Clinical and genetic characteristics of Japanese nephronophthisis patients

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Abstract

Background: Nephronophthisis (NPH) is a disease characterized by renal medullary cyst formation. NPH accounts for 4 to 5% of end-stage renal disease occurring in childhood.

Method: We investigated the clinical context and characteristics of renal and extra-renal symptoms, as well as the NPHP genes with genomic DNA extraction, PCR and determination of NPHP gene sequence, in 35 Japanese patients with clinical and histologic features suggesting NPH.

Results: NPH occurred fairly uniformly throughout Japan irrespective of region or gender. In 3 families, NPH affected siblings. Age of patients ranged from 2 to 38 years (median; 12.5. Renal abnormalities attributable to NPH discovered through mass screening, such as urine tests in school. However, NPH accounted for less than 50% of children with abnormal findings, including incidentally discovered renal dysfunction during evaluation of extra-renal symptoms or during routine check-ups. Typical extra-renal manifestations led to discovery including anemia and delayed physical development. The urine often showed low gravity specific density and low molecular weight proteinuria. Frequent renal histologic findings included cystic dilation of tubules, mainly in the medulla and irregularity of tubular basement membranes. Genetic abnormalities of NPHP1 were not common, with large deletions frequently noted. Compound heterozygotes showing single abnormalities in each of NPHP1, NPHP3 and NPHP4 were observed.

Conclusions: Our findings resemble those reported in Western populations.

Biography

Keisuke Sugimoto has completed his MD and PhD from Kindai University, Faculty of Medicine. He worked as a Research Fellow at Department of Nephrology, Vanderbilt University in USA. He obtained a position as Assistant Professor at Department of Pediatrics- Faculty of Medicine, Kindai University. He has published more than 20 papers associated with Nephrology.

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