



CLINICAL MANAGEMENT IN SECONDARY PSEUDOHYPOALDOSTERONISM: A CASE SERIES

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Background:

It is known that PDX-1 gene mutation is related to neonatal diabetes, pancreas agenesis and intrauterine growth retardation. Here the aim was to present a novel defined mutation in PDX-1 gene in case born with IUGR, diagnosed with neonatal diabetes and in which exocrine pancreas deficiency and gallbladder agenesis were detected.

Case Presentation:

Blood glucose was measured as 185mg/dl in the first hour after birth and insulin infusion was given at intervals to the case whose blood sugar was >250mg/dl during observation and the case was referred to our clinic.

In the physical examination of the male baby born with a weight of 1520gr in the 37th week as G1P1 from 19 year old mother with a story of 1°cousin marriage in his parents, the body weight was 1400gr(<3p), height was 42,5cm(<3p) and the head circumference was 31cm(<3p) and they were smaller when pregnancy week was considered. Referral blood sugar was found 216mg/dl and it reached up to 500mg/dl in the follow-ups.

Cholestatic giant celled hepatitis was detected in the liver needle biopsy of patient whose faecal elastase level was >200ng/ml and had direct hyperbilirubinemia. Blood sugar regulation was provided with insulin pump to the case whose satisfactory nutrition couldn't be provided due to exocrine pancreas function disorder and who was experiencing hypoglycemia and hyperglycemia even though 0.2U/kg/day SC insulin dose was given. Gallbladder imaging was impossible in ultrasonography and magnetic resonance cholangiography. A novel homozygote mutation was detected in PDX-1 gene Exon 2 with Whole Exom sequencing method [c.593G>C;p.Arg198Pro(p.R198P)]. It was observed that the parents had heterozygote mutation. The patient was discharged with insulin pump in the postnatal fourth month.

Conclusion:

PDX-1 was detected to play a very significant role including pancreas and islet functions and we think that the new mutation we defined would contribute to the literature.

