



Lecture Code : GD01-S3

Session Name : Genetic Disease

Session Topic : Genomics to Everyone!: Building up National Genomics Program

Date & Time, Place : June 19 (Thu) / 15:00-17:00 / Room 4 (Room 203)

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## **Current Initiatives and Future Directions of the Genetic Kidney Disease Research Group**

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The Genetic Kidney Disease Research Group (GKDRG) under the Korean Society of Nephrology has established itself as a national leader in promoting genomic medicine in nephrology. Since its inception in 2019, the group has actively contributed to the diagnosis, education, and research of hereditary kidney diseases, with a growing emphasis on integrating genetic testing into routine clinical practice. One of the group's key initiatives includes the development of evidence-based clinical guidelines for Alport syndrome, a major monogenic cause of chronic kidney disease (CKD). Through a multi-disciplinary task force, including pediatric nephrologists, internists, pathologists, and geneticists, the GKDRG has compiled Korea's first national clinical practice guideline tailored to domestic healthcare realities. This guideline not only supports early recognition and treatment of Alport syndrome but also promotes genetic counseling and surveillance for at-risk family members. To address the significant underdiagnosis of hereditary nephropathies, GKDRG has implemented hybrid symposia, professional training programs, and multicenter cohort studies. A major project involves establishing a national registry and biobank for genetically confirmed kidney diseases, with early data highlighting the prevalence of COL4A3–5 variants among Korean patients previously misdiagnosed with conditions such as FSGS or IgA nephropathy. In 2025, the group launched a nationwide prospective study on Alport syndrome supported by the Korean Society of Nephrology and the Renal Research Foundation. This initiative aims to identify the genotypic and phenotypic spectrum of the disease and to link patients to emerging therapies such as SGLT2 inhibitors, RAAS blockade, and RNA-based interventions. Looking ahead, the GKDRG aims to scale up precision nephrology efforts by collaborating with global consortia, standardizing variant interpretation pipelines, and advocating for broader insurance coverage of genetic diagnostics. Future symposia will expand to include AI-based variant analysis and novel therapeutic modalities for Fabry disease and other rare disorders, as

evidenced by the upcoming 2nd GKDRG Symposium in April 2025. Through a comprehensive approach that combines research, education, and clinical translation, the GKDRG continues to pioneer efforts in reducing diagnostic delays, improving patient outcomes, and advancing personalized nephrology in Korea.

**Keywords:** Genetic Kidney Disease, Alport Syndrome, Clinical Guidelines, Genetic Testing, Precision Medicine