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Case Report

Rare case report of mitochondriopathy

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ABSTRACT

We present the case of a term male neonate who was apparently well baby and deteriorated with hypoglycemic episode followed by deterioration with the child going in for respiratory failure and subsequently shock requiring ventilation and inotropic support which was refractory to all treatment, eventually resulting in a mortality. Investigative panel reports later showed that the baby had a mitochondriopathy-primary mitochondrial myopathy, which was the diagnosis responsible for the baby's clinical presentation. Although mitochondriopathies do not have a specific treatment, early diagnosis of milder variants and understanding their pathophysiology helps in further facilitating diagnostic approach for the future.

Keywords: Mitochondriopathy, Hypoglycemia, Newborn screening

INTRODUCTION

Mitochondriopathies are due to defects in mitochondrial enzymes, pathways and oxidative apparatus. They are inherited only from maternal cell line. They are classified as primary mitochondriopathies where mutations in the nuclear or mitochondrial DNA and secondary mitochondriopathies where it is due to exogenous factors.[1] Mitochondriopathies are rare and have an incidence of less than 1 per 1,000,000 and are not routinely screened for in the absence of a significant family history or recurrent pregnancy losses.

CASE REPORT

A 2.36 kg male baby was born to a primigravida mother at 38 weeks and 4 days gestation on 17-06-2022 at 10.52 am. The pregnancy was a spontaneous conception after 18 months of married life and there were no antenatal complications. Baby was vigorous at birth, noted to have thin meconium-stained liquor. Routine care was given to the baby and stomach wash given in view of meconium-stained liquor. Clinical examination ruled out gross congenital malformations and baby was observed in NICU for 1 hour before shifting to mother side, Baby was initiated on breastfeeds. Blood sugars were monitored every 4 hours for the baby according to institution protocol as baby was small for gestation age.

At 16 hours of life, baby was noted to be irritable and hence sugar monitoring was done at this time and was found to be 44 mg/dl, hence was advised to start on additional feeds due to hypoglycemia, after which baby's irritability reduced.

At 20 hours of life, baby's mother noticed that child had fast breathing and grunting. On examination, baby was noted to be pale, peripheries were cyanosed and peripheral pulses were not felt. Capillary refill time was prolonged (5 seconds). Respiratory examination showed paradoxical breathing pattern with subcostal retractions, grunting and gasping.

In view of the sick condition, baby was shifted to NICU and intubated with 3.0 uncuffed ET tube and fixed at 9 cm and mechanically ventilated in SIMV Mode (PIP-15,

PEEP-5, FiO_2-40%, rate-40). Umbilical vein catheter and umbilical artery catheters secured, 10 ml/kg bolus of normal saline given and baby started on inj. dopamine 10 mcg/kg.min and inj. dobutamine 10 mcg/kg/min. As baby condition clinically deteriorated (desaturations with bradycardia), ventilator settings increased in SIMV Mode (PIP-22, PEEP-6, FiO_2-100%, rate-40). Despite increased settings, there was clinical deterioration (desaturations with bradycardia), and hence ventilation switched to high frequency oscillatory ventilation (I:E-1:2, ^P-26, MAP-12). Baby maintained borderline saturations with above settings and started on inj. sodium bicarbonate correction at 1 mEq/kg/dose and started on inj. noradrenaline 0.5 mcg/kg/min along with 2nd bolus of 10 ml/kg NS. While in HFOV above settings, baby had a cardiac arrest with pulses not felt, BP and saturations not recordable. CPR initiated and given along with 3 inj. adrenaline and started on inj. adrenaline Infusion. Despite maximum resuscitatory efforts, baby was declared dead after 30 minutes of CPR.

Along with blood investigations, IEM Screening was sent for the baby through tandem mass spectometry (TMS). TMS revealed that the baby had a mitochondriopathy-primary mitochondrial myopathy. And on the basis of this investigation, we were able to establish a concrete diagnosis.

Table 1: Blood investigations of the baby.

Parameters	Value
Hb	14 g/dl
TC	19000 cells/mm^3
DC	N60L32
Platelet	215000 cells/mm^3
ESR	8 mm/hr
PCV	43%
Urea	27 mg/dl
Creatinine	1.53 mg/dl
Calcium	8.4 mg/dl
Na	141 mmol/L
K	4.8 mmol/L
Cl	105 mmol/Lt
SBR	4 mg/dl

Table 2: Serial ABG of the baby.

Variables	20 HOL	21 HOL	22 HOL	23 HOL
pH	7.07	7.05	6.99	6.67
pCO_2	77	76	88	95
pO_2	40	40	30	26
HCO_3	11	10	6	Not recordable

DISCUSSION

In general understanding, mitochondriopathies are due to defects in mitochondrial enzymes, pathways and oxidative apparatus. They are classified as primary mitochondriopathies where mutations in the nuclear or mitochondrial DNA and secondary mitochondriopathies where it is due to exogenous factors.[1]

Primary mitochondrial myopathy (PMM) are a rare subgroup of mitochondrial disease.[2] It affects males and females in equal proportion (1:1 ratio). PMM do not have a specific therapy and its prevalence is less than 1 in 1,000,000 population. Earlier manifestations of PMM have shown to be more severe in manifestation which include progressive cardiac manifestations such as cardiac failure, arrhythmia, sudden deaths and cardio-respiratory failure as well as metabolic abnormalities such as severe metabolic acidosis.[3]

The diagnosis of primary mitochondrial myopathy is by either Tandem Mass Spectrometry or by Exome sequencing. TMS can be recommended in older children with some developmental delay or with some other atypical clinical manifestation whereas exome sequencing is done in cases where there is a positive family history.[4]

Due to the low prevalence and rare occurance in clinical practice, mitochondriopathies are not routinely screened as a part of newborn screening unless specifically indicated.

There are limited case reports of mitochondriopathies occurring with neonatal manifestations. One such case is from Fiona et al who reported a case of infantile mitochondrial cardiomyopathy in a late preterm neonate in Germany.[5] Another case of mitochondriopathy presenting as respiratory distress was reported by Sowjanya et al from Chennai, India.[6]

Our case highlights the importance of Screening for Inborn errors of metabolism in NICU practice, as it helps to establish a definitive diagnosis for further prognostication and treatment. Early diagnosis helps in early identification and supportive treatment measures for the cases where it is applicable and where early interventions are associated with a positive outcome. Although in this case, the diagnosis was established post-mortem, it is of even more significance as the role of genetic counseling and antenatal diagnostic techniques becomes more valuable for the parents while planning for their next child.

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