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

LEANNE HUTSON, JOY OBUEKWE

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.

Clin Lab Sci 2021;34(2):14

***LeAnne Hutson**, Tarleton State University*

***Joy Obuekwe**, Tarleton State University*

***Address for Correspondence: Joy Obuekwe**, Tarleton State University, obuekwe@gmail.com*