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Case Report

AN INTERESTING CASE OF PERSISTENT HYPOXEMIA IN A NEONATE

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ABSTRACT

We report a case of 2.8 kg term female baby born of 3rd degree consanguineous marriage referred at 22 hrs of life with complaints of respiratory distress and poor feeding. Elder sibling was a female who died at 30 days of life with complaints of respiratory distress, poor feeding and shock. At admission baby was in shock. Baby was ventilated and started on inotropes. In view of persistent hypoxemia despite 100% Fio₂, baby was started on inhaled nitric oxide therapy. Liver function test, CBC, Renal function test, Electrolytes were normal. Chest X ray was normal. Baby was euglycemic. There was a progressive fall in arterial PH from 7.352 at admission to 6.886 with base excess increasing from -5 to -20.1. Baby was given IV bicarbonate infusion. Despite this baby had 3 episodes of cardiac arrest within 15 hrs of admission and baby died at 38 hrs of life.

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INTRODUCTION

Mitochondrial DNA (mtDNA) depletion syndromes (MDS) are severe autosomal recessive disorders associated with decreased mtDNA copy number in clinically affected tissues. Mitochondrial DNA (mtDNA) depletion is a prevalent cause of mitochondrial respiratory chain deficiency (MRCD) during newborn period¹. Common neonatal presentations of MRCD in newborns include neonatal hypotonia or hypertonia, feeding and respiratory difficulties, cardiomyopathy and sudden neonatal death².

CASE REPORT

We report a case of 2.8 kg term female baby born of 3rd degree consanguineous marriage referred at 22 hrs of life with complaints of respiratory distress and poor feeding. At admission baby was cyanotic with saturation 55% on room air. Baby was ventilated in SIMV mode with minimal settings and prostaglandin infusion was started awaiting 2D ECHO. Echocardiography was performed bedside to rule out congenital cyanotic heart disease as the baby was well until 20 hrs of life and showed severe right ventricular cardiomyopathy and no evidence of structural heart disease. Baby was in compensated shock despite two normal saline boluses and hence started on inotropes, Dopamine and Dobutamine. Baby had persistent hypoxemia with saturation between 38-50%

despite 100% Fio₂ and Pao₂ 17-25mmHg. Serial arterial blood gas monitoring done. In view of oxygenation index above 20, baby was started on inhaled nitric oxide therapy and conventional ventilation continued. Chest X ray was normal. ABG showed progressive fall in arterial PH from 7.352 at admission to 6.886 with base excess increasing from -5 to -20.1. Baby was started on IV bicarbonate infusion. Lactate was 5.71mmol/L and Ammonia 87 μmol/L. Liver function test, Complete blood picture with septic screen, Renal function test, Serum Electrolytes were normal. Baby was euglycemic since birth. Baby had 3 episodes of cardiac arrest within 15 hrs of admission and baby died at 38 hrs of life. Serial ABG values, Saturation and Fio₂ summarized in Table 1. Blood was sent for metabolic studies. GALT enzyme and Total Galactose, Biotinidase enzyme, Serum 3- hydroxybutyrate, Carnitine / acyl carnitine profile and CDG screening by TIEF were normal. 17OHP was mildly elevated but not in the range for CAH. Serum free fatty acids mildly elevated. Plasma amino acids showed elevated Alanine and lysine along with mildly elevated phenylalanine, methionine and tyrosine. GC/MS of plasma showed elevated lactic acid, 3- hydroxybutyrate, succinate and fumaric acid. The results indicating a very strong possibility of Mitochondriopathy especially mitochondrial DNA depletion syndromes. Urine and CSF analysis could not be done due to the rapid deterioration of the baby. Elder sibling was a female who died at 30 days of life with similar

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complaints of respiratory distress, poor feeding and shock. Metabolic studies not performed on elder sibling. Molecular studies and autopsy offered to the family but denied due to cultural and financial reasons. We also advised them antenatal counseling and prenatal diagnosis for the subsequent pregnancy.

Table 1 Summary of investigations

Age in hrs	PH	PaO2	PCO2	FIO2	HCO3	BE	SPO2
20 hrs	7.352	24.8	35	60%	19.1	-5	45%
28 hrs	7.249	19.3	57.4	100%	19.7	-3.7	25%
32 hrs	6.872	17.9	105	100%	11.6	-16.9	14%
35 hrs	6.886	16	77.3	100%	8.9	-20.1	9%

DISCUSSION

Metabolic disease, possibly mitochondrial, was suspected in our case due to a combination of factors like: 1) Sibling with identical course and consanguinity; 2) Fulminant course without infection or other explanation; 3) Moderate elevation of lactate, persistent metabolic acidosis and hypoxemia and 4) Cardiomyopathy with cardiac arrest; 4) Non response to standard management. Mitochondrial enzymes deficiency forces reliance on glycolysis for ATP synthesis and result in lactic acidosis and energy failure³. These appear to be the consequences suffered by our baby.

She was normal at birth and became symptomatic by 18 hrs of life and developed cardio pulmonary arrest by 38 hrs of age. There appears to be a mismatch between large energy demands and rate of glycolytic ATP production in the heart.

CONCLUSION

We want to highlight that the prevalence of mitochondrial DNA depletion disorders is probably underestimated due to lack of awareness of such rare diseases, their clinical heterogeneity especially in neonatal period, and the difficulties in establishing the diagnosis. The literature and previous reports are scarce in neonatal period and this prompted us to report the present case.

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