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Classic Bartter 증후군의 임상병리학적, 유전적 소견

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Clinical, Pathologic and Genetic Findings of Classic Bartter Syndrome

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Bartter syndrome (BS) I-IV is a rare autosomal recessive disorder of salt reabsorption in the thick ascending limb of Henle's loop. BS is classified according to onset ages or mutation of genes. We report a case of classic BS in a 22-year-old woman who presented with persistent mild proteinuria for two years. She was of short stature (153 cm) and her blood pressure was normal (100/60 mmHg). Her serum potassium was decreased to a level of 2.59 mEq/L with hypokalemic metabolic alkalosis. Plasma renin activity was elevated at 27.98 ng/ml/hr but plasma aldosterone concentration was within normal limit. Twenty-four hour urinary excretion of prostaglandin E increased to 2815 ng/day. To evaluate persistent mild proteinuria and microscopic hematuria, renal biopsy was performed. Most glomeruli were enlarged with marked hyperplasia of the juxtaglomerular cells suggesting BS. Patchy tubular atrophy, interstitial fibrosis with lymphocytic infiltration, and mild to moderate artero- and arteriolosclerosis were seen. Immunofluorscence showed segmental weak granular staining for IgG and IgA and trace for C3 in the mesangium. Electron microscopy revealed a mild expansion of mesangium with rare electron dense deposits. Generic analysis revealed that the patient had both homozygous deletion of exon 1-14 and heterozygous deletion of exon 15-19 of the CLCNKB gene. Patient was confirmed as Batter syndrome, type III. Her father had a heterozygous deletion of exon 1-14 and mother had a heterozygous deletion of all exons of the CLCNKB gene. No renal symptoms or abnormal renal function was noted in parents. For one year before admission, she had been taking for hyperlipidemia at local clinic. Recently, her serum cholesterol, triglyceride and LDL-cholesterol were within upper normal limits. Arterio- and arteriolosclerosis may be related to hyperlipidemia. In conjunction of characteristic clinical, laboratory and pathologic findings, we analyzed genetic alterations in patient and her parents and diagnosed as type III BS. This case shows the importance of the renal biopsy and of molecular analysis in delineating the cause of atypical presentation associated with adult classic BS.

Key Words: 저칼륨혈증, Bartter 증후군
Hypokalemia, Bartter syndrome