Renal coloboma 증후군에서 PAX2 유전자 분석

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PAX2 Gene Analysis in Renal Coloboma Syndrome

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Introduction : The renal coloboma syndrome is caused by mutations in the PAX2 gene and is characterized by renal malformations and optic disc coloboma. We describe one family with papillorenal syndrome in two consecutive generations and evaluate the genetic defect responsible for the disease.

Case : Patient 1 is a 48 year old man who received a diagnosis of chronic renal failure at age 36 year. Abdominal computed tomography showed bilaterally small kidneys and normal urogenital system. Ophthalmoscopy and fluorescein angiography revealed optic disc coloboma and decreased visual acuity. He currently received hemodialysis.

Patient 2 is a daughter of patient 1. She was noted at age 9 year to have mild proteinuria and an elevated creatinine (2.6 mg/dL). Renal ultrasound and voiding cystography showed bilaterally small kidneys and no vesico-ureteral reflux. At ophthalmologist's examination showed bilateral optic disc coloboma and decreased visual acuity. Spine MRI showed syringomyelia at T10-T12. She subsequently developed chronic renal impairment and received hemodialysis at 15 year.

Genetic study : Fragments spanning exon 1-12 of the PAX2 gene were amplified from genomic DNA using PCR primers. The PCR products were purified and directly sequenced using the Big Dye Terminator and an ABI PRISM sequencer. Two single nucleotide polymorphisms in exon 8 of PAX2 was idenfified in patient 1 and unaffected daughter (1341C>T, 1452C>A) but not identified in patient 2.

Conclusion : We performed direct DNA sequencing of exon 1 to 12 of PAX2 gene, however, no definite mutation responsible for renal coloboma syndrome was detected in the PAX2 genes. This result suggests that the renal coloboma syndrome might be genetically heterogenous or other genes could be responsible in this family.