Case Report

A case report of congenital leukemia – AML M0

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ABSTRACT

Congenital leukemia (CL) refers to leukemia diagnosed at birth or within the 1st month of life. Incidence is reported to be 1 in 5 million. We report the case of a 22-day-old neonate, who presented to us with features of sepsis, with predominant blasts in the peripheral smear, and was subsequently detected to have CL. Although the etiology is unknown, the presence of leukemia at birth suggests possible intrauterine exposure to drugs or other toxins or the presence of genetic abnormalities. The course of CL is one of the rapid deterioration and death from hemorrhage and infection unless treated appropriately, with adequate chemo and radiotherapy. These neonates pose a huge challenge to treat due to their high mortality, owing to both the high risk of treatment-related mortality and disease relapse. Early diagnosis and prompt initiation of treatment are essential for a better prognosis.

Key words: Acute myeloid leukemia, Case report, Congenital leukemia, Neonates

Congenital leukemia (CL) is a term applied to leukemia diagnosed at birth or within the 1st month of life [1]. It is one of the most common cancers in neonates, after teratoma and neuroblastoma [2] but is nevertheless rare and almost always lethal without chemotherapy [3]. An estimated 175–200 cases are reported in the literature [4]. We report the case of a 22-day-old Indian neonate [4], who presented to us with features of sepsis, with predominant blasts in peripheral smear, and was subsequently detected to have CL.

CASE REPORT

A 22-day-old baby boy presented with complaints of fast breathing, persistence of umbilical cord, and abdominal distension, progressively increasing since birth. He was a term neonate (birth weight 3.45 kg), second order child of a non-consanguineous marriage, born to a 24-year-old mother, with antenatal history of asymptomatic COVID-19 infection in the 4th month of gestation, by normal vaginal delivery. The immediate perinatal period was uneventful.

On day 17 of life, the baby developed pus discharge from the umbilicus with poor activity, poor feeding, and fast noisy breathing. On examination, the baby was irritable, pale looking, and febrile with tachycardia and mild respiratory distress. Abdomen appeared distended with a liver span of 7 cm and a spleen palpable 5 cm below the left costal margin. No evidence of facial dysmorphism. Further evaluation revealed a hemoglobin of 11.7 g/dL, a total count of 1.33 lakh/µL with a lymphocyte predominance of 81%, and normal platelet levels. Peripheral smear showed lymphocytosis with predominant blasts. The sepsis workup was negative [Figure 1].

Cytochemical stains on peripheral smear - myeloperoxidase staining was negative for the majority of blasts (3% blast positivity) and periodic acid-schiff showed classical block positivity. Flow cytometry of peripheral blood showed positivity for CD 13, CD 117, CD11c, CD123, CD34, and HLA DR with co-expression of CD5 and CD7. Fluorescence in situ hybridization done on peripheral blood showed the presence of the PML RARA (promyelocytic leukemia/retinoic acid receptor alpha) fusion gene. A final diagnosis of congenital acute myeloid leukaemia (AML) with minimal differentiation (AML M0 French-American-British [FAB] Type) was made.

The baby was started on palliative chemotherapy with etoposide and cytarabine. After 3 rounds of chemotherapy, there was a considerable reduction in counts. While on chemo, the baby developed multiple secondary infections. Despite all supportive treatment, he succumbed to severe pneumonia and sepsis on day 66 of life. After the patient expired, verbal consent was obtained from the parents for public discussion of the patient's case.

DISCUSSION

The pathophysiology and prognosis of leukemia occurring in this age group are different from those in cases occurring later in life. Although the etiology is unknown, the presence of leukemia

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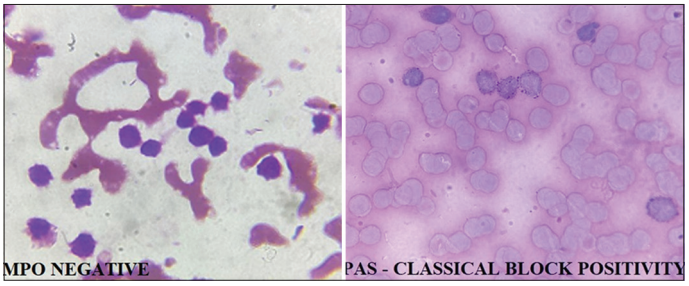

Figure 1: Peripheral smear showing predominant blasts, 2–3 times the size of mature lymphocytes with scant to moderate cytoplasm, some showing cytoplasmic pseudopod formation and round to irregular nucleus with opened up chromatin, showing 2–3 prominent nucleoli. No definite Auer rods seen

at birth suggests possible intrauterine exposure to drugs or other toxins or the presence of genetic abnormalities. Familial neonatal leukemia is extremely rare and no child born to a mother with leukemia has been found to have the disease during the neonatal period [5].

The clinical signs of leukemia may be evident at birth with hepatosplenomegaly, petechiae, and ecchymosis. In those infants who develop the disease within the 1st month (not at birth), the symptoms are usually ill-defined with low-grade fever, diarrhea, hepatomegaly, and failure to gain weight [6]. The criteria for diagnosis of CL are (a) disease presentation at or shortly after birth (<30 days), (b) proliferation of immature white cells, (c) infiltration of the cells into extra hematopoietic tissues, (d) absence of any other condition that mimics CL [4].

Most CL are of myeloid origin, unlike pediatric leukemia which is usually lymphoid. The predominant subtypes of AML in neonates are myelomonocytic, monocytic, and megakaryocytic leukemia (classification M4, M5, and M7 according to FAB) [7]. Etiological considerations in CL have included chromosomal defects, intrauterine environmental insults, viral infections, and exposure to radiation in pregnancy [8].

The outcomes seen in congenital AML are generally bleak, with no significant change in the past 2 decades of AML therapy. These neonates pose a huge challenge to treat due to their high mortality, owing to both the high risk of treatment-related mortality and disease relapse. Early diagnosis and prompt initiation of treatment are essential for a better prognosis in these cases.

Once the diagnosis of CL is established, an intensive multiagent chemotherapy regimen should be initiated. There is no specific treatment protocol for the treatment of neonatal ALL or AML. The therapy of congenital lymphoblastic leukemia remains the same as for older children, with the precaution that dosage needs to be altered to prevent undue toxicity of hypotonia, poor cry, and flaccid paralysis with vincristine [9]. Chemotherapy with steroids, vincristine, L-asparaginase, 6-mercaptopurine, and methotrexate together with anthracyclines and cytarabine are usually used [10]. The prognosis for CL is poor, with only 23% surviving at 24 months [11]. However, rare cases of CL with spontaneous remission have been described, most of which were associated with Downs' Syndrome or Noonan's syndrome [12].

CONCLUSION

Although rare, CL should be considered in a child who presents with hepatosplenomegaly and clinical features of sepsis. Being a largely underreported and underdiagnosed scenario, there is a growing need for more research focus in this area.

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