# Autosomal dominant tubulointerstitial kidney disease-UMOD

## Description

Autosomal dominant tubulointerstitial kidney disease-*UMOD* (ADTKD-*UMOD*) is part of a group of disorders (collectively called autosomal dominant tubulointerstitial kidney disease or ADTKD) that cause a slow loss of kidney function. In people with ADTKD-*UMOD,* the signs and symptoms of kidney disease often begin in adolescence or early adulthood. Over time, the kidneys become less able to filter fluids and waste products from the body. People with ADTKD-*UMOD* eventually develop kidney failure, which requires either dialysis to remove waste from the blood or a kidney transplant. The age at which people with ADTKD-*UMOD* develop kidney failure can vary, though the average age is approximately 45 years.

People with ADTKD-*UMOD* typically develop high levels of a waste product called uric acid in their blood. Normally, the kidneys transfer uric acid from the blood into urine, which then removes it from the body. People with ADTKD-*UMOD* are unable to remove uric acid from the blood effectively. In about 50 percent of people with ADTKD-*UMOD*, uric acid builds up in the joints and causes a form of arthritis called gout, typically in late adolescence or early adulthood. Gout is characterized by a sudden onset of severe joint pain and redness, often starting in the big toe. Untreated episodes of gout typically worsen over time.

## Frequency

ADTKD-*UMOD* is believed to account for fewer than 1 percent of all cases of kidney failure. Researchers aren't sure how common ADTKD-*UMOD* actually is, but it is considered to be one of the most common forms of kidney disease that is caused by changes in a single gene.

## Causes

Variants (also called mutations) in the *UMOD* gene cause ADTKD-*UMOD*. This gene provides instructions for making the uromodulin protein. This protein is produced by the kidneys and then released from the body in urine. Uromodulin is the most common protein found in the urine of healthy individuals. It is thought to play a role in the transport of minerals such as sodium and potassium.

Most variants in the *UMOD* gene change single protein building blocks (amino acids) in the uromodulin protein. These variants typically alter the structure of the protein, though some variants have more severe effects on protein function than others. People with *UMOD* variants that have a greater effect on protein function generally have severe kidney disease, and these individuals experience signs and symptoms of kidney disease at a younger age. Typically, *UMOD* gene variants prevent kidney cells from releasing the uromodulin protein. The buildup of uromodulin may trigger the self-destruction (apoptosis) of cells in the kidneys, leading to kidney disease and eventual kidney failure.

Learn more about the gene associated with Autosomal dominant tubulointerstitial kidney disease-UMOD

- UMOD

## Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

## Other Names for This Condition

- ADMCKD2
- ADTKD-UMOD
- ADTKD1
- Autosomal dominant medullary cystic kidney disease 2
- Autosomal dominant tubulointerstitial kidney disease 1
- Autosomal dominant tubulointerstitial kidney disease due to UMOD mutation
- Familial juvenile gouty nephropathy
- Familial juvenile hyperuricemic nephropathy 1
- FJHN
- Glomerulocystic kidney disease with hyperuricemia and isosthenuria
- HNFJ1
- MCKD2
- Medullary cystic kidney disease type 2
- UAKD
- UMOD kidney disease
- UMOD-related ADTKD
- UMOD-related autosomal dominant tubulointerstitial kidney disease
- Uromodulin-associated kidney disease

## Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Familial juvenile hyperuricemic nephropathy type 1 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C4551496/)

Genetic and Rare Diseases Information Center

- UMOD-related autosomal dominant tubulointerstitial kidney disease (https://rarediseases.info.nih.gov/diseases/10679/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- TUBULOINTERSTITIAL KIDNEY DISEASE, AUTOSOMAL DOMINANT, 1; ADTKD1 (https://omim.org/entry/162000)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=Autosomal+dominant+tubulointerstitial+kidney+disease-UMOD+OR+Uromodulin-associated+kidney+disease&sort=date)

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