**[BIOINFO 2023]**

# Whole exome sequencing and familial segregation study with targeted sequencing revealed pathogenic genes in inherited cystic kidney disease

Dajun Lee[1], Yun Kyu Oh[2,] Hayne Cho Park[2], and Jungmin Choi[1,*]

[1] *Department of Biomedical Sciences, Korea University*
[2] *Department of Internal Medicine, Seoul Metropolitan Government Seoul National University Boramae Medical Center*
[2] *Department of Internal Medicine, Kangnam Sacred Heart Hospital*

**Corresponding author: jungminchoi@korea.ac.kr*

Inherited cystic kidney disease (iCKD) is a spectrum of disorders in which clusters of renal cysts develop as the result of genetic mutation. To set up the exact methods and pipelines for defining genetic mutations of inherited cystic kidney disease, we analyzed targeted sequencing on 800 patients from May 2020 to May 2022. Furthermore, we also performed whole exome sequencing (WES) on 126 patients who did not have a causal variant of 89 known targeted genes or had secondary phenotype such as polycystic liver disease (PLD) in order to discovery novel causal gene and suggest causal variants for secondary phenotype. In addition, for patients who had more than one explainable pathogenic variants, we performed familial segregation analysis to determine which variant may have had a more causal effect. For further analysis, we are planning to collect 800 probands in the second phase from November 2022 to November 2024 as in the first phase to help researchers implement precision medicine for Korea iCKD paitnets and improve our established pipeline.