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특발성 영아 고칼슘혈증 증례

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A Case of Idiopathic Infantile Hypercalcemia

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Idiopathic infantile hypercalcinosis is a uncommon disorder of unknown etiology hypercalcemia in the asymptomatic infant. We experienced a case of newborn girl was diagnosed as idiopathic infantile hypercalcinosis with nephrocalcinosis. She was admitted at 5 days after birth with neonatal jaundice. The serum level of calcium was 13.6 mg/dL (referance range 8.4-10.2), ionized calcium 6.3 mg/dL (reference 4.07-5.17), urine calcium/creatinine ratio 1.47 (reference 0.03-0.81), 25(OH)D 51.2 ng/mL (referance range 20-60), and 1,25(OH)2D 45.42 pg/mL (referance range 15-75). Neonatal jaundice was resolved but hypercalcemia with hypercalciuria was persist. PTH level was suppressed below 1.2 pg/mL. Renal ultrasonogram and CT showed the nephrocalcinosis. On physical exam, she did not show the dysmorphic features with Williams syndrome. Also FISH testing for the elastin gene microdeletion on chromosome 7 that detects over 99% of individuals with Williams syndrome was negative. She had no familial history of the renal disease. Sequence analysis of CYP24A1, the key enzyme of 1,25-dihydroxyvitamin D3 degradation was performed and CYP24A1 mutations were not identified. She was treated with thiazide which inhibits calcium excretion to urine and ketoconazole, a general inhibitor of P450 enzymes, which are necessary for the metabolism of vitamin D compounds. After 3 months of therapy, PTH level was normalized. Nephrocalcinosis still persisted even though hyprcalciuria and hypercalcemia resolved.

Key Words: 고칼슘혈증, 고칼슘뇨증, 신결석
Hypercalcemia, Hypercalciuria, Nephrocalcinosis