A Case of Organic Acidemia: Are Physicians Aware Enough?

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ABSTRACT

Diversity of clinical presentation of inborn errors of metabolism (IEM) gives a diagnostic challenge to the practicing physicians. In recent years there have been dramatic advances in the diagnosis and treatment of these fatal diseases, which shows improved prognosis of many of these conditions. The need for screening for IEM arises out of the fact that most cases take to irreversible effects as time progresses. Here, the main challenge is to recognize the early signs and symptoms that are also common to sick infants with other diseases. We describe a 32-day-old female infant who was finally diagnosed as a case of IEM (organic acidemia). The baby had a history of multiple neonatal intensive care unit admission. She died during her last hospital admission. The purpose of our case presentation is to provide clues to the true nature of the disease and to make physicians aware about the possibility of IEM.

Keywords: Fatal prognosis, Inborn error of metabolism, Organic academia.

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CASE REPORT

A 32-day-old female infant, 2nd issue of its nonconsanguineous parents, was referred from a private hospital to the neonatal intensive care unit (NICU) of Bangabandhu Sheikh Mujib Medical University (BSMMU), Dhaka, Bangladesh, due to respiratory distress and less activity for 1 day. She also had feeding intolerance characterized by emesis 5 to 10 times a day, mostly following feeding since 11 days of age.

The baby was delivered by a lower uterine cesarean section (LUCS). Her mother is 34 years old, G5 P2 Ab2 magnetic resonance imaging (MRI), blood group O+ ve, on regular antenatal check-up, had a history of diabetes mellitus, but no fever, rash, or any other chronic illness. In the last trimester she developed hypertension, for which LUCS was done at 36 weeks of pregnancy in a private hospital.

She gave birth to a female child weighing 1750 gm who cried immediately after birth with APGAR score of 7 at 1 minute and 9 at 5 minutes. As the baby was preterm with low birth weight, she was transferred and admitted up to 9th postnatal day in the NICU of the same hospital. After discharge, she had no complaint for 2 days at home and was nursed without difficulty. At 11th postnatal age she developed vomiting containing milk, after almost every feed. The baby was again admitted to a private hospital. During the hospital stay of 14 days, she developed hypoglycemia, convulsion, and features of sepsis and was treated with IVF, IV antibiotics, IV phenobarbitone, IV fosphenytoin, and IV NaHCO₃.

On arrival at the NICU of BSMMU on day 32, the baby was lethargic and emaciated. The following were...
noted: \( \text{SpO}_2 \): 90% at room temperature; vitals: heart rate 134/minute, RR 58/minute, temperature 36.0°C, CRT <3 seconds, CBG 3.2 mmol/L; anthropometry: OFC 30 cm, wt 1550 gm, length 35 cm. Her head, ears, eyes, nose, and oropharyngeal structures were without obvious abnormalities. Her lungs were clear, but notable intermittent grunting was present. Her heart was normal with no murmurs. Her abdomen was flat and soft, but liver palpitation of 2 cm was observed. Extremities were normal, with 1+ pulses. Genitalia had a normal female pattern. Her reflex activity was poor. Other findings were unremarkable.

Empiric antibiotics were started considering sepsis in mind after sending sepsis workup. Her ABO showed severe metabolic acidosis with an increased anion gap of 18. Bicarbonate infusion was initiated to treat the acidosis. The CBC report showed the following: Hb 13.5 gm/dL, total WBC 5730/mm\(^3\), DC-neutrophil 15%, lympho 74%, platelet count 1 lac, CRP positive, blood C/S no growth. The exact diagnosis depends on specialized enzyme assays and/or the identification of molecular defect. These methods are not very widely available, especially in our country. Such tests are, however, suitable only when there is a strong and more specific suspicion of IEM diagnosis. In our context we mainly depend on the high index of suspicion.

DISCUSSION

Inborn error of metabolism causes a group of hereditary metabolic diseases resulting from a lack of activity of 1 or more specific enzymes or defects in the transportation of proteins resulting in a block of metabolic pathway.\(^1,2\) The frequency of occurrence of organic aciduria is 1 in 15,000 according to Seymour CA et al.\(^5\) The term “organic acidemia” or “organic aciduria” (OA) applies to a group of disorders characterized by the excretion of non-amino organic acids in the urine.\(^6\)

Most organic acidemias become clinically apparent during the newborn period or early infancy.\(^7\) After an initial period of well-being, affected children develop a life-threatening episode of metabolic acidosis characterized by an increased anion gap. This presenting episode may be mistaken for sepsis and, if unrecognized, is associated with significant mortality.\(^1,2,6,8\) In our case as well we faced the same difficulties. The usual clinical presentation includes vomiting, poor feeding, neurologic symptoms such as seizures and abnormal tone, and lethargy progressing to coma.\(^2,9\) Appropriate laboratory testing for metabolic disorders should be performed in any infant who exhibits these findings.\(^8\)

CONCLUSION

An IEM could be a diagnosis of inclusion rather than exclusion. It is frequently underestimated by the doctors in neonatal and intensive care units of national health clinics or in private clinic. An increase in the rate of identification of these disorders is directly related to clinical judgment and a lifesaving difference can be made by appropriate and timely care for the small number of babies with 1 of these disorders.

REFERENCES