

The significance of WT1 gene expression in blasts of acute leukemias and myelodysplastic syndrome (MDS) has been repeatedly studied, though with inconsistent results. In our study, we focused on determining WT1 expression using quantitative PCR in samples from pediatric patients with acute leukemias, myelodysplastic syndrome, aplastic anemia, and anaplastic large cell lymphoma. Control samples included bone marrow from healthy donors, umbilical cord blood samples, and regenerating bone marrow samples, which express the WT1 gene at very low or undetectable levels.

WT1 gene expression in blasts of both acute myeloid and lymphoblastic leukemias showed a wide range of values. In lymphoblastic leukemias, high expression was associated with the presence of the MLL/AF4 fusion gene, whereas patients positive for BCR/ABL and TEL/AML1 showed low WT1 expression. T-cell lymphoblastic leukemias exhibited higher levels of WT1 expression than B-cell acute lymphoblastic leukemias.

In myeloid leukemias, high expression was associated with less differentiated subtypes (particularly subtype M3), whereas M5 subtype leukemias expressed WT1 at significantly lower levels. A statistically significant worse prognosis was observed in children with ALL who had either very high or very low WT1 expression. In pediatric AML patients, significantly better leukemia-free survival was found in those with WT1 expression below 100 normalized WT1 copies.

Further monitoring and a larger patient cohort are necessary to assess the predictive value of WT1 gene expression for relapse. In particular, the question of whether WT1 levels could be used for measurable residual disease (MRD) monitoring and prognostic assessment during specific treatment time points in pediatric AML is to be answered by an international study, in which we are participating in coordination.

Patients with aplastic anemia show very low or undetectable WT1 transcript levels, whereas patients with myelodysplastic syndrome, as a group, exhibit higher WT1 gene expression.