Sex Chromatin Survey in Mentally Handicapped Children in Mexico

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Sex chromatin studies in institutions for mentally handicapped children show a considerably higher frequency of abnormalities as compared with observations on the general population. The published material is summarized in Table I.

The present research was undertaken in order to determine the frequency of possible X chromosome numerical aberrations, by means of a sex chromatin study, in a group of mentally handicapped children attending specialized teaching institutions in Mexico City.

Material and Methods

One thousand mentally handicapped children (623 males and 377 females), 6 to 14 years of age, with an IQ of 50 to 80, were included in the survey.

Sex chromatin was determined in epithelial cells from oral mucosa. Slides were stained by the Feulgen reaction and counterstained with Fast green. Cases with 10% or more Barr bodies were considered to be positive and to have two X chromosomes, while those with less than 5% were considered to be negative and to have either XY or XO sex chromosome complements. A total of 200 nuclei was studied in every case, and the counts were checked by two observers in each instance.

### TABLE I

<table>
<thead>
<tr>
<th>Source</th>
<th>Males No. Tested</th>
<th>Males No. with Abnormal Sex Chromatin†</th>
<th>% Abnormal</th>
<th>Females No. Tested</th>
<th>Females No. with Abnormal Sex Chromatin</th>
<th>% Abnormal</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ferguson-Smith</td>
<td>325</td>
<td>4(4+)</td>
<td>1-23</td>
<td>-</td>
<td>-</td>
<td>-</td>
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<tr>
<td>Prader, Schneider, Züblin, Francés, and Riedi</td>
<td>336</td>
<td>8(8+)</td>
<td>2-38</td>
<td>-</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>Ferguson-Smith</td>
<td>663</td>
<td>8(8+)</td>
<td>1-20</td>
<td>-</td>
<td>-</td>
<td>-</td>
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<tr>
<td>Barr, Shaver, Carr, and Plunkett</td>
<td>1535</td>
<td>14(11+; 3 +)</td>
<td>1-11</td>
<td>-</td>
<td>-</td>
<td>-</td>
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<tr>
<td>Cornwall</td>
<td>409</td>
<td>3(3+)</td>
<td>0-73</td>
<td>-</td>
<td>-</td>
<td>-</td>
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<tr>
<td>De la Chapelle and Hortling</td>
<td>342</td>
<td>3(3+)</td>
<td>0-87</td>
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<td>-</td>
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<tr>
<td>Ferguson-Smith</td>
<td>916</td>
<td>9(7+; 2 +)</td>
<td>1-20</td>
<td>-</td>
<td>-</td>
<td>-</td>
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<tr>
<td>Fraser, Campbell, MacGillivray, Boyd, and Lennox</td>
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<td>-</td>
<td>-</td>
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<tr>
<td>Mosier, Scott, and Cotter</td>
<td>1252</td>
<td>10(10+)</td>
<td>0-81</td>
<td>-</td>
<td>-</td>
<td>-</td>
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<tr>
<td>Shapiro and Ridler</td>
<td>900</td>
<td>6(6+)</td>
<td>0-66</td>
<td>-</td>
<td>-</td>
<td>-</td>
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<tr>
<td>Breakley</td>
<td>157</td>
<td>1(1+)</td>
<td>0-63</td>
<td>140</td>
<td>0</td>
<td>0</td>
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<tr>
<td>Carr, Barr, and Plunkett</td>
<td>40</td>
<td>1(1+)</td>
<td>2-50</td>
<td>-</td>
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<td>-</td>
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<tr>
<td>Gustavson and Åkesson</td>
<td>48</td>
<td>0</td>
<td>0</td>
<td>40</td>
<td>0</td>
<td>0</td>
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<tr>
<td>Israelsohn and Taylor</td>
<td>1556</td>
<td>7(7+)</td>
<td>0-45</td>
<td>-</td>
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<td>Johnstone, Ferguson-Smith, and Handmaker</td>
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<tr>
<td>Sanderson and Stewart</td>
<td>245</td>
<td>2(2+)</td>
<td>0-81</td>
<td>240</td>
<td>2(1+; 1 +)</td>
<td>0-83</td>
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<tr>
<td>Hamerton, Jagiello, and Kirman</td>
<td>229</td>
<td>1(1+)</td>
<td>0-43</td>
<td>196</td>
<td>2(1+; 1 +)</td>
<td>1-00</td>
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<tr>
<td>Maclean et al.</td>
<td>2607</td>
<td>28(23 + ; 4 +; 1 + +)</td>
<td>1-07</td>
<td>1907</td>
<td>9(8 + ; 1 -)</td>
<td>0-52</td>
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<tr>
<td>Forsman and Hambert</td>
<td>760</td>
<td>20(20 +)</td>
<td>2-63</td>
<td>-</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>Breg et al.</td>
<td>835</td>
<td>8(7 + ; 1 + +)</td>
<td>0-96</td>
<td>727</td>
<td>6(5 + ; 1 -)</td>
<td>0-85</td>
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<td>Present study</td>
<td>623</td>
<td>5(4 + ; 1 + +)</td>
<td>0-8</td>
<td>377</td>
<td>2(1-)</td>
<td>0-26</td>
</tr>
</tbody>
</table>

* Quoted by Israelsohn and Taylor.
† (+ + ; + + + ;+) single, double, triple or negative sex chromatin. + I isochromosome X.
‡ Some mosaic forms.
Chromosome studies were carried out in 4 cases with discrepant sex chromatin, according to a slight modification of the method of Moorhead, Nowell, Mellman, Battips, and Hungerford (1960) for culture of lymphocytes from peripheral blood. In one case, skin culture was set up according to the method of Hsu and Kellogg (1960).

Results

Of the 623 males studied, 5 showed a discrepancy between the phenotype and sex chromatin finding. Four of these were chromatin positive with single masses, while one had double sex chromatin (0·8%). Of the 377 females, one was chromatin negative (0·27%).

Chromosome studies on cultured lymphocytes from the sex chromatin positive boys revealed a 47, XXY karyotype in two instances: while another positive boy had a 48, XXXY chromosome constitution both in lymphocyte and fibroblast cultures. The sex chromatin negative girl had a 45, X chromosome constitution obtained from a single blood culture. Chromosome studies were not carried out on the remaining two positive boys (Fig. 1, 2, 3) (see Table II).
Fig. 2. (a) Metaphase of a case with 48, XXYY chromosomal constitution. (b) Karyotype of the same cell.

Discussion

Published reports have shown that there is little variation in the type of sex chromatin abnormalities found among mentally handicapped children (Table I), and it is interesting to note that the majority of female defectives had two sex chromatin bodies rather than none. In the present series, however, no females with double sex chromatin were observed.

Two instances of possible XO subjects were described by Maclean, Mitchell, Harnden, Williams, Jacobs, Buckton, Baikie, Court Brown, McBride, Strong, Close, and Jones (1962), and Breg, Castilla, Miller, and Cornwell (1963), and a further example has been recorded in the present study.

Chromosome studies on four of the sex chromatin positive boys in the present series revealed two to have XXY and one an XXYY sex chromosome
complement; the boy with double sex chromatin probably had an XXXY sex chromosome constitution.

The findings in the present study, on a sample of mentally handicapped children from a heterogeneous Mexican population, thus show no significant difference either in type or frequency of chromosome abnormalities as compared with other series from several different countries. The finding of a single XO girl and the failure to find any XXX females is perhaps of interest.

**Summary**

A sex chromatin survey of 1000 mentally handicapped children attending special schools in Mexico City has been carried out. Ages were between 6 and
14 years, and IQs between 50 and 80. Five boys out of 623 had positive sex chromatin, and one of these had two Barr bodies. One girl had negative sex chromatin. The entire group accounted for 0.6% of sex chromatin abnormalities. Chromosome studies in three boys with sex chromatin showed two with 47, XXX and another with 48, XXXXY chromosome complements. The chromatin negative girl had a 45, X sex chromosome constitution.

REFERENCES
Sex chromatin survey in mentally handicapped children in Mexico.
H Márquez-Monter, H Santiago-Payán and S Kofman-Alfaro

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