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CONGENITAL LEUKEMIA: A RARE CASE REPORT

Pathology

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ABSTRACT

Congenital Leukemia (CL) is a type of leukemia that develops intra uterine. It is a rare but well developed entity which presents after birth. Due to the doubling time of leukemic cells, the disease becomes clinically evident after birth or shortly thereafter. These leukemias comprise approximately 0.8 % of all childhood leukemias. Incidence is reported to be 1 in 5 million. We present a case of congenital acute myeloid leukemia manifesting from the third day of birth in a female child.Diagnosis of acute myeloid leukemia was suspected by the presence of blasts in the peripheral blood smear and was confirmed by flow cytometry.

KEYWORDS

Congenital Leukemia, childhood leukemias.

INTRODUCTION:

Congenital Leukemia is a rare leukemia which presents at birth. Incidence is reported to be 1 in 5 million[1]. Most CL are Myeloid in origin unlike pediatric leukemias which are Lymphoid[2]. There is a 20 fold increased risk of leukemia in individuals with Down syndrome (DS). The incidence rate of acute leukemias during the first 5 years of life is 50 times the rate among individuals without Down syndrome[3]. The main subtypes of AML in this age group are myelomonocytic, monocytic, and megakaryocytic leukemia (French-American British [FAB] classification M4, M5, and M7, respectively[4]. The clinical findings and biologic features of perinatal leukemia are distinctively different from those of older children and adults [4]. Leukemia in the newborn has an unexplained natural tendency to undergo spontaneous remissions lasting months or sometimes years[5]. Although the prognosis for the neonate with leukemia is generally discouraging, the outcome is much worse for patients, especially for females, with ALL than AML.

Case Report:

A female child weighing 2.4 kg was born to an elderly female in a non consanguineous marriage following a lower segment cesarean delivery and second in birth order .There was no history of exposure to cytotoxic drugs or radiation during the antenatal period to the mother. The baby was referred to our hospital with distention of abdomen and petechial spots over the trunk and body on her second day of birth.

On examination, there was pallor with no signs of cyanosis and facial dysmorphism. Per abdominal examination revealed liver which was palpable 4 cm below the right costal margin and spleen 3 cm below the left costal margin. The baby was admitted in the NICU and routine investigations were performed. Sepsis, ABO incompatibility, TORCH infections and congenital syphilis were ruled out. The hematological testing revealed a hemoglobin of 11.7g/dl and total leukocytes count of 1,65,000/mm3 . Platelets count was 100,000/mm3 . The peripheral blood smear showed normocytic normochromic blood picture with marked leucocytosis, mild shift to left, and 76% blasts. The blasts were large to medium in size with high N:C ratio, variable amount of cytoplasm, fine nuclear chromatin, two to three nucleoli and presence of cytoplasmic vacuolations. Bone marrow aspiration showed 82% blasts.Flow cytometry was performed which revealed positivity for CD38, HLA-DR , CD34, CD13, CD33 and CD11c with negativity for T and B cell markers except for CD7 which showed aberrant positivity. Based on the morphology and immunophenotyping report, the patient was given a diagnosis of Acute Myeloid Leukemia. Karyotyping was done which showed a normal 46 XY pattern.

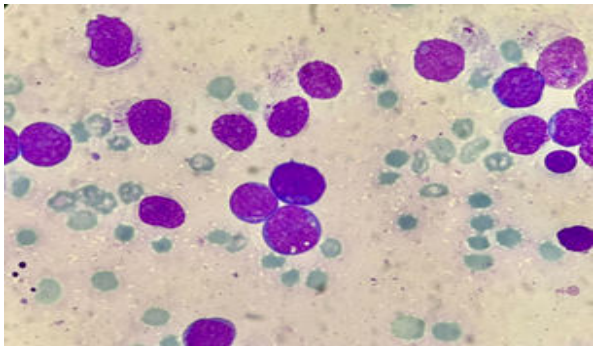

Fig 1: Photomicrograph showing blasts in peripheral smear

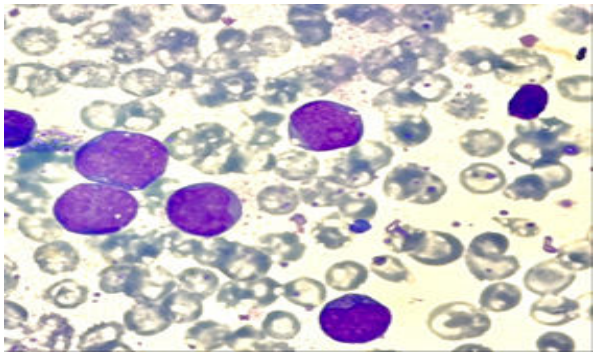

Fig 2: Photomicrograph showing blasts in peripheral having vacuoles (400x, Leishman's stain). Smear (400x, Leishman's stain)

DISCUSSION:

Congenital leukemia (CL) is defined as manifestations of leukemia within the first 4 weeks of life. CL is one of the most common cancers in neonates, following teratoma and neuroblastoma, and is lethal without chemotherapy[6]. The presentation of our case was within the

first two days of birth. The criteria for diagnosis of congenital leukemia is proliferation of immature white cells, infiltration of these cells into bone marrow, absence of any other disease that can cause leukemoid reaction mimicking leukemia i.e. congenital syphilis, blood group incompatibility and TORCH infection [7,8].

Clinical signs of leukemia may be evident at birth with hepatosplenomegaly, petechiae and ecchymosis. Twenty five to thirty percent of infants with CL have specific cutaneous infiltrates (leukemia cutis) which usually appear as firm blue or red nodules ('Blueberry Muffin')[2].

Our case also had similar presentation at birth which included hepatosplenomegaly and petechiae all over the body. The differential diagnosis of CL are sepsis, intrauterine infections (TORCH), hemolytic disease of the newborn (HDN) and transient myeloproliferative disease (TMD)[2,9,10].Infections were ruled out by serology and culture. HDN shows large number of erythrocyte precursors in the peripheral smear, which was absent in our case. TMD of the newborn is seen usually in association with Downs syndrome whereas in our case the karyotyping was normal.

The prognosis for CL is poor, with only 23% surviving at 24 months[4,5]. However, rare cases of CL with spontaneous remission have been described, most of which were associated with Downs´ Syndrome. Many infants die of respiratory distress secondary to pulmonary leukostasis and bronchopneumonia.

In summary, we present a third day old neonate with CL. Our child hardly showed any improvement in her blood picture and succumbed to the leukemic process by 3 weeks of age.

Thus, leukemia must be kept in mind in a newborn with clinical features of sepsis and leukocytosis.

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