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## **Long-read Whole Genome Sequencing Found WDR19 Gene as a New Causative Gene of Polycystic Kidney Disease**

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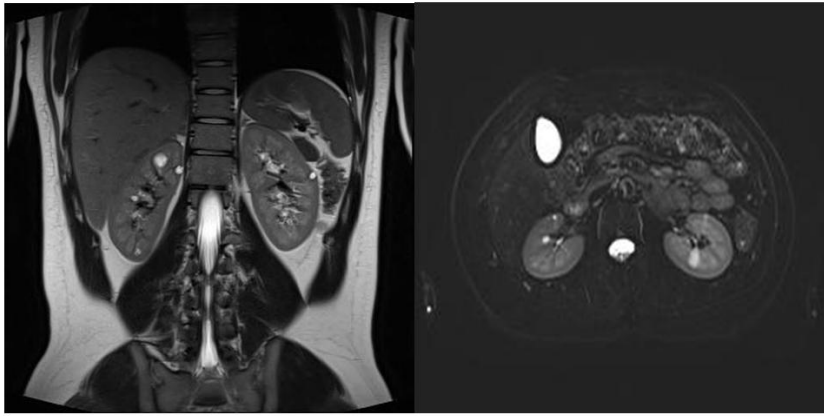
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**Case Study :** Polycystic kidney disease (PKD) is a genetic disorder that causes multiple renal cysts. PKD1 and PKD2 are known as major genes of PKD, but various minor genes have been recently identified as causative genes. Here, we report a case where WDR19 was identified as a new causative gene for PKD through long-read whole genome sequencing and family segregation study. A 47-year-old female patient was referred to the nephrology department for multiple renal and hepatic cysts. She had been treated for high blood pressure for 3 years. An abdominal CT scan demonstrated atypical PKD with lopsided type (Mayo 2) and several small hepatic cysts (Figure 1a). She had normal renal function (serum Cr 0.51 mg/dL) and there was no urinary abnormalities. She denied any family history of PKD. We performed targeted exome panel sequencing to screen for ciliopathy genes. The result of panel sequencing revealed multiple non-frameshift deletion in WDR19 gene. Subsequently, we performed long-read whole genome sequencing and family segregation study to confirm the pathogenic variant. Final gene analysis revealed non-frameshift deletion of WDR19 gene (Figure 2). In family segregation analysis, her 18-year-old second son demonstrated multiple renal cysts (Figure 1b), while her husband and the first son did not have renal cysts. We confirmed that the pathogenic variant was only segregated to her second son. The patient had undergone blood tests every six months and abdominal CT scans annually for 4 years. Her renal function had been stable and the number and size of cysts remained stationary. This case suggests that WDR19 is a causative gene for atypical PKD and indicates a favorable prognosis.

Figure 1.png



(a)



(b)

Figure 1.png  
**PKD WDR19 nonframeshift deletion**  
**(chr4:39189755-60, chr4:39189768)**

