CASE REPORT

Chromosome Studies in an Infant with Acute Erythremic Myelosis

By W. KROLL AND K. SCHLESINGER

In acute erythremic myelosis (Di Guglielmo disease\(^1\)), only few chromosomal studies have so far been reported.\(^2\) We would like to present a case of Di Guglielmo's disease with chromosomal abnormalities found in the bone marrow in a small child.

CASE REPORT

The 2-3/12-year-old boy (H.R.) was admitted to the University Children's Hospital of Heidelberg in May 1968, because of petechial skin bleedings, anemia, enlarged lymph nodes and hepatosplenomegaly. On admission the hemoglobin was 3.4 Gm. per cent, erythrocytes 1.0 mill/mm\(^3\), reticulocytes 3 per cent, leukocytes 3000/mm\(^3\). The hypocellular bone marrow showed 14.5 per cent granulopoietic cells, four per cent apparently normal erythropoietic cells, 71 per cent nonpathological reticular cells and 10.5 per cent atypical cells. These atypical cells looked undifferentiated in their morphological appearance, almost resembling proerythroblasts. The cytoplasm reacted strongly with basophilic stain; however, the nuclei were relatively small as compared to nuclei of normal proerythroblasts, and the cytoplasm was increased. PAS staining of the atypical cells showed negative results and 85 per cent of them had a markedly increased iron content. The cytoplasm contained vacuoles and some nuclei large nucleoli. The results lead to the interpretation that these atypical cells were pathologically altered erythroblasts.

A remission after treatment with prednisone, vincristine and 6-mercaptopurine only lasted for about 6 weeks. Atypical erythroblasts again appeared in the peripheral blood and predominated in the bone marrow. There was no concomitant increase in myeloblasts (Sept. 3, 1968). From the same aspirate, chromosome analysis was done in direct bone marrow preparation.

The boy succumbed to a final pneumonia on 23 September after four months of illness.

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Direct bone marrow preparations were prepared in adaption to the method of Kiossoglou.\(^6\) Only 22 mitoses could be analyzed. No gaps or chromatid breaks were seen. None of the cells was euploid. In each metaphase there were two marker chromosomes: one A\(1\) chromosome had an elongated long arm (A\(q^+\)) and one extra acrocentric chromosome had the length of a D group chromosome. An additional chromosome, which was possibly an F chromosome, was observed in 11 metaphases. Monosomy of a G chromosome was present in 11 mitoses (Table 1).

Kiossoglou et al.\(^3\) described three cases of acute erythremic myelosis, all of...
Table 1.—Numerical and Structural Chromosome Aberrations Found in 22 Analyzed Metaphases from Bone Marrow

<table>
<thead>
<tr>
<th>Number of Metaphases</th>
<th>Deviation from Normal Karyotype</th>
<th>Karyotypes of Abnormal Cells</th>
</tr>
</thead>
<tbody>
<tr>
<td>22</td>
<td></td>
<td></td>
</tr>
<tr>
<td>6</td>
<td>A 1q+</td>
<td>+1</td>
</tr>
<tr>
<td>1</td>
<td>B</td>
<td>-1</td>
</tr>
<tr>
<td>2</td>
<td>C+X +1</td>
<td>+1</td>
</tr>
<tr>
<td>7</td>
<td>D</td>
<td>+1</td>
</tr>
<tr>
<td>1</td>
<td>E</td>
<td>+1</td>
</tr>
<tr>
<td>4</td>
<td>F +2</td>
<td>-1</td>
</tr>
<tr>
<td>1</td>
<td>G -1</td>
<td></td>
</tr>
</tbody>
</table>
them showing a monosomy G in each cell. Castoldi et al.2 found in one case, a partial deletion of a G chromosome. They discussed the probability of a true relation between the monosomy G or the partial deletion of a G chromosome and the erythremic myelosis. However, there are so far too-few corresponding chromosomal findings to settle this point. Some cases have been described without any chromosomal changes3,7 or with inconstant aneuploidy.5,7 The

Fig. 1.—A. Group 2 with Alq⁺. B. Karyotype with Alq⁺, D⁺, F⁺, from bone marrow.

Fig. 2.—Atypical cells with two or more nuclei, several large nucleoli and vacuoles in cytoplasm.
CHROMOSOME STUDIES IN AN INFANT

report is intended to serve as a further contribution to this topic. When more cases have been cytogenetically examined, it may perhaps become possible to decide if a constant chromosomal abnormality is present in this rare disease.

REFERENCES

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