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Diagnosis of Genetic kidney disease

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Whole exome sequencing (WES) using massively parallel sequencing techniques has expanded the spectrum of genetic defects in a variety of genetic disorders. To date, more than 7,000 genetic disorders, involving more than 4,000 genes, have been identified, and the numbers are increasing. The diagnostic rate of WES has been found to range from 30% to 40%, a variation that may be attributed to the numbers and phenotypes of enrolled patients and the anthropologic characteristics of study cohorts.

Genetic causes have been identified for ~10% of adult chronic kidney diseases and >70% of pediatric renal diseases. Considering more than 200 genes are responsible for a variety of genetic kidney diseases, genomic studies such as target gene panel testing, WES and whole genome sequencing using massivelly parallel sequencing techniques have been recommended for the diagnosis.

However, WES are time-consuming and labor-intensive, requiring clinical geneticists and bioinformaticians to match large numbers of candidate variants with various clinical symptoms in each subject analyzed . Moreover, in the absence of supporting data, many variants remain variants of uncertain significance (VUS), limiting the ability to confirm genetic diagnoses.

In this talk, genetic diagnostic process using a new streamlined automated interpretation software system, EVIDENCE (3Billion, Inc., Seoul, South Korea), will be presented and the results of pilot study using WES in patients suspected as having genetic kidney diseases