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TRANSIENT NEONATAL DIABETES MELLITUS WITH MODERATE DIABETIC KETOACIDOSIS (DKA)-A CASE REPORT

KEY WORDS: Neonatal Diabetes Mellitus

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ABSTRACT

Background: Neonatal Diabetes Mellitus (NDM) is a rare monogenic disorder of glucose metabolism presenting within the first six months of life. It can be either transient (TNDM) or permanent (PNDM). TNDM usually remits by 12–18 months but may recur later in life. Presentation with diabetic ketoacidosis (DKA) in neonates is uncommon and poses significant diagnostic and therapeutic challenges. **Case Presentation:** We report the case of a 1½-month-old female infant, born at term to consanguineous parents, who presented with fever, respiratory distress, and poor feeding. Laboratory investigations revealed severe metabolic acidosis (pH 7.16, HCO_3^- 5.1 mmol/L), hyperglycaemia (RBS 278 mg/dL), and strongly positive urine ketones, consistent with moderate DKA. She was managed with intravenous fluids, insulin infusion, and antibiotics for possible sepsis. The infant had a prior history of transient hypothyroidism and macroglossia, suggesting a syndromic association. The child improved with therapy, and the clinical picture was consistent with Transient Neonatal Diabetes Mellitus (TNDM) presenting atypically with moderate DKA. **Conclusion:** Early identification of TNDM is vital to prevent life-threatening complications such as DKA. In neonates with unexplained hyperglycaemia and dehydration, NDM should be strongly suspected. Genetic evaluation and long-term endocrinologic follow-up are essential for appropriate management and prognosis.

INTRODUCTION

Neonatal Diabetes Mellitus (NDM) is a rare form of monogenic diabetes that presents within the first six months of life, with an estimated incidence of 1 in 90,000 to 160,000 live births[1]. It results from genetic defects affecting pancreatic -cell development, insulin secretion, or regulation. NDM is broadly classified into Transient Neonatal Diabetes Mellitus (TNDM) and Permanent Neonatal Diabetes Mellitus (PNDM), depending on whether insulin dependence resolves or persists beyond infancy[2]. TNDM accounts for approximately 50–60% of all neonatal diabetes cases and is characterized by intrauterine growth retardation, hyperglycemia appearing within days to weeks after birth, and spontaneous remission typically by 12 to 18 months of age[3]. However, relapse can occur later in childhood or adolescence, often presenting as type 2–like diabetes. Genetic and epigenetic abnormalities, most notably involving chromosome 6q24, or activating mutations in KCNJ11 and ABCC8 genes encoding ATP-sensitive potassium (KATP) channel subunits, underlie most cases of TNDM . Clinical manifestations range from mild hyperglycaemia to life-threatening diabetic ketoacidosis (DKA), though DKA presentation in neonates remains rare due to partial insulin activity. Early diagnosis and appropriate insulin management are essential to prevent metabolic complications and ensure optimal growth and neurodevelopmental outcomes.

Case Description

A 1½-month-old female infant, the second child of third-degree consanguineous parents, presented with fever for three days, cough and cold for two days, difficulty in breathing since the day of admission, and refusal to feed. She was born at term by normal vaginal delivery with a birth weight of 2.2 kg and had no perinatal complications. There was no family history of diabetes or congenital anomalies. During the neonatal period, the child had been admitted for evaluation of macroglossia, where she was diagnosed with transient hypothyroidism and started on Thyronorm 25 µg once daily. At six days of life, she was again admitted with respiratory distress and fever; investigations revealed C-reactive protein 18.5 mg/L, hemoglobin 15.3 g/dL, total leukocyte count 14,900/mm³, platelet count 1.28 lakh/mm³, blood urea 56 mg/dL, and serum potassium 7.2 mmol/L. She was diagnosed with transient hyperglycemia and transient hypothyroidism with dysmorphic features and was managed conservatively before discharge.

During the current admission, the infant appeared febrile, tachypneic, tachycardic, drowsy, and dehydrated, with signs of pallor, lymphadenopathy, and hepatomegaly. Arterial blood gas analysis revealed pH 7.16, HCO_3^- 5.1 mmol/L, PCO_2 14 mmHg, and PO_2 182 mmHg, indicating severe metabolic acidosis. Her random blood sugar was 278 mg/dL, and urine ketones were strongly positive, confirming moderate diabetic ketoacidosis (DKA). A concurrent possibility of sepsis was considered based on her clinical presentation.

The infant was managed with intravenous fluids (normal saline at 12 cc/hour), broad-spectrum antibiotics (Cefotaxime and Amikacin), regular insulin infusion via syringe pump (1.25 mL/hour in sterile water), and supportive care with close monitoring of electrolytes, glucose, and hydration status. She gradually improved and was stabilized over the following days. The constellation of hyperglycemia, DKA, transient hypothyroidism, and macroglossia was consistent with Transient Neonatal Diabetes Mellitus (TNDM) presenting unusually with moderate DKA, warranting further endocrinologic and genetic evaluation for 6q24-related abnormalities and family counselling.

DISCUSSION

Transient neonatal diabetes mellitus (TNDM) is an uncommon cause of hyperglycaemia in early infancy, usually appearing within the first six weeks of life and remitting by 12–18 months. The pathophysiology primarily involves abnormal expression or methylation defects of imprinted genes on chromosome 6q24, or activating mutations in KCNJ11 and ABCC8, which encode the KATP channel subunits Kir6.2 and SUR1, respectively. These genetic abnormalities lead to pancreatic β-cell dysfunction and impaired insulin secretion. Unlike permanent neonatal diabetes mellitus (PNDM), β-cell function in TNDM tends to recover as epigenetic or metabolic regulation normalizes with age.[5-6]

Presentation with diabetic ketoacidosis (DKA) in TNDM, as in this case, is rare due to residual insulin secretion in most affected neonates. However, intercurrent infections, dehydration, or delayed diagnosis can precipitate metabolic decompensation. The coexistence of macroglossia, transient hypothyroidism, and dysmorphic features further suggests 6q24-related imprinting abnormalities. Management requires meticulous correction of dehydration, gradual normalization of blood glucose through insulin infusion, and avoidance of rapid osmotic shifts. Long-term follow-up is vital, as up to 50% of TNDM cases may relapse later in adolescence or adulthood as type 2–like diabetes. Genetic counselling of the family is recommended to guide prognosis and recurrence risk.[7-8]

CONCLUSION

Early recognition of transient neonatal diabetes mellitus is critical to prevent complications such as DKA. In neonates presenting with unexplained hyperglycaemia, DKA, and failure to thrive, a high index of suspicion for NDM is essential. Genetic workup and follow-up are recommended for long-term management.

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