

TRANSIENT NEONATAL DIABETES MELLITUS: New Case

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Introduction:

Neonatal diabetes mellitus (NDM) is a monogenic form of diabetes that occurs in the first 6 months-of-age. It is a rare condition occurring in only one in 100,000-300,000 live births. Clinically, NDM subgroups include transient (TNDM) and permanent NDM (PNDM). TNDM is most frequently caused by abnormalities in the imprinted region of chromosome 6q24.

Case Report:

A 18-day-old male was referred from another clinic due to diabetic ketoacidosis (DKA). The patient was born to healthy first-degree cousins at 38 weeks of gestation with a birth weight of 2500 g and birth length of 42 cm. Physical examination did not reveal any dysmorphic features. He appeared extremely dehydrated, tachypneic and lethargic. Laboratory investigations revealed ketonuria, acidosis (pH: 7.02, HCO₃: 2.9 mmol/L) and hyperglycemia (plasma glucose 828 mg/dL). The patient was then hydrated with intravenous fluids and treated with an insulin and sodium bicarbonate. His serum c-peptide was 0.15 ng/dL (normal range, 0.9-7.1), HbA1c was 6.7% (normal range, 4-6%). Anti-GAD and anti-insulin antibodies were negative. Abdominal ultrasonography demonstrated a normal pancreas anatomy. After hydration therapy intravenous insulin infusion changed to subcutaneous neutral protamine Hagedorn (NPH). At the age of 5 months, the patient entered remission at which stage insulin treatment was withdrawn. The patient is currently 11-months-of-age. His HbA1c is 4.7% and his growth and physical development are normal. *ABCC8*, *KCNJ11*, *INS*, and *EIF2AK3* genes were sequenced and no mutations were detected. In addition to these, a novel mutation determined on chromosome 6q24 due to paternally duplication, confirming a diagnosis of TNDM.

Conclusion:

A total 70% of TNDM is caused by defects causing overexpression of paternally expressed genes in the imprinted region of chromosome 6q24. Correctly identifying monogenic NDM is important for facilitating accurate diagnosis, appropriate therapy and genetic testing for at risk family members.