III. SUMMARY

Some of the topics relevant to solid tumors are also relevant to the hematological malignancies, for example, the endpoint of survival, prognostic factors regarding outcome for untreated patients, and predictive factors that predict the success of drugs. But the hematological malignancies are distinguished by the fact that the concept of metastasis is not applied, by the phenomenally high cure rate for some of the leukemias (HCL, APL), by the fact that the hematological neoplasms may be unusually difficult to distinguish from each other, and by the fact that the RECIST criteria are not used.

IV. CYTOGENETICS OF HEMATOLOGICAL CANCERS

a. Introduction

The human chromosomes are numbered 1−22, with two additional chromosomes called X and Y. Chromosomes 1−22 occur as two copies in every somatic cell. The X chromosome occurs as two copies in every female somatic cell, but only once in every male somatic cell. The Y chromosome occurs only once in every male somatic cell. Thus, the sex chromosomes in males are XY, and the sex chromosomes in females are XX. Altogether, human somatic cells have 46 chromosomes (207). During mitosis, the genome condenses to form chromosomes that can be seen using a light microscope. The appearance of these chromosomes is called the *karyotype*.

In a point in the cell cycle, that is, during metaphase, each of the chromosomes can be seen to have two arms, the p arm and the q arm. The letter p refers to the short arm, while q refers to the long arm. Within each arm, numbers are assigned to large areas called regions, and another set of numbers is used to refer to bands within the regions. Numbering starts from the centromere, and increases as one moves towards the tip of each arm (208).

To provide an example, the term "14q32" refers to the second band in the third region of the q arm of chromosome 14 (209). (It is not the case that the number 32 is read as thirty-two. Instead, it is read as three-two.) Another example is as follows. The breast cancer gene *BRCA1* is located at 17q21.31. This means that the gene is located on the q arm of chromosome 17, in region 2. Within region 2, the gene is located in band 1. Collectively, this may be called, "band two, one." Within band 21, the gene resides in sub-band 31. Regarding the number 31, the 3 refers to a sub-band, and the number 1 refers to a sub-band within sub-band 3 (210).

b. Cytogenetics for Diagnosis and Prediction—AML

The following cytogenetic markers are used for predicting outcome for AML (211). These markers predict favorable prognosis, intermediate prognosis, and poor prognosis, as indicated:

[207]Tijio JH, Levan A. The chromosome number in man. Hereditas 1956;42:1−6.

[208]Pasternak JJ. An introduction to human genetics. 2nd ed. Hoboken, NJ: John Wiley and Sons, Inc.; 2005. p. 27.

[209]Jorde LB, Carey JC, Bamshad MJ, White RL. Medical genetics. 3rd ed. St. Louis, MO: Mosby; 2003. p. 108.

[210]Pasternak JJ. An introduction to human genetics. 2nd ed. Hoboken, NJ: John Wiley and Sons, Inc.; 2005. pp. 27−8.

[211]Gregory TK, Wald D, Chen Y, Vermaat JM, Xiong Y, Tse W. Molecular prognostic markers for adult acute myeloid leukemia with normal cytogenetics. J. Hematol. Oncol. 2009;2:23.