AML with t(8;21)(q22;q22); (RUNX1–RUNX1T1)

Myeloblasts show a characteristic perinuclear hofs and b salmon colored cytoplasmic granules. These features are typically more prominent in the bone marrow and less apparent in the peripheral blood.

Flow cytometry histograms of the bone marrow from a patient with AML with t(8;21)(q22;q22)

The leukemic blasts (red) express CD34, CD38, CD13, HLA-Dr, partial CD19, CD56, CD45 and myeloperoxidase, characteristic of AML with a t(8;21). The blasts lack expression of CD20 and CD16.
Acute Myeloid Leukemia With Recurrent Genetic Abnormalities

AML with t(8;21)(q22;q22) (RUNX1–RUNX1T1) and AML with inv(16) (p13.1q22) or t(16;16)(p13.1;q22) (CBFB–MYH11) comprise the core binding factor leukemias, with disruption of the core binding factor α and β subunits, respectively. Core binding factor leukemias are associated with a favorable prognosis in children and adults, especially when treated with repetitive cycles of high-dose cytarabine (HiDAC) post-remission. Cases of t(8;21) AML with a WBC $>20 \times 10^3/\text{mm}^3$ ($>20 \times 10^9/\text{L}$) at presentation appear to behave more like intermediate risk disease and may benefit from allo-SCT in first remission. Mutations of KIT in core binding factor AML are common (20%–25%). In adults, KIT mutations in exons 8 and exon 17 appear to worsen prognosis. It is unclear if they have a similar prognostic effect in children, or whether t(8;21) AML with KIT mutation benefits from allo-SCT in first remission. Mutations in FLT3 are very uncommon in core binding factor leukemia. Additional cytogenetic abnormalities are present in the majority of t(8;21) AML, most commonly including loss of a sex chromosome, or partial deletion of the long arm of chromosome 9 [del(9q)]. The presence of an unfavorable additional cytogenetic abnormality, such as monosomy 7, may adversely impact prognosis.

RT-PCR may detect RUNX1–RUNX1T1 transcripts in the absence of any clinical disease. The mRNA can be detected in some stem cells, mature monocytes, and hematopoietic progenitors during remission. Quantitative PCR, measuring the kinetics of RUNX1–RUNX1T1 transcripts, appears more useful for monitoring minimal residual disease.

46.1.2 AML With inv(16)(p13;q22) or t(16;16)(p13;q22), (CBFB–MYH11)

AML with an inv(16)(p13;q22) or t(16;16)(p13;q22) comprises 10% of adult AML, and approximately 6% of childhood AML. The inv(16)(p13;q22) is a pericentric inversion of chromosome 16 i46.6. The genes at the breakpoint junction are the β subunit of CBF factor