

Abstract Submission No.: A-1214

Unilateral polycystic kidney with PKHD1 gene mutation

Soon-Kil Kwon¹, Hyewon Woo², Bum Sang Cho³

¹Department of Internal Medicine-Nephrology, Chungbuk National University Hospital, Korea, Republic of

²Department of Pediatrics, Chungbuk National University Hospital, Korea, Republic of

³Department of Radiology, Chungbuk National University Hospital, Korea, Republic of

Case Study : A 19-year-old female visited the outpatient clinic to follow up on kidney imaging. She had been diagnosed with incidental left hydronephrosis which was found at fetal ultrasonography and followed yearly until 6 years old, and she had no medical problems including urinary tract infections. There was no family history of kidney disease including cystic disease and her parents' kidney sonographic findings were normal. Her physical examination findings, blood and urine tests were all normal, and The sonographic finding when she was 2 years old showed only a 0.6 cm sized simple cyst in the right kidney and severe hydronephrosis of the left kidney, however, dynamic abdominal and pelvic CT performed at 19-year-old showed innumerable cysts of the right kidney, and residual mild dilatation of left renal pelvocalyx which was thought the sequelae of previous severe hydronephrosis. CT showed that the left kidney morphology was normal except for the previously diagnosed mild calyceal dilatation, but a polycystic change was found throughout the total right kidney (Fig. 1). A next-generation sequencing (NGS) for polycystic kidney disease confirmed a heterozygous mutation of c.7769T>G, p.(Met2590Arg) in the PKHD1 gene which was a variant of unknown clinical significance (VUS). PKD1, PKD2, and other PCKD-related gene tests were negative (Table 1). In this case, the PKHD1 mutation was confirmed through NGS analysis in an incidentally found unilateral polycystic kidney. Although the unilateral polycystic kidney is not clinically significant, it is necessary to confirm that it is one of the various heterozygotes of the PKHD1 gene.

Figure1.png

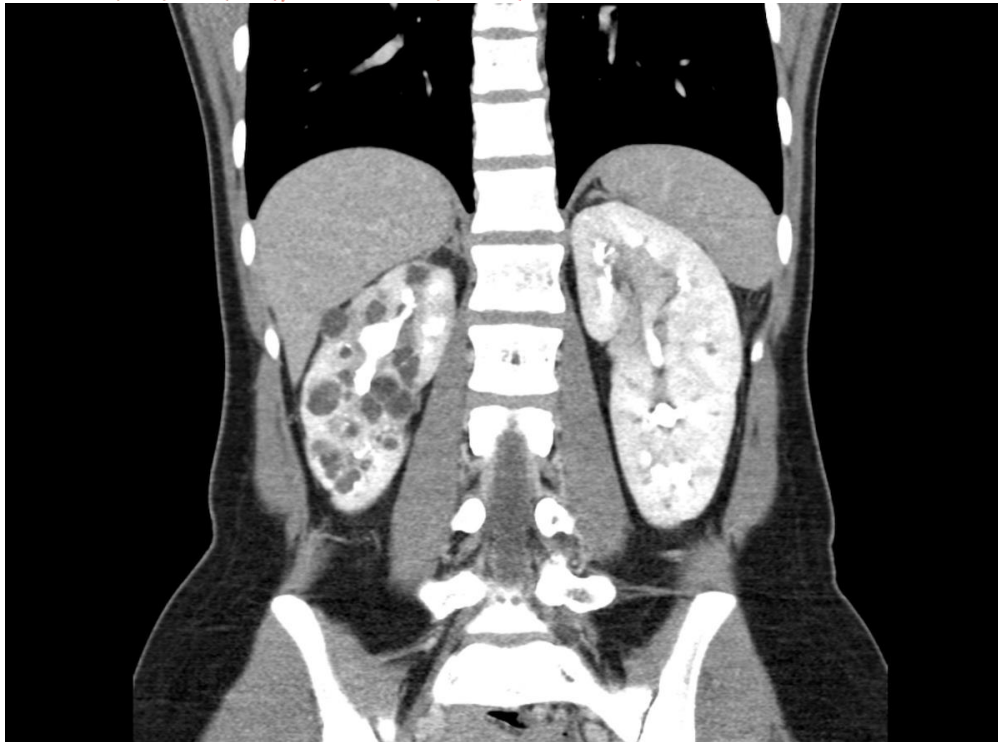


Figure1.png

Table 1. Result of next-generation sequencing polycystic kidney disease panel* and variant interpretation

Gene	Zygosity	Variant	Classification	CADD (hg38 v1.7)	Phenotype	Phenotype MIM No	Inheritance
<i>PKHD1</i>	Heterozygous	c.7769T>G, p.(Met2590Arg)	VUS	0.912	Polycystic kidney disease 4, with or without hepatic disease	263200	AR
<i>PKD1</i>	Not detected				Polycystic kidney disease 1	173900	AD
<i>PKD2</i>	Not detected				Polycystic kidney disease 2	613095	AD
<i>GANAB</i>	Not detected				Polycystic kidney disease 3	600666	AD
<i>DZIP1L</i>	Not detected				Polycystic kidney disease 5	617610	AR
<i>DNAJB11</i>	Not detected				Polycystic kidney disease 6 with or without polycystic liver disease	618061	AD
<i>HNF1B</i>	Not detected				Renal cysts and diabetes syndrome	137920	AD
<i>ALG8</i>	Not detected				Polycystic liver disease 3 with or without kidney cysts	617874	AD
<i>LRP5</i>	Not detected				Polycystic liver disease 4 with or without kidney cysts	617875	AD

*Gene list included in NGS panel is ANKS6, CEP164, CEP83, COL4A1, DNAJB11, DZIP1L, GANAB, HNF1B, INVS, MAPKBP1, NPHP1, NPHP3, NPHP4, PKD1, PKD2, PKHD1, TMEM67, TSC1, TSC2, TTC21B, UMOD, VHL, WDR19, ALG8, ALG9, CEP290, COL4A4, ETFA, FLCN, LRP5, NOTCH2, PAX2, PMM2, and SEC61A1.

Abbreviation: CADD, combined annotation dependent deletion; AR, Autosomal recessive; AD, Autosomal dominant; VUS, Variant of Uncertain Significance