Familial juvenile hyperuricemic nephropathy (FJHN) and medullary cystic kidney disease type 1 (MCKD1) and type 2 (MCKD2) are autosomal dominant tubulointerstitial nephropathies characterized by combinations of hyperuricaemia, gouty arthritis, progressive renal insufficiency, and in some but not all families, medullary cysts. The phenotypic expression of these diseases is inconsistent, overlaps and indicates broader genetic and allelic heterogeneity. Their pathophysiology was mainly unknown. Previous studies localized FJHN/MCKD genes to chromosomes 16p11 and 1q21. This thesis was primarily aimed at identification of molecular bases and mechanisms underlying FJHN/MCKD. To follow this aim, we focused on collection and characterization of FJHN/MCKD patients and families, identification of disease causing genes in affected families, characterisation of identified proteins and their mutated forms and the isolation and characterisation of interacting partners of newly identified proteins. We employed and established numerous molecular genetic, molecular biological and biochemical methods. We gathered one of the largest sets of families with FJHN/MCKD in the world. In about 26% of families we identified *UMOD* (uromodulin encoding) gene mutations and characterised by various approaches 6 uromodulin mutant proteins. In 1 family we defined new genetic locus on chromosome 1q41, identified the mutation in *REN* (renin encoding) gene and characterised by various approaches renin mutant protein. In the rest of families, we could not prove the genetic linkage to any of known or newly identified FJHN/MCKD loci. Nevertheless, we provided evidences that alteration of uromodulin biology is common to genetically heterogenous FJHN/MCKD and suggested several pathogenic mechanisms leading to the disease. We were unable to identify any uromodulin interacting proteins by two different protein-protein interaction approaches. As a practical consequence, we perform routinely the selective screening of hyperuricaemic conditions and provide unique, complex diagnostic service for this group of disorders.