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ELECTROLYTE MANAGEMENT AND MONITORING TO PREVENT COMPLICATIONS MMA: A CASE REPORT

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ABSTRACT
Methylmalonic acidemia (MMA) is a rare metabolic disorder caused by the body's inability to break down certain proteins and fats, leading to a buildup of toxic methylmalonic acid. MMA symptoms vary and can cause serious complications if not treated appropriately. Purpose : to explanation of electrolyte management and monitoring to prevent complications on patient with methylmalonic acidemia. Method : this study used case study. Collecting data used nursing assessment. An 8 year old patient with MMA, cough accompanied by vomiting, decreased appetite, and dehydration came to the emergency room in critical condition. The patient showed signs of dehydration, metabolic acidosis, and electrolyte disturbances that required immediate treatment. Results: Management of MMA involves adequate hydration, management of hypoglycemia, correction of metabolic acidosis, and careful monitoring of electrolytes. Additional therapies such as L-Carnitine supplementation and intensive clinical monitoring are also necessary to prevent life-threatening complications. Conclusion: Rapid and appropriate treatment of the metabolic crisis in MMA is essential to prevent serious complications and improve the patient's prognosis. Regular monitoring of electrolytes and clinical parameters as well as comprehensive therapy can help improve the quality of life of individuals with MMA.

Keywords: electrolyte management; metabolic crisis; methylmalonic acidemia; monitoring

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INTRODUCTION

Methylmalonic acidemia (MMA) is a rare hereditary metabolic disorder characterized by the body's inability to properly break down certain proteins and fats (Gupta et al., 2023). This results in a buildup of toxic levels of methylmalonic acid in the blood and tissues. MMA is usually caused by a deficiency of the enzyme methylmalonyl-CoA mutase (MCM), a defect in the transport or synthesis of its coenzyme adenosyl-cobalamin, or a deficiency of the enzyme methylmalonyl-CoA epimerase (Baumgartner et al., 2014).

Symptoms of methylmalonic acidemia can vary from vomiting, dehydration, hypotonia, developmental delays, excessive fatigue, to failure to grow and develop at the expected rate (Sowa, 2022). Long-term complications can include eating problems, intellectual impairment, movement problems, chronic kidney disease, and inflammation of the pancreas. Without treatment, this disorder can cause coma and death in some cases (R.-Y. Chen et al., 2020). MMA usually appears in infancy or childhood, although there are cases where symptoms may

appear in the neonatal period or in adulthood (Molema et al., 2021). MMA symptoms in infants usually appear in the first few months or years of life, whereas in atypical and "benign" or adult cases, symptoms may appear at an older age (Zhang et al., 2020).

In MMA incidence varies across different populations. Estimated incidence in Western populations ranges from 1:48,000 to 1:61,000 births for MMA. In mainland China, the incidence of MMA is approximately 1:4,000 to 1:26,000 births. Patients with MMA experience a significant mortality rate, and the prognosis for long-term survival is low (Kang et al., 2020). While the exact incidence of MMA in Saudi Arabia in live births is unknown; however, newborn screening results indicate that 1 in every 12,178 living newborns may be affected by this disease and most of it is caused by mutase deficiency (Collado, 2020).

MMA can affect various body systems, including metabolic disorders, where patients with MMA will experience something called a metabolic crisis. Some early symptoms of a metabolic crisis include decreased appetite, vomiting, extreme drowsiness or lack of energy and decreased muscle tone (Yang et al., 2020). Other findings in the blood and urine include high levels of acidic substances in the blood called metabolic acidosis, high blood ammonia levels, high glycine levels in the blood and urine, high levels of methylmalonic acid and propionic acid along with high levels of other dangerous substances (T. Chen et al., 2021).

MMA is a rare and serious metabolic disorder, which can cause metabolic crises and severe electrolyte imbalances (Molema et al., 2021). Metabolic crisis in MMA often requires prompt and appropriate management to prevent life-threatening complications (Armstrong, 2021). Hence its importanceelectrolyte management and monitoring to prevent complicationsfrom MMA regarding the diversity of symptoms, early diagnosis, therapeutic management as well as education and awareness about the complications of MMA. The purpose of this study is to explanation of electrolyte management and monitoring to prevent complications on patient with methylmalonic acidemia

METHOD

This study employed a case study method focused on a single patient diagnosed with MMA. This approach was chosen to provide an in-depth analysis of electrolyte management and monitoring to prevent complications associated with this condition. Data were collected by conducting assessments on the patient with Methylmalonic Acidemia. The data analysis was carried out descriptively with the aim of identifying patterns of electrolyte changes and linking them to the medical interventions taken. Additionally, a literature review was conducted to compare the findings of this study with similar cases previously reported. The results of this case study were expected to provide practical insights into effective electrolyte management strategies and potentially reduce the risk of complications in patients with MMA.

The data collection process for this study was conducted through a detailed and systematic approach focusing on a single patient diagnosed with Methylmalonic Acidemia (MMA). First, the patient's medical history was comprehensively gathered, including medical records, family history, and previous diagnostic test results. Next, direct clinical observations and routine monitoring were conducted to track the patient's vital signs and physical condition. Laboratory tests were performed to measure key biochemical parameters such as electrolytes, urea, creatinine, glucose, ammonia, lipase, blood gases, and complete blood count (CBC) with differential. Urine samples were also analyzed for ketones and specific gravity to assess metabolic status. Additionally, the patient's response to various treatments and interventions

was meticulously documented, including the administration of medications, nutritional support, and instances of metabolic crises. This extensive data collection process enabled a thorough analysis of the patient's condition, treatment outcomes, and the effectiveness of electrolyte management strategies in preventing complications associated with MMA.

RESULTS

The client came to the ER on 03/19/2024 An. R, 8 years old, Saudi Arabia with a diagnosis of MMA, Gall bladder stone, stage III CKD, vitamin D deficiency, iron deficiency anemia, the patient was installed with a port a cath, and a gastrostomy tube, with complaints of cough accompanied by vomiting after coughing, decreased appetite and decreased oral intake. Conscious awareness, GCS 14/15, there is an increase in respiratory rate (Kussmaul) breathing, alert but lethargic, visible dehydration, sunken eyeballs, prolonged CRT more than 4 seconds, tachycardia, tachypnea, ABG results, PH: 7.36, PCO2: 27.9, HCO3:15.9, RBS :121, ammonia 150 mmol/dl. Results other laboratories: Na 139, Chloride 100, Potassium (K) 5.6; Lactate 5.7, creatinine 140.5, urea 25.7, BUN 0.2, leukocytes 4800 and GFR 30 mg/dl. Judging from the signs and symptoms, as well as the results of blood gas, ammonia and potassium analysis, the patient is experiencing a metabolic crisis that requires immediate treatment. The therapy received (according to the MMA protocol) the patient was given IVF Normal Saline bolus 20 cc/kg/ 30 min, and continued with IVF D10% N2 maintenance 85 ml/day, IV NaHCO3 15 mmol/ 8 hours, Omeprazole 17 mg IV, IV L carnitine 800 mg/8 hours, patient instructed NPO, ondansetron 2 mg IV. After 24 hours of treatment, the client's metabolic acidosis can be corrected. With the results of blood gas analysis PH: 7.35, PCO2: 40, PO2: 35, HCO3: 20, ammonia: 80 mmol/dl, Lactate: 2.7, Potassium: 4.7.

DISCUSSION

MMA is a rare genetic disease typically manifesting in early infancy, often becoming apparent by the second or third day of life. Initial symptoms include lethargy, anorexia, vomiting, and dehydration, progressing to potentially life-threatening complications such as seizures, coma, and even death. Clinical features commonly include metabolic acidosis, ketosis, lactic acidosis, and hyperammonemia (Molema et al., 2021). Neurological deficits, including psychomotor retardation, cognitive impairment, movement disorders, and epilepsy, are common among affected individuals (Armstrong, 2021). In a recent case, MMA was diagnosed in a one-week-old infant who presented with an inability to breastfeed. The diagnosis was confirmed through a series of metabolic disorder examinations during hospitalization in the Neonatal Intensive Care Unit (NICU) (Dao et al., 2021). The patient underwent routine treatment due to frequent disease relapses, necessitating the installation of a catheter port for intravenous (IV) access at the age of four and a gastrostomy tube for nutritional access at the age of two (Molema et al., 2021).

Clinical monitoring in MMA cases, as outlined in the MMA Guidelines from King Faisal Hospital, Saudi Arabia, involves a thorough assessment covering various aspects of patient health. This monitoring protocol encompasses evaluating mental status using the Glasgow Coma Score to assess neurological function and monitoring vital signs, with a particular focus on blood pressure (BP) and temperature, to detect any signs of physiological distress (Luciani et al., 2021). Additionally, fluid balance is assessed to ensure adequate hydration status and prevent dehydration. The protocol includes assessing for evidence of bleeding, especially in cases with low platelet levels, and monitoring for symptoms of infection, particularly in neutropenic patients, to promptly identify and manage any infectious processes (Brox-Torrecilla et al., 2021; Yu et al., 2021).

Regular monitoring of biochemical parameters such as electrolytes (sodium, potassium, chloride, bicarbonate, calcium, phosphate, and magnesium), urea, creatinine, glucose, and ammonia levels is conducted to evaluate renal and metabolic function (Sowa, 2022). Lipase levels are also monitored to screen for pancreatic involvement. Blood gas analysis is performed to evaluate acid-base status and respiratory function, while a complete blood count (CBC) with differential helps assess for any abnormalities in blood cell counts (T. Han et al., 2022). Urine testing for ketones and specific gravity is conducted to monitor metabolic status and hydration levels. This comprehensive monitoring approach allows healthcare providers to promptly detect and address any complications or changes in the patient's condition, ensuring optimal management of MMA and minimizing the risk of adverse outcomes (Wang et al., 2022).

Despite typical presentations, this case presented an uncommon feature of hyperkalemia (potassium 5.6), contrary to conventional expectations where hyperkalemia is not a common feature of MMA except during metabolic decompensation (Gupta et al., 2023). Additionally, the patient's condition was complicated by gallbladder stones and stage III kidney failure, exacerbating metabolic acidosis. Effective management of MMA requires early diagnosis and prompt treatment initiation, with regular monitoring playing a crucial role in improving patient outcomes (T. Chen et al., 2023). Newborn screening, utilizing biomarkers such as Propionyl Carnitine (C3), has proven instrumental in facilitating early detection, particularly in late-onset cases (Head et al., 2023). Specialized care in a specialist center, along with interventions tailored to the patient's metabolic needs, is essential for preventing metabolic crises and minimizing associated morbidity and mortality (Hakimzadeh et al., 2024). Electrolyte imbalances, including hyperkalemia, often accompany metabolic decompensation in MMA patients and require careful management (Yuan et al., 2024). Hyperlactatemia, a common feature of MMA, contributes to metabolic acidosis, exacerbating renal dysfunction in cases of kidney failure. Metabolic crises demand immediate interventions, including hydration, glucose supplementation, correction of metabolic acidosis with sodium bicarbonate, and, in severe cases, dialysis (Arhip et al., 2024)

During the recovery phase, several interventions are employed to support the patient's recovery from metabolic crises. Firstly, broad-spectrum antibiotics are administered to expedite recovery from acute crises. In this case, the patient received intravenous ceftriaxone 800mg (Bayat et al., 2024). To facilitate recovery, the patient is advised to remain NPO (nil per os) until improvements in mental status are observed. If significant neurological impairment is absent, enteral feeding via nasogastric (NG) or gastrostomy tube (GT) is initiated using a patient-specific formula excluding offending amino acids (Fraser & Venditti, 2016). This approach ensures a higher energy intake and reduces the risk of malnutrition. In instances where enteral feeding cannot be initiated within 48 hours, early initiation of total parenteral nutrition (TPN) is recommended to prevent malnutrition (Gupta et al., 2023). It's important to note that while on TPN, only moderate protein restriction is necessary, and consultation with a team of metabolic specialists is advised (Sowa, 2022). Patients receive specialized formulas from dieticians and metabolic clinics tailored to their individual needs (Yu et al., 2021).

Long-term management involves nutritional support, with specific attention to glucose and protein intake, often supplemented with intravenous L-carnitine. Recovery from metabolic crises necessitates antibiotic therapy, enteral feeding, and close monitoring of clinical parameters. Patient education, genetic counseling, and regular follow-up care, including speech and language therapy, are vital components of comprehensive MMA management.

Education for patients and caregivers encompasses various aspects crucial for managing MMA effectively. Firstly, guidance on dietary practices, including food and drink preparation, is provided based on recommendations from a metabolic consultant (Zhou et al., 2018). Understanding the potential side effects of medications is essential for ensuring safe and appropriate use. Moreover, education on the proper care and management of a nasogastric tube (NGT) is imparted, particularly pertinent for patients who have undergone gastrostomy tube installation (He et al., 2021) Recognizing signs and symptoms of disease exacerbation and knowing when to seek immediate medical attention is emphasized to prevent complications (Zhou et al., 2018). Follow-up care is emphasized, involving a multidisciplinary approach. Patients are encouraged to attend metabolic clinics for comprehensive support from dietetic, medical, and nursing professionals (Wang et al., 2022). Regular blood tests, including assessments of amino acids, are conducted during outpatient department (OPD) visits. Additionally, urine samples are typically checked monthly to monitor metabolic status (Baker, 2015).

Vitamin B12 plays an important role in the human body's metabolism, especially in the process of DNA formation, red blood cell synthesis, and nervous system function as well as maintaining electrolyte balance in the body (B. Han et al., 2016). In the context of patients with high levels of methylmalonic acid (MMA), vitamin B12 has a crucial role due to its association with amino acid and fatty acid metabolism. Amino Acid Metabolism where Vitamin B12 is needed for the conversion of methylmalonyl-CoA to succinyl-CoA via the enzyme L-methylmalonyl-CoA mutase (Yu et al., 2021). In vitamin B12 deficiency, the activity of this enzyme is impaired, leading to increased MMA production. Therefore, high MMA levels can be an indicator of vitamin B12 deficiency in later patients (Collado, 2020). In addition, vitamin B12 is also involved in fatty acid metabolism, especially in the formation of odd-chain fatty acids and branched amino acids (Fraser & Venditti, 2016). Because MMA is an intermediate in the metabolism of propionic acid, which is produced from odd-chain fatty acid metabolism, MMA levels can also reflect impaired fatty acid metabolic activity (Peng et al., 2019).

CONCLUSION

Methylmalonic Acidemia (MMA) is a rare hereditary metabolic disorder that can cause various serious complications such as metabolic acidosis, vitamin D deficiency, anemia and neurological disorders. Management of MMA includes various aspects such as adequate hydration, treating hypoglycemia, treating metabolic acidosis by administering sodium bicarbonate, and providing special nutrition that avoids certain amino acids. Regular monitoring of electrolytes and clinical parameters is essential to identify and prevent complications that may arise from MMA. Additional therapy such as administration of L-Carnitine, antibiotics, dialysis, and careful clinical monitoring are also needed in the treatment of MMA. With proper monitoring and treatment, it is hoped that it can reduce the risk of complications and improve the quality of life for individuals suffering from Methylmalonic Acidemia.

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