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신생아 시기에 생존한 신세뇨관 발생 이상 1례

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A Case of Renal Tubular Dysgenesis Surviving the Neonatal Period

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Renal tubular dysgenesis is a developmental abnormality of the renal proximal tubules characterized by Potter phenotype and severe hypotension in the early neonatal period. Here we report a male infant with renal tubular dysgenesis. Fetal ultrasonography revealed oligohydroamnios and slightly enlarged, hyperechogenic kidneys at 29 weeks of gestation. He was delivered via vagina at 32^{+4} weeks of gestation with a birth weight of 1,960 gm ($10-50^{th}$ percentile). He had a hypocalvaria with widening of suture and fontanelle, hyperflexible joints with high serum alkaline phosphatase level. In addition, generalized renal tubulopathy (polyuria, hyponatremia, decreased renal function (GFR <30 mL/min/1.73m^2)) and severe refractory hypotension were observed. Endocrinologic studies showed a high plasma renin activity (>22.3 ng/ml/hr; normal range 2.0-15.2), low angiotensin-converting enzyme (ACE) concentration (<5 U/L; normal range 8.3-21.4). These findings suggested decreased function of ACE, and the ACE gene analysis revealed compound heterozygous mutations, c.1454delC causing p.Pro(CCT)485Leu(CTT)fs in exon 9 and c.G776A causing p.Arg(CGC)259His(GAG) in exon 5. The former mutation is inherited from his father, and the latter from his mother. There was no mutation in the REN (encoding rennin) gene.

Key Words : 신세뇨관 발생이상, 안지오텐진전환효소 유전자
Renal tubular dysgenesis, ACE gene mutation