The Case of Transient Neonatal Diabetes Mellitus Associated with 6q24

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The most common form of transient neonatal diabetes mellitus (TNDM) is 6q24-associated TNDM. It is seen in the first 2 months of life, lasts more than 2 weeks, and mostly requires an insulin treatment.

A girl baby was admitted to the newborn intensive care unit on her first day of life since her blood glucose level was 350 mg/dL and due to “lack of sucking”. The baby was born to a 22-year-old healthy mother at 35th week of gestational age with birth weight of 2025 g (-3.75 SDS). The patient does not have any family history of diabetes; the parents are not relatives. On her physical examination, there was not any pathology other than being small for gestational age and having macroglossia. When serum blood glucose level was 380 mg/dL, blood ketone was negative, serum insulin level was <2 mIU/mL, and C-peptide was 0.408 ng/mL. Regarding the differential diagnosis, metabolic scanning, TORCH, and sepsis examinations were done and they were all normal. On her second day of life, an insulin infusion of 0.01 U/kg/hour was started. On follow-up, the insulin infusion was given with a gradually increasing rate, and on her 4th day of life, she was provided with an insulin infusion pump which has a continuous glucose measurement system. Normoglycemia was attained by basal rates settled with respect to the glucose levels. In terms of persistent neonatal diabetes, genetic analysis of ABCC8, KCNJ11, INS, and EIF2AK3 were normal. When she was 20 days old, insulin therapy was stopped since normoglycemia was attained. There was not any hyperglycemia in the follow-up. 6q24 was investigated in terms of transient neonatal diabetes and paternal uniparental isodisomy was detected due to a maternal mutation loss. Regarding the probable renal pathologies which may accompany transient neonatal diabetes, a renal ultrasound imaging was performed. On renal ultrasonography, bilateral grade 1 hydronephrosis was found. Cardiac examination revealed peripheral pulmonary stenosis of 12 mmHg gradient and the patient was taken into a follow-up program. The case had a mild hyperglycemia (capillary glucose 120-175 mg/dL) during an episode of gastroenteritis with fever at age 4 months and she is still being followed without treatment.

Although seen really rarely, TNDM is important in terms of not requiring therapy, having possible accompanying cardiac, renal pathologies, and having tendency to recur in late life.

Key words: Transient neonatal diabetes, 6q24