42. A Cri-du-Chat Infant Born to a Mother Carrying a 5/17 Balanced Translocation

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Since the initial communication on the short arm deletion of a B chromosome in the cri-du-chat syndrome by Lejeune et al. (1963), many allied and variant chromosome abnormalities have been presented in this syndrome (Makino 1975). Recent familial cytogenetic surveys have provided evidence to show that there are some cri-du-chat infants who were born to parents one of whom is a carrier of B/C, B/D or B/G translocations (De Capoa et al. 1967, Singh et al. 1973, Jackson and Ban 1970), or of an insertional translocation, ins(17; 5) (Berger et al. 1974). The present article describes cytogenetic profiles of a cri-du-chat syndrome baby and his mother who is a carrier of reciprocal translocation between the terminal portion of a short arm of no. 5 and the long arm of no. 17.

Case report. The propositus is an infant (♂) born on July 14, 1976, as the second son between a 31-year-old mother and a 33-year-old father. The first pregnancy in 1972 terminated in a spontaneous abortion at the end of 4-month-pregnancy. The second pregnancy yielded a normal male boy, now 2 years old. The third pregnancy in 1975 resulted in a spontaneous abortion at the 13th week. Both parents are phenotypically normal and not consanguineous, there being no evidence in them for mental deficiency and for exposure to atomic bomb. The father has neither familial disorders nor congenital anomalies in his relatives, while the mother has an elder sister who is a carrier of a possible balanced translocation including a no. 5 chromosome, and has a cri-du-chat syndrome baby. Their chromosomes were investigated by Kadotani et al. (1971), though banding analyses were not performed (see pedigree in Fig. 1).

The propositus showed the birth weight of 2250 g and the height of 43 cm at 37 weeks of gestation. A high pitched cry was noted immediately after birth. The clinical diagnosis of the cri-du-chat syndrome was possible on the basis of this and the other characteristic physical anomalies, such as bilateral epicanthic folds, hypertelorism, antimongoloid slant, broad base of nose, lowset malformed

* Chromosome Research Institute, Hokkaido University, Sapporo.
ears, micrognathia, high-arched palate, flat occiput, short neck, bilateral simian creases, and hypotonia. The heart, lungs and abdominal viscera were apparently normal except for undescended testes.

*Cytogenetic findings.* Peripheral blood cultures for chromosome study were prepared from the propositus, his parents, his elder brother, and his maternal grandmother (Table I). Chromosome analyses were performed according to the conventional Giemsa staining method and the trypsin G-banding technique after Seabright (1971).

The propositus showed the karyotype with 46 chromosomes including a chromosome 5p-, characteristic to the cri-du-chat syndrome. The deleted segment of the 5p- chromosome corresponded to the bands from 5p13 to 5pter (Fig. 2), according to the Paris nomenclature. No other chromosomal abnormalities were noted. The elder brother, the father and the maternal grandmother of the propositus showed no chromosome change, each having apparently a normal karyotype. In contrast, the mother was chromosomally abnormal in having two unusual elements: one was represented by a deletion of the short arm of no. 5 chromosome and the other by the elongated long arm of no. 17 chromosome. She does not have any clinical sign for the cri-du-chat syndrome. The banding technique revealed that the extra chromosomal material on the long arm of no. 17 chromosome was remarkable by showing a banding pattern identical to the terminal segment of the short arm of a normal no. 5 chromosome. Then, it appears that the deleted short arm of no. 5 is translocated to the long arm of no. 17 chromosome (Fig. 3).

![Fig. 1. Pedigree involving the proband.](image-url)

<table>
<thead>
<tr>
<th>Individual</th>
<th>Chromosome counts</th>
<th>No. of cells karyotyped</th>
<th>Karyotype</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>44 45 46 47</td>
<td>Giemsa G-band</td>
<td></td>
</tr>
<tr>
<td>Propositus</td>
<td>2 3 12 0</td>
<td>5 19</td>
<td>46, XY, 5p-</td>
</tr>
<tr>
<td>Mother</td>
<td>1 11 89 1</td>
<td>32 22</td>
<td>46, XX, rep (5; 17) (p13; q23)</td>
</tr>
<tr>
<td>Father</td>
<td>0 0 23 1</td>
<td>6 0</td>
<td>46, XY</td>
</tr>
<tr>
<td>Grandmother</td>
<td>0 0 23 1</td>
<td>15 6</td>
<td>46, XX</td>
</tr>
</tbody>
</table>
Remarks and conclusion. The proband showed the karyotype of 46,XY,5p-. The deleted segment of no. 5 chromosome includes bands p13→pter of the short arm. The feature is consistent with the findings from the recent banding studies showing that the band 5p15 is lacking in all reported cases of the cri-du-chat syndrome (Niebuhr 1972, Berger et al. 1974, Jackson et al. 1975, Gracia et al. 1976). The mother of the proband is most probably a carrier of a balanced translocation involving the terminal part of a short arm of no. 5 and the long arm of no. 17. She had one case of the cri-du-chat syndrome, one normal boy and two abortions. The two abortions could be accounted for by the gametic segregation of the abnormal no. 17 chromosome (17q+mat) resulting in a partially trisomic status for the short arm of no. 5 chromosome, although other types of meiotic segregations including nondisjunctions are conceivable for the zygotic unbalance.

It is of particular interest to note that the maternal aunt of the proband who was phenotypically normal and had a cri-du-chat baby was predicted as a carrier of a possible balanced translocation involving the terminal part of a short arm of no. 5 and the long arm of no. 17. She had one case of the cri-du-chat syndrome, one normal boy and two abortions. The two abortions could be accounted for by the gametic segregation of the abnormal no. 17 chromosome (17q+mat) resulting in a partially trisomic status for the short arm of no. 5 chromosome, although other types of meiotic segregations including nondisjunctions are conceivable for the zygotic unbalance.

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(Kadotani et al. 1971). Although the banding analysis was not carried out in the aunt, her karyotype is assumed to be the same as that of the mother. Since the maternal grandmother was chromosomally normal, it is most likely that the balanced translocation was inherited through the maternal line, probably originated from the maternal grandfather, though he was not available for chromosome study.

It is then concluded that, the propositus possesses the karyotype, 46,XY,der(5),rcp(5;17)(p12;q23)mat characteristic of the cri-du-chat syndrome, and his mother is a carrier of a balanced translocation, 46,XX,rcp(5;17)(p13;q23). The translocation is probably originated from the maternal grandfather.

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References