

## Genetics of human kidney and urinary tract malformations

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Congenital Anomalies of the Kidney and the Urinary Tract (CAKUT) account for 40-50% of pediatric end-stage kidney failure worldwide. CAKUT can occur as familial or sporadic disease with highly variable phenotypic expression. For the past 40 years, the diagnostic approach to CAKUT has relied on description of anatomic defects, which poorly discriminates subtypes of disease and provides little prognostic information. By traditional linkage analysis we identified loci on chromosomes 1p32-33, 10q24-26 and 12p11-q13 in our largest families and demonstrated significant genetic heterogeneity for this trait. Due to paucity of fundamental insight about primary pathogenesis, therapeutic options are also severely limited.

Recent technology development allow for identification of rare mutations at a genome-wide level in small families and even in sporadic individuals, with great potential for the identification of susceptibility genes for CAKUT. We developed a comprehensive strategy by using novel technologies for the identification of rare structural variants and point mutations by high-density chips genotyping and next-generation sequencing.

We showed that almost 20% of patients with renal hypodysplasia/agenesis carry a diagnosis of a known or novel genomic disorder, with important implications in diagnosis, genetic counseling and stratification of risk for developing severe extra-renal manifestations, such as autism, mental retardation, and others. Moreover, by combination of linkage analysis and next-generation sequencing we identified a novel gene predisposing to kidney and urinary tract malformations.

High-throughput technologies for search of rare structural variants and point mutations will lead to the development of novel diagnostic and therapeutic strategies for kidney and urinary tract malformations.

### Biography

Simone Sanna-Cherchi has completed his M.D. at the age of 25 years at the University of Parma, Italy, where he also complete his residency in Internal Medicine and Fellowship in Nephrology. He then completed a 5-years post-doctoral training at the Division of Nephrology at Columbia University Medical Center in New York, where he was then appointed as Faculty. He has published more than 25 papers in top-tier, high-impact journals and serves as editorial board members of several reputed journals, including Kidney International. His work has been funded by governmental and non-governmental agencies, including the NIH, AHA, ASN, and Telethon Foundation.

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## Anti-DS-DNA positivity in a patient with prostate cancer and acute kidney injury: A case report

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**Objective:** Patients with malignancies may develop autoimmune and rheumatic manifestations as a result of generation of autoantibodies, paraneoplastic syndromes, direct invasion of joints and muscles by the tumour cells, or combination chemotherapy. We present a case with prostate cancer and acute kidney injury (AKI) mimicking rapidly progressive glomerulonephritis with a positive test result for anti-double-stranded deoxyribonucleic acid antibodies (anti-dsDNA ab)

**Case Presentation and Intervention:** The 78 years old male patient applied to our center with the complaint of generalized bone pain since approximately two months, and rapid onset of weakness, oliguria and dysuria for three days. He had been diagnosed metastatic prostate cancer (PC) for two years and at the time of application had received high dose of naproxen sodium because of severe bone pain for 5 days. Serum creatinine level was elevated on admission (2.4 mg/dl). Urinalysis revealed microscopic hematuria, granular casts and proteinuria of 1.2 g/d. Immunologic tests including anti-dsDNA antibody were done regarding acute nephritic syndrome. The subject was positive for anti-dsDNA ab with the value of 96 IU/ml (normal, <10 IU/ml). Renal biopsy was planned however the patient refused the procedure. After 6 days of oliguric period, his clinical condition and renal functions returned to almost normal with supportive care within 12 days.

**Conclusion:** Malignant diseases may be a trigger for either the generation of some autoantibodies. Attention must be paid when interpreting the anti-dsDNA positivity in malignant patients with AKI mimicking RPGN.