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A Case Series on Transient Abnormal Myelopoiesis in Down Syndrome

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Introduction: Transient Abnormal Myelopoiesis (TAM) is a myeloproliferative disorder that can develop in 10% to 15% of newborns with Down syndrome. It may present with a high count of blast cells and can resemble congenital leukemia. This series discusses two cases of TAM, both of which achieved sustained remission a few months after their diagnosis. **Case Report:** In the first case, a baby boy with Down syndrome presented with congenital pneumonia at birth, bilateral eye conjunctivitis and an enlarged liver. Blood film showed 34% blast cells, and thrombocytopenia. After a month of stay he was discharged with a normalized blood count and no detectable blast cells in the peripheral blood film. While the second case involved a baby girl with Down syndrome who was treated for congenital pneumonia. Her total white blood cell count was elevated, with 33% blast cells characterized by prominent nucleoli and bluish, blebbing cytoplasm. After a week, her blast cells increased to 80%. Immunophenotyping performed showed 72% of blast cells which displayed heterogenous positivity for CD34, CD33, HLADR, CD61 (in 54% of blast population), CD41a (in 54% of blast population), CD36, CD56, CD123, and CD9 (megakaryocytic phenotype). They were negative for cy MPO, B and T cell markers. She started treatment with low-dose Cytarabine. **Discussion:** Down syndrome is associated with TAM and and myeloid leukemia associated with Down Syndrome (ML-DS). The detection of *exon2/3 GATA1* mutation via sequencing is essential in the diagnosis of both TAM and ML-DS however this test is not readily available at our center. TAM typically resolves within three months. However, close monitoring is essential because multiorgan failure can lead to a mortality rate of up to 20%.

Keywords: Neonates; non-disjunction trisomy 21; transient abnormal myelopoiesis