DOI: 10.7860/IJNMR/2017/24550.2199

Case Report

Neonatology Section

Transient Abnormal Myelopoiesis in Down Syndrome

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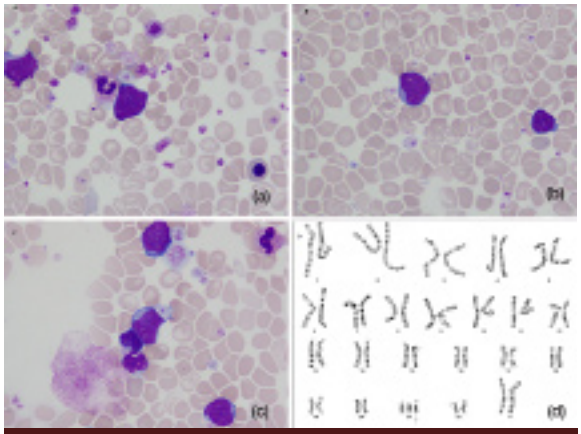
ABSTRACT

We report peripheral blood film and cytogenetic results of a five day old neonate who presented with history of high grade fever, vomiting and jaundice. Peripheral blood film showed presence of 30% blast cells which were large in size having abundant granular cytoplasm. Karyotyping results revealed presence of trisomy 21. She was diagnosed as a case of Down syndrome with congenital leukemia. Approximately 10% of such cases present as Transient Abnormal Myelopoiesis. Persistence of blast cells beyond six months is an indication for treatment. As the diagnosis was made retrospectively, this case highlights the importance of peripheral smear review which showed presence of abnormal cells prompting for further investigations.

Keywords: Congenital leukemia, Neonate, Peripheral smear

CASE REPORT

We report peripheral blood film and cytogenetic results of a five day old neonate, after taking consent from the parents. He was presented with history of high grade fever, vomiting and jaundice for two days. Her weight at birth was approximately 3kg and rest of the physical examination was unremarkable. Her complete blood counts at 3rd day of life showed Hb: 14.8 g/dl, HCT: 44.9%, MCV: 93.9fl, MCH: 31.0pg, WBC: 29 X 10^9/l and Platelets: 482 X 10^9/l. Peripheral blood film was reviewed which revealed 30% blast cells. These blast cells were large in size having abundant granular cytoplasm. The nuclei of these blast cells showed open chromatin and prominent nucleoli. Subsequently Immunophenotyping by flow cytometry was performed which was consistent with acute myeloid leukemia. Based on peripheral film findings, blood chromosome sample was also sent which showed trisomy 21 in all 20 counted cells [Table/Fig-1a-d]. Considering the age at presentation, the patient was labeled as a case of Transient Abnormal Myelopoiesis. Further, history regarding the consanguinity was taken which was negative in this case. Mother's antenatal work-up included ultrasound abdomen and pelvis while anomaly scan was not performed. The child did not have any dysmorphic features. Unfortunately the baby could not survive and succumbed to death on the seventh day of life.



[Table/Fig-1a-d]: Blast cells seen on peripheral blood film at 100X. Trisomy 21 on peripheral blood chromosomes report.

DISCUSSION

Congenital leukemias diagnosed in the first month of life are rare malignancies which account for less than 1% of childhood cancers [1]. Children with Down syndrome (DS) are predisposed to develop congenital leukemia particularly acute myeloid leukemia [2]. Approximately 10% of patients born with DS develop Transient Abnormal Myelopoiesis (TAM) [3]. Development of TAM is due to the mutations in the second exon of GATA1 gene [4].

These patients require regular follow-up and supportive care rather than upfront chemotherapy. Spontaneous remission occurs in three to six months of life [5]. However, persistence of blast cells beyond seven months is an indication of treatment with chemotherapeutic agents.

Our patient was born through an uneventful spontaneous vaginal delivery and developed rising white blood cell counts on third day of life. Peripheral blood chromosomes performed later trisomy 21. We retrospectively diagnosed DS in view of peripheral blood film findings.

CONCLUSION

Based on the findings of peripheral smear, we retrospectively diagnosed Down syndrome associated Transient Abnormal Myelopoiesis. Our patient could not survive beyond seventh day of his life. The treatment includes surveillance with complete blood counts till 6 months of age. Persistence of leukemia beyond six months is an indication for treatment.

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FINANCIAL OR OTHER COMPETING INTERESTS:
None.

Date of Publishing: **Jan 01, 2017**