137. Chromosome Banding Patterns in 27 Cases of Acute Myeloblastic Leukemia*1

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(Communicated by Sajiro Makino, M. J. A., Nov. 12, 1976)

Recent studies with chromosome banding techniques have afforded some evidence of possible non-random karyotypic changes in certain cases of hematological disorders including AML.2),5) We wish to present here karyotypic data on 27 cases of AML in which banding analyses were successfully made.

Materials and methods. The 27 cases here described were selected from 61 cases of AML surveyed in our laboratory during the past 5 years. Out of them 2 unusual cases with the possible Ph1-translocation have been reported elsewhere.8) Air-dried chromosome slides were first examined by conventional Giemsa staining, and then subjected to more detailed analyses with the Q-banding or G-banding methods. The karyotypes were described according to the standard nomenclature by the Paris Conference (1971).

Results. The results of karyotype analyses in the 27 cases were summarized in Table I, together with some clinical and hematological data. There were 11 karyotypically abnormal cases, while the remaining 16 cases did not show any appreciable karyotypic change even with banding analyses. Trisomy 8 was a common abnormality in 2 cases (cases 1 and 2), though the latter case had an additional extra element possibly originated from a no. 22 chromosome. This case had been diagnosed as having a 47, XX, +C karyotype when examined about one year ago on the basis of non-banded Giemsa preparations of bone marrow cells. Another case of trisomy (case 3) was due to an extra no. 12 chromosome (Fig. 1, a). Cases 4 and 5 both had deletions, but of different types, i.e., del(3) and del(9), respectively (Fig. 1, d and c). Cases 6–10 had the same abnormality as represented by a translocation between the long arm of a no. 8 and a no. 21. Three of them (cases 8–10) were accompanied by a missing sex-element of either an X or the Y. The 8/21 translocation in cases 6–9 was observed in almost 100% of the cells analyzed, while case 10 showed a considerable number of cells with a normal karyo-

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*1 Contributions from the Chromosome Research Unit, Faculty of Science, Hokkaido University, Sapporo. Supported by a Grant-in-Aid for Cancer Research from the Ministry of Education, Science and Culture, Japan.
type. The proportion of the normal to abnormal cells was 5:7 in the marrow culture. Case 11 had a translocation between a no. 9 and the Y (Fig. 1, b). The break points estimated by the banding analysis in the above mentioned structurally altered chromosomes are shown in Table I.

Discussion. Although a certain extent of sampling bias is expected in the present series, the common occurrence of an 8/21 translocation in the 5 cases studied deserves special attention. This abnormality which had been described, before the advent of banding techniques, as a 46, −C, +D, +E, −G or a C/G translocation was recently disclosed to be caused by a specific rearrangement, t(8;21) (q22;q22), in a considerable number of AML cases analyzed by banding.1,3,4,6,7,10 A missing sex chromosome was also a common feature in some of the cases having the 8/21 anomaly. Oshimura et al. (1976)3 estimated that about 10% of the AML cases reported had the 8/21 translocation. The incidence in our series was not less than 8.2% (5/61), taking all the material collected into consideration. The frequency of karyotypically abnormal cells in AML may vary from case to case, depending on the stage and course of the disease, with or without therapy, and some technical variables related to whether samples are processed directly or cultured. Whether the 46, XY cells which coexisted with the 45, X, −Y, t(8;21) cells in the marrow culture of case 10 represent leukemic cells is unknown, even though the other case (case 9) having the
### Table I. Karyotypes and some clinical and hematological data of the 27 cases studied

| Case No. | (code)     | Specimen    | Karyotype                              | Age (year) | Survival (week) | WBC ($\times 10^9$) | % Blast in PB | Auer body | Remission |
|----------|------------|-------------|----------------------------------------|------------|-----------------|---------------------|---------------|-----------|-----------|
| 1        | (211, CT)  | PB(c)       | 47, XX, +8                             | 21         | 31              | 12.0                | 75            | ?         | No        |
| 2        | (368, AT)  | BM(c)       | 48, XX, +8, +?22                       | 65         | 55              | 2.1                 | 95            | +         | Complete  |
| 3        | (109, KH)  | PB(c)       | 47, XY, +12                            | 37         | 17              | 10.9                | 81.5          | -         | Partial   |
| 4        | (411, TS)  | BM(c)       | 46, XY, del(3)(pter→q21::)              | 31         | 56              | 9.7                 | 82            | +         | Partial   |
| 5        | (434, MI)  | PB(c)       | 46, XY, del(9)(pter→q22::)              | 44         | >6              | 25.5                | 87.5          | -         | Undefined |
| 6        | (208, SM)  | BM(d, c)    | 46, XX, t(8;21)(q22;q22)                | 28         | 44              | 9.5                 | 53            | +         | No        |
| 7        | (214, IS)  | BM(d), PB(c)| 46, XY, t(8;21)(q22;q22)               | 34         | 1               | 13.9                | 84            | -         | Undefined |
| 8        | (303, II)  | BM(c), PB(c)| 45, X, −X, t(8;21)(q22;q22)            | 24         | 32              | 18.5                | 85            | +         | Complete  |
| 9        | (340, HO)  | BM(c)       | 45, X, −Y, t(8;21)(q22;q22)             | 26         | >8              | 7.0                 | 94.5          | +         | Complete  |
| 10       | (230, ES)  | BM(c)       | 46, X/45, X, −Y, t(8;21)(q22;q22)       | 17         | 65              | 8.9                 | 81            | -         | Complete  |
| 11       | (240, KY)  | PB(c)       | 46, XY, t(9;11)(p11; p11)              | 37         | 32              | 95.6                | 97.5          | -         | Partial   |
| 12       | (304, HK)  | BM(d, c)    | 46, XX                                 | 57         | 16              | 6.3                 | 28            | -         | No        |
| 13       | (315, TI)  | PB(c)       | 46, XX                                 | 69         | 2               | 6.6                 | 38            | -         | No        |
| 14       | (324, YN)  | BM(d), PB(c)| 46, XX                                | 29         | 53              | 5.5                 | 66            | -         | Partial   |
| 15       | (346, JO)  | BM(d)       | 46, XX                                 | 41         | 2               | 6.4                 | 7             | +         | No        |
| 16       | (370, YI)  | BM(c)       | 46, XX                                 | 18         | >65             | 1.7                 | 55.5          | -         | Partial   |
| 17       | (184, KS)  | BM(d)       | 46, XY                                 | 39         | 217             | 8.9                 | 22            | -         | Complete  |
| 18       | (220, NM)  | BM(c), PB(c)| 46, XY                                | 57         | 3               | 10.5                | 93            | ?         | No        |
| 19       | (229, TM)  | BM(c), PB(d)| 46, XY                                | 46         | 11              | 18.3                | 54            | +         | Undefined |
| 20       | (262, MS)  | BM(c), PB(c)| 46, XY                                | 23         | 34              | 101.6               | 94.5          | -         | Partial   |
| 21       | (283, MY)  | BM(d)       | 46, XY                                 | 20         | 8               | 7.0                 | 29            | -         | Partial   |
| 22       | (322, MN)  | PB(c)       | 46, XY                                 | 38         | 28              | 19.3                | 80            | -         | Partial   |
| 23       | (391, NK)  | BM(d)       | 46, XY                                 | 71         | 12              | 10.0                | 57.5          | ?         | Undefined |
| 24       | (392, MW)  | BM(c)       | 46, XY                                 | 27         | 48              | 6.5                 | 72            | +         | Complete  |
| 25       | (394, RA)  | BM(c)       | 46, XY                                 | 51         | 44              | 28.1                | 89.5          | +         | Complete  |
| 26       | (400, SS)  | BM(c)       | 46, XY                                 | 27         | 34              | 2.6                 | 95.5          | +         | Complete  |
| 27       | (415, KH)  | BM(c), PB(c)| 46, XY                                | 72         | >23             | 3.1                 | 5             | -         | Partial   |

BM: bone marrow, PB: peripheral blood, c: cultured, d: direct preparation.
same abnormality did not show any mosaicism in the marrow culture.

Trisomy 8, another common change here encountered, is not considered specific for AML. This abnormality has been reported in some other malignant diseases including hematological disorders.\(^2,5,10\) The additional trisomy for a possible no. 22 chromosome in case 2 which was acquired during the progression of the disease seems interesting since such a secondary change of stemline karyotype is a rather rare event in AML. Trisomy 12, del(3), del(9) and t(9; Y) are hitherto undescribed abnormalities in AML.

Despite several attempts having been made to correlate the karyotypic profiles and clinical features in AML, the results obtained in different surveys appear to be rather variable and conflicting. While this may be accounted for by many factors, rigid cytogenetic characterization of individual cases seems to be a matter of prime importance. Thus, certain common clinical features have been suggested to exist in AML patients with the 8/21 translocation, referring to the median survival and age distribution of the patients as well as some hematological pictures.\(^1,7,9\)

Acknowledgments. We sincerely thank Emeritus Professor Dr. Sajiro Makino, Drs. C. Mikuni, I. Maekawa, T. Morioka, H. Sakai, T. Watanabe, T. Kobayashi, Y. Gocho and S. Ohtsuka.

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