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Various Clinical Aspect and Genetic Counselling of Hereditary Kidney Disease

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Genetic kidney disease is a rare disease entity, accounting for ~ 10% of adults with end stage kidney disease. On the other hand, diverse genetic etiologies have been found in a significant proportion (~70%) of pediatric or young adult patients with kidney disease. To date, more than 200 monogenic or chromosomal structural abnormalities have been known as underlying kidney diseases. In these regards, genetic testing has been recommended in children or young adult with steroid resistant nephrotic syndrome, congenital malformations of kidney and urinary tract, cystic disease or kidney disease with extrarenal manifestations. However, as the genetic diagnostic technologies develop remarkably, their utility expands in the diverse fields of clinical nephrology. Moreover, the genetic diagnosis gives a substantial impact on patient care with requirement for the appropriate genetic counseling. In this talk, diverse genetic testing modalities will be introduced with the implications of genetic testing in clinical genetic diagnostics and genetic counseling.