

A Case of Autosomal Dominant Polycystic Kidney Disease in Conjunction with Horseshoe Kidney

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Introduction: Autosomal dominant polycystic kidney disease (ADPKD) is one of common hereditary disorder due to mutations in genes that express polycystin 1 (PKD1) and polycystin 2 (PKD2). It has been reported that the prevalence ranges from one in 400 to 1000. The prevalence of the horseshoe kidney (HSK), a congenital renal fusion anomaly during embryogenesis, ranges from one in 400 to 600. Coexistence of both distinct clinical conditions is extremely rare. Polycystic HSK is estimated to affect between one in 134,000 and 8,000,000. A case of polycystic HSK had been reported in South Korea. However, the case was not confirmed as ADPKD because genetic testing was not performed. To our best knowledge, ADPKD diagnosed by genetic test accompanied by HSK has been shown for the first time in South Korea. We report a case to cultivate the current medical literature regarding this rare entity.

Case Report: A 24-year-old man referred to nephrology outpatient clinic in Jeonbuk National University Hospital for health check-up before joining the army. There was no significant medical illness and physical examination was unremarkable. His father of age 45 and his uncle of age 46 were diagnosed as ADPKD several years ago, currently confirmed end stage renal disease, and both started on hemodialysis. The patient had been hypertensive (150/90 mmHg) from the age of 20. Computed tomography (CT) revealed enlarged kidneys with multiple cysts, fused by their lower poles, presenting polycystic HSK (Figure1). There were also multiple cysts in liver. The renal function was noticed to be normal (serum creatinine of 0.85mg/dL, eGFR of 122ml/min/1.73m²) and there was no evidence of any proteinuria. His gene sequencing for PKD1 was as follows: heterozygous pathogenic variant c.165_171del (p.Leu56ArgfsTer15) which genetically confirms the diagnosis of ADPKD type 1. The patient started on telmisartan for hypertension and is being followed-up annually.

Discussion: Concomitant presence of ADPKD and HSK is likely to lower the age of renal failure and therefore warrants closer surveillance of these patients.

