C** case 3

some, a region that is invariably present in the genomes of those XX or X males who have Y-DNA (Vergnaud et al. 1986; Petit et al. 1987; Page et al. 1987a; this report).

That X-Y interchange is not likely to account for 45,X maleness is suggested by the fact that the single X was maternal in all four patients where the parental origin has been determined (present patient 3; de la Chapelle et al. 1986, case 1; Maserati et al. 1986; Gal et al. 1987).

Another aetiological factor in 45,X males is mosaicism involving an intact or structurally abnormal Y chromosome. Molecular and cytogenetic studies of one such patient (de la Chapelle et al. 1986, case 1) disclosed the extremely circumscribed nature of the clone containing the Y chromosome. Indeed, 45,X/46,XY mosaicism with quite variable phenotype is not infrequent (Bühler 1980; Davis 1981; Magenis and Donlon 1982). In the strict sense patients in whom even very few cells with a Y chromosome are detected can no longer be considered to be 45,X males. However, the proven existence of patients with such low-grade mosaicism in which the Y chromosome is barely detectable, as in a case of ours (de la Chapelle et al. 1986, case 1) makes it likely that other patients who are mosaics will remain undetected.

Breakpoints in the translocation chromosomes; loss of one derivative chromosome

The small size and lack of distinct bands of the Y chromosome make it difficult to characterize accurately by cytogenetic methods. Thanks to the molecular map of the Y (Vergnaud et al. 1986; Page 1986) it was possible to ascertain the breakpoints in our patients with the resolution afforded by the probes we used. Our results show that in all three individuals the translocation comprises a contiguous portion of the Y chromosome ranging from the tip of the short arm (characterized by the pseudoautosomal DNA probe pDP230) to the long arm. The centromere is contained in interval 4B represented by probe pDP97 and fragment D recognized by probe 50f2. Since these were present in all three males we conclude that the breakpoint must have been in Yq. However, the breakpoint occurred at a different site in each patient (Table 1).

We have not studied the breakpoints in the autosomes involved by DNA methods. However, we hypothesize that the derivative (14p+ and 15p+) chromosomes in our cases have the centromere of the autosome and thus have two centromeres. This would explain why the other derivative chromosome presumably formed at the translocation is eliminated; it lacks a centromere. We conclude that the likely basis for the loss of one chromosome in 45,X males is the fact that the translocation leads to the formation of one dicentric chromosome, which is retained, and one acentric chromosome, which is lost.

Phenotypic correlations; mapping of the fertility factor

The most severely affected of our patients (case 3 with penoscrotal hypospadias and psychomotor retardation) has the smallest portion of the Y chromosome (intervals 1–4B), while patient 2 who is azoospermic but otherwise normal has intervals 1–5. Case 1 has intervals 1–6 and is clinically normal and fertile; moreover, a study of his pedigree (Subrt and Blehova, 1974) shows that his normal son and father also were 45,X,15p+. In contrast, his grandson, who has the same karyotype, has multiple congenital abnormalities, the aetiology of which may be unrelated to the Y;15 translocation.

These findings argue that the Y chromosomal gene related to fertility (*AZF* for azoospermia factor; Tiepolo and Zuffardi 1976) is located in interval 6 on Yq. We base this suggestion on the fact that case 1 has intervals 1–6 and is fertile while case 2 has intervals 1–5 and is azoospermic. A similar portion of the Y as in our patient 2 occurred in the azoospermic patients described by Disteche et al. (1986) and by Gal et al. (1987) lending further support to this hypothesis. In addition, among XYq− males (Vergnaud et al. 1986) case 26 who had intervals 1–6 of the Y chromosome was fertile while patients who had intervals 1–4B (cases 22–24) and 1–5 (case 25) were sterile.

The family of case 1 in which 45,X males are fertile is of great interest. In this family an alternative mode of sex determination might be said to operate, in which the autosomes 15 have become sex chromosomes. This kind of sex determination operates in some species of monkeys where females are XX, while males are X with a probable Y;autosome translocation (Ma et al. 1980). Meiotic studies in the family of case 1 might give important clues as to the meiotic behavior of the chromosomes involved.

Loss of the short arm of one or several acrocentric chromosomes can occur without any phenotypic consequences (e.g. Nielsen and Rasmussen 1976; Gahmberg et al. 1980). To date no genes apart from those encoding ribosomal RNA have been assigned to these chromosome arms (Cox and Gedde-Dahl 1985) which display heterochromatic characteristics and are markedly polymorphic (ISCN 1985). Involvement of an acrocentric chromosome has now been found in five of the seven 45,X males studied by molecular methods. However, in two cases, a non-acrocentric chromosome was involved (Maserati et al. 1986; Magenis et al. 1987). The resulting loss of euchromatic chromosome material in these instances led to phenotypic abnormalities that could be predicted from what is known about such deletions (18p− and 5p−) when they occur without a translocation. Findings in three further patients in whom a Y;5p translocation chromosome is present but who have not been studied by molecular methods (Vignetti et al. 1977; Tolksdorf et al. 1980; Seidel et al. 1981) corroborate this conclusion.

There seems to be preferential involvement of certain portions of certain autosomes in the translocations of 45,X males. We propose that it is not by coincidence that in so many 45,X males in whom a Y; autosome translocation has been found or suspected, the autosomal point of translocation is in the short arm of an acrocentric chromosome. Non-random association of acrocentric chromosomes with the sex pair occurs in male meiosis (Stahl et al. 1984) and could be a causative factor.

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