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Anaesthetic Management of a Child with Methylmalonic Acidemia Undergoing Bilateral Orchidopexy – A Case Report

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Abstract

Background: Methylmalonic Acidemia (MMA) is an inborn error of metabolism caused by a deficiency in methylmalonyl CoA mutase, leading to increased protein catabolism, hyperammonemia and metabolic acidosis. This condition presents challenges to the anaesthesiologist for perioperative management due to metabolic instability, particularly during surgeries.

Case Presentation: A 1-year-old male child, weighing 8.5 kg and diagnosed with MMA, was posted for bilateral orchidopexy for undescended testes. Anaesthetic management was focused on preventing hypoglycemia, managing surgical stress with good analgesia, and avoiding metabolic acidosis. L-carnitine and methylcobalamin were administered preoperatively. General anaesthesia with inhalational induction was given, and a pre-incision caudal block for analgesia was performed. The child recovered without any complications.

Conclusion: We report a case of successful anaesthetic management of a child with MMA, emphasizing the importance of knowing the pathophysiology of the disease to prevent metabolic complications.

Keywords: Methylmalonic acidemia, Bilateral orchidopexy, Pediatric anaesthesia, Metabolic disorder.

Introduction

Methylmalonic Acidemia (MMA) is an inborn error of metabolism that affects the body's ability to break down certain proteins and fats. It is caused by a deficiency of the enzyme methylmalonyl CoA mutase. The resulting accumulation of methylmalonic acid leads to hyperammonemia, severe metabolic acidosis, and neurological deficits. Perioperative management of patients with MMA is challenging due to the potential for metabolic decompensation, hyperammonemia, and hypoglycemia during surgery and in the immediate post-operative period.

Case Presentation

A 1-year-old male child, weighing 8.5 kg, presented with a known diagnosis of MMA for bilateral orchidopexy for bilateral undescended testes. MMA was diagnosed by tandem mass spectrometry and genetic testing (Fig. 1). The scans showed that both the testes were in the inguinal region and hence the decision of open repair was taken (Fig. 2).

In order to minimize the metabolic derangements, the NPO period was strictly limited to 6 hours prior to the surgery (As per ASA guidelines) to avoid hypoglycemia and clear liquids were allowed upto 2 hours before the surgery, and 0.9% DNS (dextrose saline) infusion at 34 ml per hour was continued in the High Dependency Unit (HDU) till the baby was shifted to the Operation theatre. Pre operative Pediatrician consultation was obtained regarding the administration of L-carnitine and Methylcobalamin.

In the operating room, after connecting standard ASA monitors ECG, NIBP and Pulse oximeter, Blood sugars were checked and was 65 mg/dL, yet the child didn't show any signs of hypoglycemia and was active and pink, so intravenous bolus of 10 mL of 5% dextrose was given to avoid further hypoglycemia. The patient was premedicated with Inj glycopyrrolate 40 mcg IV and Inj Fentanyl 20 mcg IV and inhalation induction with sevoflurane which initially was 8% on the dial and later decreased to 2% was performed. Endotracheal intubation was achieved with a 4.0 mm uncuffed ET tube after Inj Atracurium 5 mg IV administration and was secured after 5 point auscultation. Anaesthesia was maintained using Oxygen: Air 50%: 50% and Sevoflurane 2%. Analgesia was achieved with a land mark guided caudal block using the modified Armitage formula, which was given prior to the incision. The baby didn't require any additional doses of Neuro muscular blocking drugs. Blood sugars were checked twice intraop and were above 120 mg/dl. IV fluid 0.9% Dextrose Normal Saline (DNS) was continued as the maintainance fluid intraoperatively at 34 ml per hour. Perioperative hypothermia was prevented by using Forced air warming device throughout the perioperative period. Intraoperative temperature monitoring was not done. Arterial blood gas analysis (ABG) was done and showed pH of 7.30 with Bicarbonate levels of 16.3, $PaCO_2$ levels of 34.0 and base deficit of 8.8 with normal lactate levels for which no correction was done (Fig. 3). The total duration of surgery was 45 mins and the child was later extubated without any adverse events (Fig. 4, 5).

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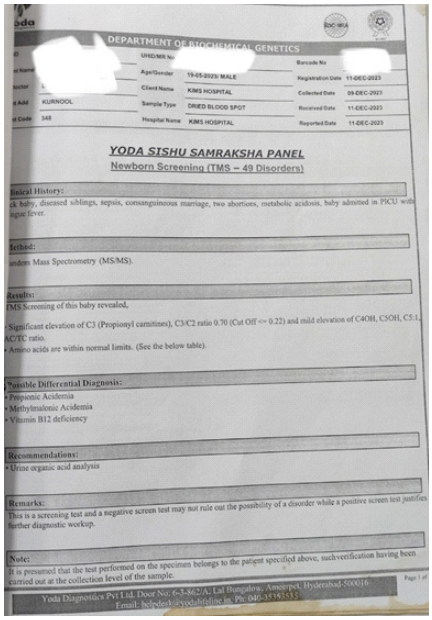


DEPARTMENT OF BIOCHEMICAL GENETICS

YODA SISHU SAMRAKSHA PANEL
Newborn Screening (TMS – 49 Disorders)

Clinical History:
ok baby, diseased siblings, sepsis, consanguinous marriage, two abortions, metabolic acidosis, baby admitted in PICU with ague fever.

Method:
andem Mass Spectrometry (MS/MS).

Results:
MS Screening of this baby revealed,
- Significant elevation of C3 (Propionyl carnitines), C3/C2 ratio 0.70 (Cut Off <= 0.22) and mild elevation of C4OH, C5OH, C5:1-
AC/TC ratio.
- Amino acids are within normal limits. (See the below table).

Possible Differential Diagnosis:
- Propionic Acidemia
- Methylmalonic Acidemia
- Vitamin B12 deficiency

Recommendations:
- Urine organic acid analysis

Remarks:
This is a screening test and a negative screen test may not rule out the possibility of a disorder while a positive screen test justifies further diagnostic workup.

Note:
It is presumed that the test performed on the specimen belongs to the patient specified above, subverification having been carried out at the collection level of the sample.

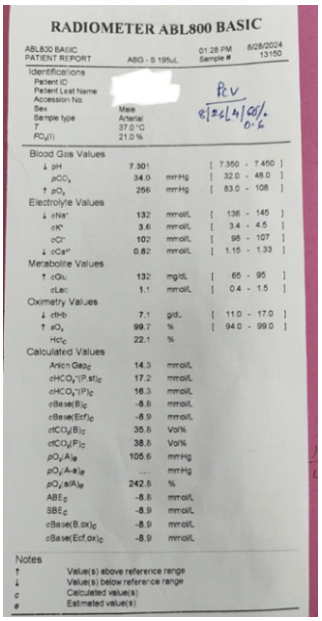
Yoda Diagnostics Pvt Ltd, Door No: 6-3-862/A; Lal Bungalow, Ameerpet, Hyderabad-500016.
Email: helpdesk@yodadiagnostics.in; Ph: 040-45553333

Figure 1: Tandem Mass spectrometry results showing elevate levels of Propionyl carnitines (C3) and a C3/C2 ratio of 0.70 (normal cutoff <0.22), indicating a possible diagnosis of Methyl malonic acidemia with differential diagnosis of Propionic acidemia and Vitamin B12 deficiency.

Discussion

Methyl malonic acidemia is a rare autosomal recessive disorder caused by mutations affecting the enzyme methylmalonil-CoA mutase, leading to the accumulation of methylmalonic acid and toxic metabolites. This results in severe metabolic disturbances, including metabolic acidosis, hyperammonemia, hypoglycemia, and hyperkalemia, all of which are of significant concerns during anaesthesia management [1].

The perioperative period in MMA patients can trigger metabolic crisis due to stress, fasting, or surgery, resulting in decompensation



RADIOMETER ABL800 BASIC

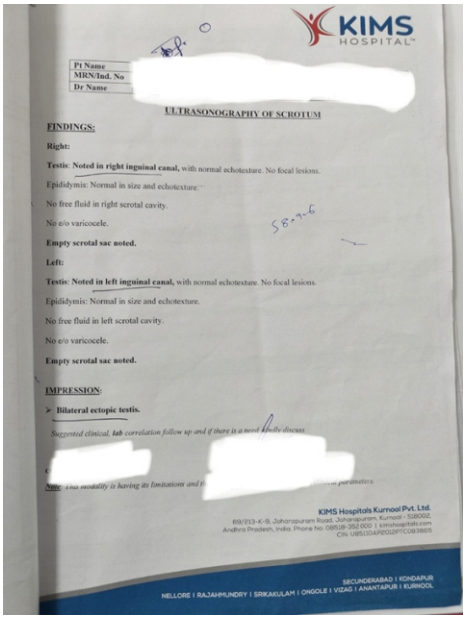
ABL800 BASIC PATIENT REPORT — ABG - S 195uL — 01:28 PM 8/28/2024 — Sample # 13150

Identifications
Patient ID
Patient Last Name
Accession No.
Sex: Male
Sample type: Arterial
T: 37.0 °C
FO2(I): 21.0 %

Blood Gas Values			
↓ pH	7.301		[7.350 - 7.450]
pCO2	34.0	mmHg	[32.0 - 48.0]
↑ pO2	266	mmHg	[83.0 - 108]
Electrolyte Values			
↓ cNa+	132	mmol/L	[136 - 145]
cK+	3.6	mmol/L	[3.4 - 4.5]
cCl-	102	mmol/L	[98 - 107]
↓ cCa++	0.82	mmol/L	[1.15 - 1.33]
Metabolite Values			
↑ cGlu	132	mg/dL	[65 - 95]
cLac	1.1	mmol/L	[0.4 - 1.5]
Oximetry Values			
↓ ctHb	7.1	g/dL	[11.0 - 17.0]
↑ sO2	99.7	%	[94.0 - 99.0]
HctC	22.1	%	
Calculated Values			
AnionGapC	14.3	mmol/L	
cHCO3-(P,st)c	17.2	mmol/L	
cHCO3-(P)c	16.3	mmol/L	
cBase(B)c	-8.8	mmol/L	
cBase(Ecf)c	-8.9	mmol/L	
ctCO2(B)c	35.8	Vol%	
ctCO2(P)c	38.8	Vol%	
pO2(A)e	105.6	mmHg	
pO2(A-a)e	...	mmHg	
pO2(a/A)e	242.8	%	
ABEc	-8.8	mmol/L	
SBEc	-8.9	mmol/L	
cBase(B,ox)c	-8.9	mmol/L	
cBase(Ecf,ox)c	-8.9	mmol/L	

Notes
↑ Value(s) above reference range
↓ Value(s) below reference range
c Calculated value(s)
e Estimated value(s)

Figure 3: Arterial blood gas analysis showing a pH of 7.30, $PaCO_2$ of 34.0 mm Hg, HCO_3 of 17.2 mmol/L, with a base deficit of 8.9, lactate levels of mmol/L, and normal electrolyte and glucose levels, indicating mild metabolic acidosis without lactic acidosis.



KIMS HOSPITAL

Pt Name
MRN/Ind. No
Dr Name

ULTRASONOGRAPHY OF SCROTUM

FINDINGS:

Right:

Testis: Noted in right inguinal canal, with normal echotexture. No focal lesions.

Epididymis: Normal in size and echotexture.

No free fluid in right scrotal cavity.

No e/o varicocele.

Empty scrotal sac noted.

Left:

Testis: Noted in left inguinal canal, with normal echotexture. No focal lesions.

Epididymis: Normal in size and echotexture.

No free fluid in left scrotal cavity.

No e/o varicocele.

Empty scrotal sac noted.

IMPRESSION:

➢ Bilateral ectopic testis.

Suggested clinical, lab correlation follow up and if there is a need kindly discuss.

KIMS Hospitals Kurnool Pvt. Ltd.

SECUNDERABAD | KONDAPUR
NELLORE | RAJAHMUNDRY | SRIKAKULAM | ONGOLE | VIZAG | ANANTAPUR | KURNOOL

Figure 2: Ultrasound scan image of the scrotum showing both the testes locate within their respective inguinal canals, confirming the diagnosis of Bilateral undescended testes.

marked by metabolic acidosis and electrolyte disturbances. Hyperkalemia, in particular, has been associated with life-threatening arrythmias in these patients, as seen in a documented case of severe arrythmias following the induction of anaesthesia due to hyperkalemia [2].

Certain drugs should be avoided in MMA patients, particularly Propofol, which is controversial due to the risk of Propofol Infusion Syndrome (PRIS), a condition characterized by metabolic acidosis, rhabdomyolysis and cardiac failure. While some suggest that an induction dose is not harmful, majortiy advise caution [3].

Succinylcholine, a depolarizing muscle relaxant, is also typically avoided in these patients due to its potential to cause hyperkalemia,

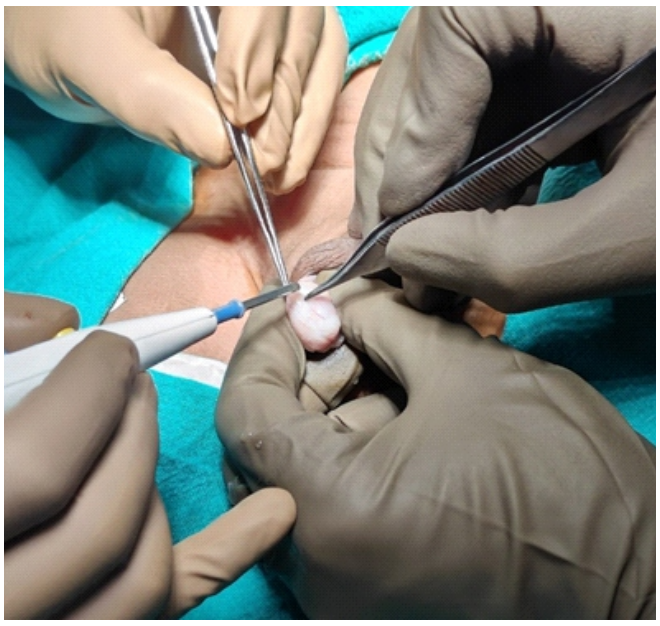


Figure 4: Intraoperative image showing the surgical procedure of placing the testis into the scrotum during orchidopexy.

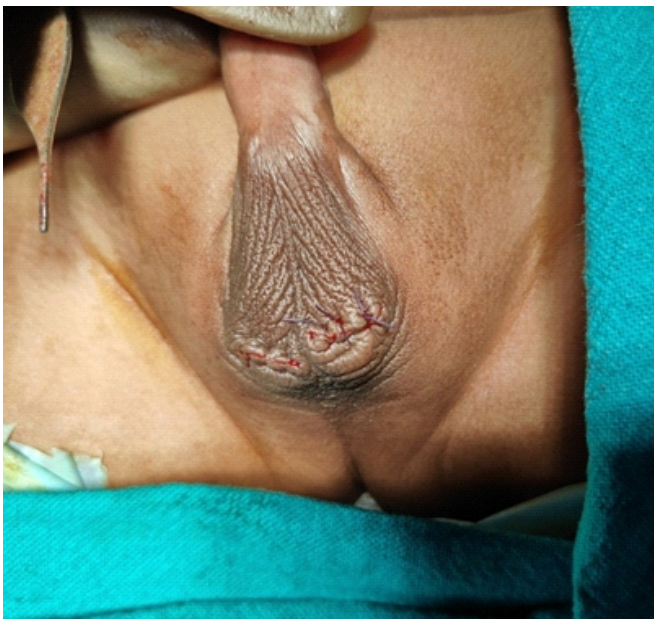


Figure 5: Post operative image showing both the testes successfully placed in their respective scrotum after completion of bilateral orchidopexy.

exacerbating the metabolic disturbance already present in these patients [4].

Preoperative administration of L-carnitine and Cobalamin is essential in the management of these patients. L-carnitine helps by facilitating the removal of toxic organic acids and improving fatty acid metabolism, while cobalamin serves as a co-factor in reducing methylmalonic accumulation in responsive forms of MMA. Their combined use helps stabilize metabolic function before surgery [5].

MMA patients require close peri-operative monitoring to prevent metabolic crises. Blood glucose, acid-base status, and potassium levels must be monitored to avoid hypoglycemia, acidosis and hyperkalemia. Hyperkalemia, though rare, can be fatal, as shown in reports of acute arrythmias caused by severe potassium imbalance in these patients [2, 5].

Conclusion

We report a case of successful anaesthetic management in a child with Methylmalonic acidemia undergoing bilateral orchidopexy. A multidisciplinary approach, careful perioperative monitoring, an appropriate metabolic management were key to preventing complications an ensuring smooth recovery.

Clinical message

Methylmalonic acidemia presents significant anaesthetic challenges due to the risk of metabolic decompensation. Key aspects of management include avoiding certain drugs like Propofol and Succinylcholine, vigilant monitoring for acidosis and electrolyte imbalances, and administering L-carnitine and Cobalamin preoperatively. A multidisciplinary approach involving anaesthesiologists and pediatricians, is crucial to prevent life-threatening complications.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given his/her consent for his/her images and other clinical information to be reported in the Journal. The patient understands that his/her name and initials will not be published, and due efforts will be made to conceal his/her identity, but anonymity cannot be guaranteed.

Conflict of interest: Nil **Source of support:** None

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