

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 (165).

To provide an example, the term “14q32” refers to the second band in the third region of the q arm of chromosome 14 (166). (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 (167).

### a. Cytogenetics for diagnosis and prediction – AML

The following cytogenetic markers are used for predicting outcome for AML (168). These markers predict favorable prognosis, intermediate prognosis, and poor prognosis, as indicated:

- Favorable prognosis: inv(16); t(15;17); and t(8;21)
- Intermediate prognosis: no identifiable abnormal cytogenetics
- Poor prognosis: monosomy 5; monosomy 7; and 11q23.

These cytogenetic markers result in the creation of a number of fusion genes, as indicated in Table 17.3. AML patients with no identifiable abnormal cytogenetics (using the microscope) have been classified according to a number of specific genetic mutations, as determined by DNA sequencing, where these genetic mutations provide prognostic value. The prognostic values of these mutant genes are also indicated in the table.

### b. Cytogenetics for diagnosis and prediction – ALL

Cytogenetic characteristics may be the most important prognostic factor for ALL, according to Cortes and Kantarjian (169). These abnormalities can take the form of

<sup>165</sup> Pasternak JJ. *An Introduction to Human Genetics*. 2nd ed. Hoboken, NJ: John Wiley and Sons, Inc.; 2005; p. 27.

<sup>166</sup> Jorde LB, Carey JC, Bamshad MJ, White RL. *Medical Genetics*. 3rd ed. St. Louis, MO: Mosby; 2003; p. 108.

<sup>167</sup> Pasternak JJ. *An Introduction to Human Genetics*. 2nd ed. Hoboken, NJ: John Wiley and Sons, Inc.; 2005; pp. 27–28.

<sup>168</sup> 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.

<sup>169</sup> Cortes JE, Kantarjian HM. Acute lymphoblastic leukemia. A comprehensive review with emphasis on biology and therapy. *Cancer*. 1995;76:2393–2417.