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JUVENILE MYELOMONOCYTIC LEUKEMIA PRESENTING WITH RESPIRATORY DISTRESS AND SPLENOMEGALY IN A NEONATE: A CASE REPORT

Dr. Tagilipelli Sai Teja* Department of Pediatrics, Rajarajeswari Medical College & Hospital, Bangalore*Corresponding Author

Dr. Y Sree Krishna Professor Department of Pediatrics, Rajarajeswari Medical College & Hospital, Bangalore

ABSTRACT

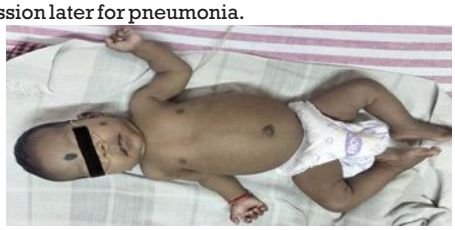
Juvenile Myelomonocytic Leukemia (JMML) is a rare clonal hematopoietic stem cell disorder of early childhood characterized by excessive proliferation of myelomonocytic cells. It accounts for approximately 1–2% of all childhood leukemias and commonly presents with leukocytosis, monocytosis, anemia, thrombocytopenia, and splenomegaly. We report a case of a term female neonate with respiratory distress at 45 hours of life requiring NICU admission. Examination revealed splenomegaly with thrombocytopenia. Peripheral smear showed leukocytosis with monocytosis and immature myeloid cells.Metabolic and newborn screening were normal, and genetic evaluation was planned. Early recognition is important as hematopoietic stem cell transplantation is the only curative treatment.This case highlights the importance of considering JMML in neonates with unexplained leukocytosis and splenomegaly.

INTRODUCTION

Juvenile Myelomonocytic Leukemia (JMML) is a rare clonal hematopoietic stem cell disorder of early childhood characterized by excessive proliferation of myelomonocytic lineage cells, with overlapping features of myelodysplastic and myeloproliferative neoplasms. It accounts for approximately 1–2% of pediatric leukemias, with an incidence of about 1.2 cases per million children per year, and typically presents in infants and young children, with neonatal cases being extremely rare.The disease is driven by dysregulation of the RAS signaling pathway, leading to abnormal proliferation of monocytic lineage cells. Unlike acute leukemias, blast counts remain low, while mature myelomonocytic cells predominate. This results in suppression of other cell lines, causing anemia and thrombocytopenia. Clinically, JMML presents with leukocytosis, monocytosis, and splenomegaly. Diagnosis is based on clinical and hematological criteria along with genetic confirmation. Early recognition is crucial, as hematopoietic stem cell transplantation (HSCT) remains the only curative therapy.

CASE REPORT

A term female neonate, was born at 37+3 weeks via normal vaginal delivery with a birth weight of 2.7 kg and good APGAR scores. The antenatal period was uneventful except for maternal anemia requiring transfusion. At 45 hours of life, the baby developed respiratory distress with tachypnea and retractions, requiring NICU admission. She was managed with high-flow nasal cannula oxygen and empirical antibiotics for suspected early-onset sepsis, later escalated due to persistent symptoms. On examination, significant splenomegaly (4 cm below left costal margin) was noted. Laboratory investigations showed leukocytosis (14,700/mm^3) with marked monocytosis (30%) and thrombocytopenia (1.01 lakh/mm^3). CRP was negative, and metabolic and newborn screening were normal. Peripheral smear revealed immature myeloid cells. Differential diagnoses including sepsis, metabolic disorders, and hematological conditions were considered. Whole exome sequencing confirmed JMML. The baby was initially stabilized and discharged but required readmission later for pneumonia.



DISCUSSION

JMML is a rare pediatric myeloproliferative disorder caused by mutations in the RAS pathway, leading to excessive proliferation of myelomonocytic cells. It is characterized by persistent monocytosis, splenomegaly, and cytopenias, with low blast counts distinguishing it from acute leukemia. Neonatal presentation is uncommon and often mimics sepsis, leading to diagnostic delays. In this case, persistent splenomegaly and monocytosis prompted further evaluation beyond initial sepsis management.The differential diagnosis includes infections, metabolic disorders, immune deficiencies, and HLH. Genetic testing, particularly whole exome sequencing, plays a key role in confirming diagnosis and guiding management. HSCT remains the only curative treatment, with emerging mutation-directed therapies offering additional options in selected cases. Prognosis depends on factors such as age, platelet count, and genetic mutations. This case highlights the importance of considering JMML in neonates with unexplained splenomegaly and monocytosis, ensuring early diagnosis and timely management.

CONCLUSION

Juvenile Myelomonocytic Leukemia (JMML) is a rare neonatal malignancy that may mimic common conditions such as sepsis, leading to diagnostic delays. The presence of persistent splenomegaly, monocytosis, and thrombocytopenia should raise suspicion for an underlying hematological disorder.Early diagnosis through timely hematological and genetic evaluation is essential, as hematopoietic stem cell transplantation remains the only curative treatment. This case highlights the need for a high index of suspicion to enable prompt diagnosis and improved outcomes.

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