121. A Chromosome Study on the 751 Cases of the Congenital Defectives and their Relatives

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This paper describes a summary of the direct combination-work undertaken between the clinical doctors and cytogeneticists for the studies of the clinical diagnosis and genetic advices for congenital defectives. During a period from November 1975 to the end of October 1985, 751 cases were collected for chromosomal researches. This article is a product of the collaboration-work with the institutions for the congenital defectives and the Kadotani Medical Research Foundation. The 751 cases under study consisted of 205 cases from the Hiroshima City Welfare Center for the Handicapped Children’s Center,1,2 92 cases from the Kojika-Gakuen,3 45 cases from the Tomoe-Gakuen,4 89 cases from the Kamo National Sanatorium,5 45 cases from the Wakaba-Ryoikuen,6 91 cases from the Roppo-Gakuen, and 184 cases from the Kadotani Medical Research Foundation.

The chromosome studies were performed with the standard blood cultures. The chromosomes were analysed following the conventional Giemsa, the G-, Q-, and C-banding procedures.

Results and remarks. Five hundred and eighty four out of the 751 cases herein concerned were the congenital defectives and the remaining 167 cases were their relatives. Four hundred and sixty seven out of 584 cases with the congenital defects were found to possess the normal karyotypes, leaving 117 cases showing the abnormal karyotypes, being 20.0% in incidence. Among the abnormalities, 11 out of 117 cases were found to be transmitted through the parental line. Out of 117 cases with the chromosome abnormalities, 102 cases showed the autosomal abnormalities, while the remaining 15 cases had the sex-chromosome abnormalities. The data so far obtained are summarized in Table I, and critical details for 35 cases of them are referred to in the literature listed (7–41). One of the chromosome aberrations is shown in Fig. 1. Details of some of these abnormal cases will be published elsewhere in the near future.

Then it is apparent that the chromosome study serves as an important tool for the clinical diagnosis of the congenital defectives and their relatives seeking for genetic advices.

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Table I. Chromosome abnormalities from 117 cases out of 584 cases with the congenital defects

<table>
<thead>
<tr>
<th>Autosomal abnormalities</th>
<th>Sex-chromosome abnormalities</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Deletion and trisomy</strong></td>
<td></td>
</tr>
<tr>
<td>Karyotype</td>
<td>No. of cases</td>
</tr>
<tr>
<td>46, XY, 5p—</td>
<td>1</td>
</tr>
<tr>
<td>46, XY, der(5), rec(5;17) (p13;q23) mat</td>
<td>1</td>
</tr>
<tr>
<td>46, XY, del(5) (q15q31)</td>
<td>1</td>
</tr>
<tr>
<td>46, XY, del(5) (q22q31)</td>
<td>1</td>
</tr>
<tr>
<td>46, XY/47, XY, +8</td>
<td>1</td>
</tr>
<tr>
<td>46, XY, del(9) (p22pter)</td>
<td>1</td>
</tr>
<tr>
<td>46, XY, del(9) (q11q13)</td>
<td>1</td>
</tr>
<tr>
<td>46, XY, del(10) (p14pter)</td>
<td>1</td>
</tr>
<tr>
<td>47, XY, +del(15) (q15qter)</td>
<td>1</td>
</tr>
<tr>
<td>46, XX, del(18) (p1105)</td>
<td>1</td>
</tr>
<tr>
<td>47, XX or XY, +18</td>
<td>4</td>
</tr>
<tr>
<td>47, XX or XY, +21</td>
<td>58* 16</td>
</tr>
<tr>
<td>46, XX/47, XX, +21</td>
<td>1</td>
</tr>
<tr>
<td>47, XY, +22</td>
<td>1</td>
</tr>
<tr>
<td>47, XY, +del(22) (q13qter)</td>
<td>1</td>
</tr>
<tr>
<td>47, XX, +del(22) (q13) mat</td>
<td>1</td>
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<tr>
<td><strong>Total</strong></td>
<td><strong>76</strong></td>
</tr>
</tbody>
</table>

Ref.: References. * Two cases out of 58 cases were sibling from their parents with the normal karyotypes. ** Chromosome formulas of the six cases with inv(9) were 46, XY, inv(9) (p11q13) pat, 46, XY, inv(9) (p13q13) pat, 46, XX, inv(9) (p11q13), 46, XY, inv(9) (p11q13), 46, XX, inv(9) (p13q21), and 46, XX, inv(9) (under study).
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References