

Autosomal dominant tubulointerstitial kidney disease (ADTKD) describes a group of diseases that affect the tubules of the kidney. These conditions have the following characteristics: (1) They are inherited in an autosomal dominant manner; this means that a parent has a 50% chance of passing the disease on to their children. Often many family members are affected. (2) Chronic kidney disease develops. This initially is noticed as an elevated blood creatinine level. There are no symptoms from an elevated creatinine until it is very high, so often the elevated creatinine is found out on blood testing at a doctor's office or in the hospital. Often, doctors are uncertain why the creatinine is elevated. As chronic kidney disease progresses, and the creatinine rises more, patients develop symptoms of fatigue, anemia, and feel cold all the time. Decreased appetite and fluid retention develop as the patient nears the need for dialysis. (3) Dialysis or kidney transplant is required sometime between the 4th and 7th decade of life. (4) Several types of the disease are associated with elevated uric acid concentrations in blood and gout, which sometimes starts in the teenage years. In ADTKD-UMOD and ADTKD-REN, some – but not all - family members are affected by gout. All types of autosomal dominant tubulointerstitial kidney disease are very uncommon.