

## Nephronophthisis의 진단과 치료

서울대학교병원 소아청소년과

강 희 경

### Diagnosis and Management of Nephronophthisis

Hee Gyung Kang

Pediatrics, Seoul National University Children's Hospital

Nephronophthisis (NPHP) is an autosomal recessive progressive tubulointerstitial nephropathy. It is the most common genetic cause of end-stage renal disease (ESRD) in childhood and adolescence. Patients typically present with polyuria, nocturia, growth retardation, and anemia with an insidious onset of chronic renal failure. Diagnostic criteria of NPHP is as follows; insidious onset of ESRD in the first 3 decades of life without evidence of previous renal damage. Renal ultrasonographic findings show increased echogenicity, normal or slightly decreased size for age and corticomedullary differentiation loss. Renal pathology shows chronic tubular interstitial disease. NPHP is often accompanied by impairment of other systems, such as vision (retinitis pigmentosa), cerebellum (cerebellar vermis aplasia), and liver (fibrosis), comprising various syndromes including Joubert syndrome and Meckel Gruver syndrome (MKS). Molecules critical to function or structure of primary cilia are involved in the pathogenesis of this condition, and collective term of nephronophthisis-related ciliopathy (NPHP-RC) is used. Genetic diagnosis is available for common causative genes (upto 30 % of the patients) at research laboratory and next generation sequencing technique is being applied to test rare or novel genes of ciliopathy. There is no curative treatment for NPHP, and no recurrence of NPHP has been reported after kidney transplantation.