Case Report

Neonatal diabetes – A rare case

S Y Ingale[1], Jaiom Dagar[2*], C D Aundhakar[3]

[1,2,3,4,5] Department of Paediatrics, Krishna Institute of Medical Sciences, Karad, Maharashtra, INDIA.
Email: jai.dagar05@gmail.com

Abstract

Context: Neonatal diabetes is a rare endocrinal disorder with an incidence of about 1 in 100,000 live births. It is defined as diabetes diagnosed in the first 6 months of life and is categorized into permanent neonatal diabetes and transient neonatal diabetes**. Case report:** We present a case of neonatal diabetes due to a mutation in KATP channel which revealed a heterozygous mutation in exon 21 of *ABCC8*. **Conclusion:** An infant who presents with nonspecific symptoms that cannot be attributed to other illnesses should have a simple urine test for glucose. The genetic diagnosis has implications on treatment as patients can switch from insulin therapy to oral hypoglycaemic drugs.
Keywords: Insulin-dependent diabetes mellitus, infancy, diabetic ketoacidosis, management, diagnosis.

[*]**Address for Correspondence:**
Dr. Jaiom Dagar, Resident, Department of Paediatrics, KIMS, Karad, District-Satara-415110, Maharashtra, INDIA.
Email: jai.dagar05@gmail.com
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INTRODUCTION

Neonatal diabetes mellitus (NDM) is a form of insulin requiring monogenic diabetes characterised by onset of hyperglycaemia within in the first 6 months of life and is usually of genetic origin. Insulin-dependent diabetes mellitus (IDDM) is the second most common chronic disease in childhood.[1]The peak ages of onset are between 7 and 9 years and 11 and 13 years.[1]Although presentation of diabetes in an infant under the age of 1 is rare, it does occur. Estimates of prevalence range from 0.5% to 1% of all children with IDDM.[2] Early recognition of diabetes in this age group can prevent the high morbidity and mortality associated with this disease.[3] Diabetes in an infant is often misdiagnosed initially because the symptoms of vomiting and dehydration are often attributed to a viral illness. An infant is more likely to present in diabetic ketoacidosis (DKA) and coma than an older child. This increases the mortality associated with diabetes by 50% in childhood.[3] we present a case of infant with type 1 diabetes manifested as diabetic ketoacidosis.

CASE REPORT

A 4 month old male infant, 1st issue of 2nd degree consanguineous marriage, was referred to our hospital with severe respiratory distress since last 3 days, for need of mechanical ventilation. He also had 1 episode of tonic seizure associated with unrolling of eyes. Past medical history and family history were unremarkable. Father is 28 yr old with no history related to DM and HT. Birth history- birth weight 2.59 kgs, delivered by L.S.C.S for severe oligohydroamnios to a 32 yrs old gravid 3 mother, with no significant antenatal and perinatal history. Physical examination revealed - a sick baby, gasping respiration, vitals – HR 172/min, SPO_2 80% with 3 litre/min of oxygen. Patient was intubated and put on SIMV mode of mechanical ventilation, The following report were available : urea 154 mg/dl, creatinine 1.8 mg/dl, Na^+ 156 m eq/L, K^+ 6.3 meq/L, Hb 12.1 gm%, WBC 21,130/cu mm, Neutrophils 90% and Lymphocytes 10%, platelet 1.50 lakh/cumm, bsl >500 mg/dl, urine: sugar 4+, acetone 2+, albumin 3+, reducing substances 3+, serum ammonia 25. ABG showed severe metabolic acidosis. He was put on IV H.INSULIN (short acting) drip and IV antibiotics. Neurodevelopment examination was normal. Glycated haemoglobin value is 15.6%. (Reference range: 4.0-5.9%). Structural diseases of pancreas were ruled out by ultrasound of abdomen. Glutamic acid decarboxylase-65 antibody (GAD-65) and insulinoma-associated antibody (IA-2) were negative; the fasting C-peptide was 0.90ng/mL, (reference range: 0.8-

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3.8 ng/mL). Genetic tests for mutation in KATP channel were carried out. This revealed a heterozygous mutation in exon 21 of *ABCC8*.

DISCUSSION

Diabetes is a global health and development crisis. Incidence of NDM particularly in Indian population is not studied so far but a number of case studies have been published from India [4]. This condition can be either transient (TNDM), with a period of remission or permanent (PNDM), requiring lifelong treatment. Infants suffering from diabetes mellitus present with signs and symptoms confused with other more common illnesses in this age group. A physician examining an ill appearing dehydrated infant, without any obvious cause for the dehydration, should quickly screen the urine for glucose and ketones. Diagnosis of diabetes is a problem when an infant has only hyperglycemia or ketonuria. Febrile illnesses, convulsions, and dehydration can cause this laboratory abnormalities[5], even in developed countries, some 15–70% of all newly diagnosed infants and children with diabetes present with DKA.[6] In DKA, the blood glucose is more than 300 mg/dL, serum bicarbonate is < 15 mEq/dL, ketonuria is present, and pH is <7.3. A number of molecular causes are responsible for NDM, of which, mutations in the genes (*KCNJ11* and *ABCC8*) encoding the potassium ATP channel (KATP) are a major cause, less frequent causes of NDM includes mutations in the genes namely *HNF4A, PTF1A, HNF1B, SLC2A2, GATA6, FOXP3* and *RFX6* (15-20). Very rarely insulin resistance syndromes like Leprechaunism, Rabson-Mendenhall Syndrome also present with diabetes in the infancy. Management of neonatal diabetes is very challenging. Initial management of the infant with diabetes depends on whether DKA is present. Children with mutations of *KCNJ11* and *ABCC8* who are responsive to sulphonylurea have a good metabolic control and show improvement in the neurological impairment.[7]This switching over to sulphonylurea can be undertaken in such children irrespective of the chronological age. The change over to sulphonylurea can be done as a rapid in patient procedure over a 2 weeks or it can be done slowly over a period of 28 days. The Children who are suffering from monogenic diabetes and not responding to sulphonylurea will be treated with insulin injections. Insulin requirements for an infant are generally between 0.5 and 0.8 units/kg/day. The metabolic control can be achieved by using one or two doses of intermediate acting insulin per day. Neonatal diabetes can also be treated by insulin pump therapy. Other than metabolic control these children need to be monitored for growth and development.[8] associated comorbid states needs to be addressed. Avoiding hypoglycemia in these infants is very essential as the growing brain is more vulnerable for sequel due to hypoglycaemia. Parents should be educated to check blood glucose and urinary ketones more frequently during any illness. Any physician caring for children should have a high index of suspicion to diagnose diabetes in infancy. An infant who presents with nonspecific symptoms that cannot be attributed to other illnesses should have a simple urine test for glucose.

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