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신청소율검사, 유전자검사, 신조직검사를 통해 확진된 Gitelman 증후군의 예

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A Case of Gitelman's Syndrome Diagnosed by Labaratory Test, Renal Clerance Study, Genetic Analysis and Renal Biopsy

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Patients with Gitelman's syndrome present a variety of clinical features, such as hypokalemia, hypomagnesemia, hypocalciuria and metabolic alkalosis. Their ability to concentrate urine is normal to mildly reduced and glomerular filtration rate is within normal range. Pathophysiologically, mutation of the thiazide sensitive Na-Cl cotransporter (NCCT) gene on distal convoluted tubule is the cause of the syndrome. There have been so many cases diagnosed with Gitelman's syndrome by clinical manifestation and biochemical abnormality so far. In this study, we performed additional study to confirm the diagnosis who clinically suspicious for Gitelman's syndrome, renal clearance study and genetic analysis. which followed by pathologic confirmation by renal biopsy with immunohistochemical stain.
A 14-year-old woman had recurrent hypokalemic periodic paralysis of upper extremity and eyelid that first presented 4 years ago. At admission, her blood pressure was 120/70 mmHg, and pulse rate was 78 beats per minute, body temperature was 36.1 C. and had no signs of dehydration. The results of the laboratory investigation were as follows : pH 7.448, PaCO2 44.4 mmHg, PaO2 65.8 mmHg, bicarbonate 30.0 mEq/L, base excess 5.2 mEq/L. serum Na 140 mg/dl, K 3.1 mg/dl, chloride 95 mg/dl. magnesium 1.4 mg/dl, calcium 9.8 mg/dl, Renin activity 22.84 ng/ml/hr, aldosterone 69.34 pg/ml. 24-hour urine calcium was 18.2 mg/day. which is very low and strongly favours a diagnosis of Gitelman's syndrome. Chloride clearnce was increased more than 10 times after furosemide administation, but not increased after hydrochlorothiazide treatment. And the distal fractional chloride reabsoption was significantly decreased by furosemide injection, whereas hydrochlorothiazide had no effect on it.
Genomic DNA sequence analysis of SLC12A3 revealed that her parents were all heterogygous, carry only one SLC mutant allele, and her little brother and herself were homozyous, carrying two different mutations on the parental of SLC12A3. But her brother has no symtoms, so we could guess the clinical manifestations of GS are highly variable. The renal biopsy was performed, and the immunohistochemical stain showed us patient's NCCT was decreased than control, and the NKCC was normal.

Key Words: Gitelman 증후군, SLC12A3 유전자 검사
Gitelman syndrome, SLC12A3, Renal biopsy