A Rare Case of Neonatal Diabetes due to *COQ2* Gene Mutation

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Introduction

Neonatal diabetes is a rare form of diabetes presenting within the first six months of life. Genetic cause has been found in only 60% to 75% of patients suggesting that continuing research will identify new genes. Coenzyme Q10 is a lipophilic electron carrier located in the inner mitochondrial membrane. Primary coenzyme Q10 deficiency is a rare and clinically heterogeneous respiratory chain disorder considered to be the only treatable mitochondrial disorder, since patients have response to oral coenzyme Q10 supplementation. In infants, the most common phenotype is the severe infantile multisystemic variant with encephalopathy and nephropathy being the most common presentation. In addition, liver, cardiac and pancreatic involvements have been described in the literature.

Case Report

We describe two siblings with neonatal diabetes due to coenzyme Q10 deficiency. Patient 1 was the first child of consanguineous parents delivered at 34-week gestation with oligohydramnios and intrauterine growth restriction. He did not have any dysmorphic features at birth. There was no family history of diabetes, renal or neurological disorders. By the second day of life, hyperglycemia developed and intravenous regular insulin treatment was started with strict blood glucose monitoring. Proteinuria was detected at 24th day of life. He had no clinical evidence of pancreatic exocrine failure and abdominal ultrasonography revealed a structurally normal pancreas. He was diagnosed as neonatal diabetes and was discharged with subcutaneous NPH insulin (0.4 U/kg/d in 4 doses). At the age of 2 months, myoclonic convulsions started. Electroencephalogram showed epileptic discharges on bilateral frontal regions. Total daily insulin dose was decreased to 0.07 U/kg/d with no episode of hypoglycemia. At 4 months of age, homozygous mutation in *COQ2* gene was detected and coenzyme Q10 therapy was initiated. He remained insulin-dependent and died during a convulsive attack following 2 weeks of treatment.

Patient 2, the sister of patient 1, was born at 37 weeks' gestation. During the first day of life, hyperglycemia, respiratory distress, lactic acidosis and proteinuria were developed. Septic parameters were all negative. Insulin infusion with coenzyme Q10 replacement started in advance before the molecular analysis. Respiratory distress and lactic acidosis improved on 3rd day, treatment was switched to subcutaneous NPH insulin with full enteral nutrition. At 6th day of life, insulin treatment was stopped. Congenital nephrotic syndrome also diagnosed, required high protein diet and captopril. A clinical diagnosis of coenzyme Q10 deficiency was confirmed with the same mutation in *COQ2* gene. Currently, she is 2 months old and hyperglycemia did not occur.

Conclusion

COQ2 gene mutation is a very rare cause of neonatal diabetes. Early coenzyme Q10 therapy can treat neonatal diabetes due to the *COQ2* gene mutation.