- 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 the following table. 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. Table 18.3 discloses the prognostic value of these mutant genes.

c. Cytogenetics for Diagnosis and Prediction—ALL

Cytogenetic characteristics may be the most important prognostic factor for ALL, according to Cortes and Kantarjian (212). These abnormalities can take the form of changes in the number of chromosomes, or changes in the structure of the affected chromosomes, as shown in Table 18.4. All numbers (%) in the table are from Cortes and Kantarjian (213), unless specified otherwise.

This concerns some of the abnormalities listed in the table.

1. Numeric Abnormalities in ALL

Patients with hyperdiploid ALL, in particular those with more than 50 chromosomes, have the best prognosis. Hypodiploidy (less than 44 chromosomes), which is found in less than 2% of pediatric or adult cases, predicts a poor outcome. The rare cases with low hypodiploidy (33–39 chromosomes) and near-haploidy (23–29 chromosomes) have a particularly poor prognosis.

2. Structural Abnormality t(9;22) (Philadelphia Chromosome) in ALL

The outcome for ALL patients with blasts containing the Philadelphia chromosome is poor (214). The Philadelphia chromosome in ALL may be different from that found in CML. In ALL, this chromosome involves band 34 of the long arm of chromosome 9, splicing the proto-oncogene c-*abl* to band 11 of the long arm of chromosome 22 in the *bcr* gene. In 50–80% of cases of ALL, the breakpoint in 22q11 falls between exons b1 and b2 of the major breakpoint cluster region, as opposed to between b2 and b3 or b3 and b4 in CML. The difference is in the positions of the breakpoints occurring in the translocation, that is, breakpoints within the *BCR* gene. This difference in breakpoints results in a smaller polypeptide of 190 kDa (p190 *BCR/ABL*) in ALL, and a larger polypeptide in CML (210 kDa, p210 *BCR/ABL*) (215,216). Both polypeptides have increased tyrosine kinase activity.

[212]Cortes JE, Kantarjian HM. Acute lymphoblastic leukemia. A comprehensive review with emphasis on biology and therapy. Cancer 1995;76:2393–417.

[213]Cortes JE, Kantarjian HM. Acute lymphoblastic leukemia. A comprehensive review with emphasis on biology and therapy. Cancer 1995;76:2393–417.

[214]Cortes JE, Kantarjian HM. Acute lymphoblastic leukemia. A comprehensive review with emphasis on biology and therapy. Cancer 1995;76:2393–417.

[215]Score J, Calasanz MJ, Ottman O, et al. Analysis of genomic breakpoints in p190 and p210 BCR-ABL indicate distinct mechanisms of formation. Leukemia 2010;24:1742–50.

[216]Cortes JE, Kantarjian HM. Acute lymphoblastic leukemia. A comprehensive review with emphasis on biology and therapy. Cancer 1995;76:2393–417.