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Is there a significant advance in using genetics for Pediatric Patients?

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There have been significant advances in using genetics to diagnose and treat pediatric patients with kidney diseases.
When I began my pediatric nephrology fellowship in 2001, nephrin was recently discovered, putting podocytes in the spotlight. Since then, multiple genes related to podocytes or glomeruli that cause congenital or hereditary nephrotic syndrome or glomerulopathies have been identified, along with many other causative genes of various kidney diseases. As a result, we can now narrow down suspects through careful physical examination of extra-renal symptoms, such as ambiguous genitalia (*WT1*), a light reflex of the eye (*LAMB2*), or disproportional short stature (*SMARCAL1*), and laboratory findings such as thrombocytopenia (*MYH9*) or hypocomplementemia (*CFH*). With advances in genetics, the time required to obtain a precise diagnosis has been significantly reduced, and risky diagnostic tests such as water deprivation are no longer necessary. Once pathogenic or likely-pathogenic variants of a culprit gene are confirmed, we no longer ponder how to modify immunosuppression to achieve remission of proteinuria but focus on improving the growth and quality of life of patients. With advances in genetics, the practice of pediatric nephrology has evolved from trying empirical or symptomatic treatments based on a clinician's personal experience to providing evidence-based medicine targeting the pathophysiology of a disease. For example, complement 5-blocker for atypical hemolytic uremic syndrome or anti-FGF23 antibodies for X-linked hypophosphatemic rickets, or at least avoid ineffective, possibly harmful treatment options. When the transplantation is being considered, we can confidently counsel the risk of recurrence of focal segmental glomerulosclerosis. We can also provide cascade testing for family members and prenatal screening of younger siblings or offsprings of a proband.
Indeed, not all kidney diseases are of genetic origin, and even for genetic problems, sometimes we cannot find the (likely) pathogenic variants. Often, variants of unknown significance perplex clinicians and patients alike. In addition, only a handful of diseases have targeted therapy. Therefore, we have a long way to go to advance in genetics and clinics.