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Specific chromosomal satellite association among infertile male patients

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It has been observed that certain chromosomes tend to occupy positions in the mitotic cells close to each other.[1] The most obviously associated chromosomes are the acrocentric chromosomes, which in human karyotype involve chromosome numbers 13, 14, 15, 21, and 22. Accordingly, this phenomenon has been designed as satellite association (SA) or acrocentric association (AA). Indeed, a large body of data is now accumulating to suggest the involvement of SA in the occurrence of chromosomal non-disjunction[2] and Robertsonian translocation.[3] Furthermore, it appears that D/D translocation (13/14, 13/15 and others) is not associated with phenotypic abnormality except for a possible association with male infertility.[4]

Based on the above data together with our previous finding, which suggested a high frequency of SA in metaphases of infertile male patients,[5] we were pushed to carry out more extensive work to assign the most frequent associated chromosomes that might predispose the chromosomes to centric fusion and might have indirect effect on spermatogenesis and to conclude as well, whether those association occur at random or non-random pattern. During the 2-year period starting from October 1998 to October 2001, a total of 75 infertile male patients were subjected to the present investigation. One hundred normal controls were included and investigated in parallel. Culture media, chromosomes, cytology, satellite association scoring and G-banding techniques were conducted according to standard methods. The present investigation revealed a high frequency of association between chromosomes numbers 13-14 among the oligospermia patients compared to normal control. The chi-square analysis showed the association is highly significant (**Table 1**). Again, the 2x2 association particularly between chromosome numbers 13-13, 13-14, 13-15 and 13-21 were significant (X^2=303.77, p>0.005) (**Table 2**). No such observation has been noticed among the azoospermia patients. A high frequency of association was observed between chromosome number 13-14 among the azoospermia though it is non significant (**Table 1**).

Based on the above data, the present investigation showed that the association tendency among the oligospermic groups is non-random, specially when chromosome numbers 13 and 14 are involved. Indeed, the result of Mattei et al[6] also suggested that the unequal frequency observed in the

Table 1 - Number of association for each acrocentric chromosome in metaphase among the oligospermia patients.

Patient n	Association scored for each pair acrocentric per 25 cells					Association /cell
	13	15	14	21	22	
1	10	9	7	10	5	1.64
2	12	10	8	7	8	1.80
3	10	9	10	5	5	1.56
4	11	8	7	0	7	1.32
5	8	7	6	10	4	1.4
6	8	6	2	0	3	0.76
7	9	7	10	6	0	1.28
8	4	4	3	2	2	2.12
9	9	7	10	7	6	1.56
10	13	11	5	10	17	2.24
11	5	4	11	3	3	1.04
12	14	10	5	13	5	1.88
13	15	14	14	3	16	2.48
14	16	14	4	12	0	1.84
15	18	14	7	8	6	2.12
16	13	16	0	8	4	1.64
17	18	14	12	11	8	2.52
18	20	15	14	13	9	2.84
19	12	12	6	6	13	1.96
20	16	10	6	9	7	1.96
21	11	16	11	12	13	2.12
22	16	20	13	12	11	2.84
23	14	12	10	3	5	1.76
24	9	8	11	10	10	1.76
25	15	12	10	6	7	1.88
26	15	14	12	0	12	2.12
27	12	11	10	8	9	2
28	10	9	12	13	12	2.24
29	9	4	6	4	7	1.2
30	18	9	12	8	7	2.16
31	11	10	18	11	8	2.32
32	19	13	10	19	0	1.88
33	13	11	1	10	15	2
34	15	14	10	8	7	2.16
35	16	13	9	8	7	1.88
Total	**444**	**367**	**302**	**275**	**258**	**1.894**

$\chi^2 = 268.84$

Table 2 - Distribution of 2x2 association chromosomes among patient with oligospemia.

Patient n	Association chromosomes per 25 cell														
	13-13	13-14	13-15	13-21	13-22	14-14	14-15	14-21	14-22	15-15	15-21	15-22	21-21	21-22	22-22
1	2	6	2	0	3	4	5	4	0	2	1	3	2	4	3
2	4	7	3	6	2	3	4	5	3	2	1	1	1	2	1
3	3	5	2	3	3	4	3	3	1	1	2	3	0	3	3
4	1	4	3	4	4	2	2	1	2	0	3	2	0	2	3
5	2	5	4	4	2	3	3	4	3	1	0	2	0	1	1
6	3	4	2	0	0	0	2	0	1	1	0	0	0	2	4
7	3	5	2	7	3	1	3	4	1	0	2	1	0	0	0
8	7	8	7	4	2	5	2	2	3	0	3	2	3	3	2
9	0	8	0	0	2	3	5	6	4	0	3	1	2	4	1
10	8	6	5	9	4	4	3	4	4	0	2	2	2	3	0
11	6	1	7	5	1	0	1	3	0	1	0	0	0	0	1
12	0	8	5	0	8	1	5	4	3	2	2	4	0	3	2
13	14	7	0	0	8	3	4	5	4	3	3	3	2	4	2
14	5	8	0	0	0	2	4	4	5	2	4	4	3	3	2
15	0	9	9	0	0	4	7	6	6	0	0	6	2	4	0
16	4	10	0	0	1	3	3	5	4	2	3	2	1	2	1
17	9	6	7	0	8	4	2	3	5	2	3	4	4	2	4
18	5	4	9	8	8	6	5	4	6	2	3	2	2	4	2
19	0	7	9	8	0	2	3	4	4	1	4	2	1	2	3
20	8	7	0	0	0	4	3	3	5	3	3	5	2	3	2
21	0	8	6	2	7	2	1	4	3	3	4	3	3	5	5
22	8	6	10	6	6	3	2	5	2	2	1	3	4	4	7
23	0	9	0	0	0	1	3	6	4	3	4	1	5	4	4
24	4	5	4	3	1	2	2	4	3	4	2	4	2	3	3
25	3	3	3	2	5	1	4	4	1	3	4	2	4	4	3
26	1	5	4	4	4	3	2	5	4	4	5	3	3	5	3
27	3	4	2	5	6	2	1	6	0	0	4	5	5	4	2
28	5	6	6	2	2	2	6	6	3	2	2	4	4	3	3
29	0	7	0	0	1	1	5	5	1	1	2	3	2	1	1
30	4	6	7	7	2	2	0	1	5	4	1	5	4	2	4
31	9	7	7	0	2	1	3	4	3	4	0	4	6	3	5
32	0	8	3	0	3	4	3	4	2	5	3	3	3	3	3
33	4	4	4	2	5	3	2	5	1	4	3	3	5	4	1
34	5	6	5	3	4	2	4	5	2	5	1	1	4	3	4
35	3	7	5	1	3	1	3	1	1	5	4	2	4	4	3
Total	**133**	**216**	**142**	**95**	**110**	**81**	**121**	**139**	**99**	**74**	**80**	**95**	**85**	**103**	**88**

p<0.005, 13-14 and 13-13 (X^2=56.3), 13-14 and 13-15 (X^2=55.70), 13-14 and 13-22 (X^2=53.62), total X2=303.77, degrees of freedom = 136

distribution of Robertsonian translocation constituted an argument, supporting the view that association between acrocentric chromosomes did not occur at random. It is also suggested that the close proximity of the short arms of specific D-group and specific G-group could explain the occurrence of exchange between them, leading to Robertsonian translocation. It is well established that there are 2 factors considered to play a major role in acrocentric chromosome association. Those factors are the presence of satellite deoxyribonucleic acid in the short arm of those chromosomes and the nucleolar organizer regions activity. Indeed, the non-random distribution of Robertsonians translocation can be explained by the meiotic distribution of acrocentric association. Furthermore, the tendency for specific acrocentric chromosome to be in Robertsonian translocation could result from the homology at molecular level.

In conclusion, it seems that the non-random tendency of acrocentric chromosome is evident particularly between chromosome numbers 13-14 among the oligospermic. This tendency could be considered as predisposing factor to Robertsonian translocation. Our finding is supported by previous work, which revealed that, approximately 4% of the oligospermic patients have 45,XY,t(13,14) karyotypes.[5]

Received 1st February 2004. Accepted for publication in final form 24th May 2004.

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