Chronic myelogenous leukemia is a clonal cancer arising from neoplastic transformation of a hematopoietic stem cell. A biologic feature of the leukemic blast that has been demonstrated to be a strong predictor of treatment response is the presence of certain specific chromosome translocations. The first translocation shown to have prognostic significance is the t(9:22). It is characterized by a reciprocal chromosomal translocation t(9:22)(q34;q11). The resulting shortened chromosome 22 is classically called the Philadelphia Chromosome (Ph'). In CML, studies have found that the break in chromosome 22 is limited to a 5.8 kb section named breakpoint cluster region (bcr), specifically bcr exon 2 or exon 3, and the reciprocal break in chromosome 9 involves movement of most of the ABL proto-oncogene. Although cytogenetically indistinguishable from its CML version, the 9:22 translocation found in Acute Lymphocytic Leukemia (ALL) (some 6% of childhood ALL and 17-25% of adult ALL) has its chromosomal breakpoint in a more 5' bcr exon while the ABL sequence remains the same. This results in a 7 kb chimeric mRNA transcript and is translated into a 190 kDa protein, p190, also with increased tyrosine kinase activity.

Expression of these chimeric fusion proteins is thought to be important in the pathogenesis of these neoplasms and have recently been shown to be strong predictors of poor prognosis. The t(9:22) identifies a group of pediatric ALL patients with an extremely poor prognosis and few long term survivals. Presence of the p190 bcr/abl transcript is a predictor of poor prognosis and may indicate the need for aggressive treatment options such as bone marrow transplantation (BMT). Therefore, patients whose leukemic blasts contain the t(9:22) are typically offered the option of bone marrow transplantation (BMT) soon after remission is achieved. Bone marrow transplantation (BMT) is often the treatment of choice for adult Ph' ALL. RT-PCR can be used to monitor patients for the persistence of the bcr-abl transcript following transplantation and is the best method available to detect residual disease and predict relapse post BMT.

This procedure has been shown to detect one abnormal cell in a background of one million normal cells. Due to its sensitivity, PCR analysis is appropriate for both bone marrow and peripheral blood analysis and can be performed on recovering marrow with extremely low cell counts.