Shaping Tomorrow's Nephrology: Insight-Driven Kidney Care
KSN 2026 SEOUL, KOREA
The 46th Annual Meeting of the Korean Society of Nephrology
Jun 11 (Thu) – 14 (Sun), 2026
COEX, Seoul, Korea

Lecture Code : JS04-S3
Session Name : KSN-ESPN Joint Symposium
Session Topic : KSN-ESPN Joint Symposium
Date & Time, Place : June 12 (Fri) / 13:50-15:30 / Room 1 (GBR 101), 1F

Genetic Aspects of Renovascular Hypertension in Asian Children

Heeyeon Cho

Samsung Medical Center, Republic of Korea

Genetic Aspects of Renovascular Hypertension in Asian Children Heeyeon Cho Department of Pediatrics, Samsung Medical Center, Sungkyunkwan University School of Medicine, Seoul, South Korea Background and Clinical Profile Pediatric renovascular hypertension (RVH) is frequently associated with systemic vasculopathies. Our initial retrospective study of 25 Korean pediatric RVH patients revealed a unique epidemiological profile: Moyamoya disease (MMD) was the most prevalent underlying condition (40%), surpassing the global prevalence of Fibromuscular Dysplasia (FMD) or Takayasu arteritis. Notably, 70% of these patients were diagnosed with RVH prior to their MMD diagnosis, suggesting that renal artery involvement may be the sentinel clinical manifestation of this systemic disease. The RNF213 p.R4810K Paradigm Building on these clinical findings, we investigated the genetic basis of this association, specifically focusing on the RNF213 p.R4810K variant, a known susceptibility gene for East Asian MMD in RVH pediatric patients. In our cohort, approximately 40% of patients diagnosed with RVH tested positive for RNF213-related variants. This identifies the variant as a primary driver of renal vasculopathy in Korean children. Conversely, in the remaining 60% of RVH cases where RNF213 variants were absent, the underlying genetic etiology remains unidentified despite Whole Exome Sequencing (WES). Consequently, further research is currently underway to discover novel causative genes in this RNF213-negative subgroup. Clinical Characteristics & Treatment Challenges RNF213-associated RVH is characterized by a predominance of proximal/ostial renal artery stenosis, frequently occurring in conjunction with mid-aortic syndrome (diffuse aortic narrowing). These anatomical features contribute to poor outcomes with conventional percutaneous transluminal angioplasty (PTA). While initial medical management is essential, clinicians should consider surgical bypass when the patients show resistant

hypertension and/or renal insufficiency. Screening and Diagnostic Recommendations Given that RVH may precede cerebrovascular symptoms, we propose the following protocols for Asian pediatric populations: · Genetic Priority: In cases of pediatric RVH without clear etiology, RNF213 genetic testing is recommended. · Systemic Screening: The presence of RVH associated with the RNF213 variant necessitates comprehensive cerebrovascular evaluation to screen for occult MMD. · Future Directions: While homozygosity indicates higher severity, further research is needed to elucidate the phenotypic spectrum of heterozygous carriers and to identify the genetic drivers in RNF213-negative systemic vasculopathies. Conclusion Current evidence suggests that RNF213-associated vasculopathy may represent a significant component of the pediatric RVH landscape in Korea. Shifting the diagnostic focus toward genetic screening and recognizing the limitations of angioplasty in this population are critical steps in preventing target-organ damage and improving long-term renal and neurological outcomes.

Keywords: renovascular hypertension, pediatric, genetic, RNF213