SIGNIFICANCE STATEMENT

Clinical diagnosis of hereditary tubulointerstitial kidney disease has always been a challenge. The subform ADTKD-*MUC1* largely escapes from molecular diagnostics, because its mutational hotspot is located in an inaccessible repeat domain. Here, the authors re-evaluate the detection of *MUC1* mutations by SNaPshot minisequencing and establish immunohistochemistry for the resulting frameshift protein in human kidney samples in comparison with mucin 1 from the wild-type allele. MUC1 frameshift protein accumulates in the tubular cytoplasm of ADTKD-*MUC1* kidneys, where wild-type mucin 1 is also readily detectable. Molecular diagnosis of ADTKD-*MUC1* is possible but restricted to a limited number of laboratories. Immunohistochemistry on kidney biopsies may be a feasible method for nongenetic selection of patients for further diagnostics.