Acute Myeloid Leukemia – Down Syndrome: A Case Study from a Children's Hospital

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ABSTRACT

Congenital conditions like Down syndrome have been associated with increased risk for clonal disorders that affect megakaryocytic lineage. The World Health Organization has classified acute myeloid leukemia – Down syndrome (ML-DS) as a specific subtype of acute myeloid leukemia. Studies have shown that children with ML-DS have a better prognosis than children without Down syndrome with a long-term survival rate of 74%–91%. One possible explanation for this differentiation is that the megakaryoblast cells in ML-DS have increased sensitivity to cytotoxic drugs like cytarabine-based therapy. This has been attributed to the GATA1-mutant isoform decreased expression of cytidine deaminase and the overexpression of the cystathionine-β–synthase, a chromosome 21–localized gene present in ML-DS megakaryoblast cells. This case study followed the course of diagnosis and treatment of a 17-month-old patient with ML-DS. GATA1 mutation was confirmed in this case, and flow cytometry identified a megakaryoblast population expressing cluster differentiation markers of CD13, CD33, CD34, CD7, CD36, CD41, CD61, CD71, and CD117 in approximately 43.5% of the sample. As a result of the flow cytometry, physicians confirmed ML-DS as the diagnosis. The patient received 2 rounds of a chemotherapy treatment that included a combination chemotherapy regime with intrathecal chemotherapy to prevent relapse. Within 1 month of treatment, the patient was in remission and has remained with negative minimal residual disease to date.

ABBREVIATIONS: ML-DS - acute myeloid leukemia – Down syndrome.

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