REN-related kidney disease

Description

REN-related kidney disease is an inherited condition that affects kidney function. This condition causes slowly progressive kidney disease that usually becomes apparent during childhood. As this condition progresses, the kidneys become less able to filter fluids and waste products from the body, resulting in kidney failure. Individuals with *REN*-related kidney disease typically require dialysis (to remove wastes from the blood) or a kidney transplant between ages 40 and 70.

People with *REN*-related kidney disease sometimes have low blood pressure. They may also have mildly increased levels of potassium in their blood (hyperkalemia). In childhood, people with *REN*-related kidney disease develop a shortage of red blood cells (anemia), which can cause pale skin, weakness, and fatigue. In this disorder, anemia is usually mild and begins to improve during adolescence.

Many individuals with this condition develop high blood levels of a waste product called uric acid. Normally, the kidneys remove uric acid from the blood and transfer it to urine so it can be excreted from the body. In *REN*-related kidney disease, the kidneys are unable to remove uric acid from the blood effectively. A buildup of uric acid can cause gout, which is a form of arthritis resulting from uric acid crystals in the joints. Individuals with *REN*-related kidney disease may begin to experience the signs and symptoms of gout during their twenties.

Frequency

REN-related kidney disease is a rare condition. At least three families with this condition have been identified.

Causes

Mutations in the *REN* gene cause *REN*-related kidney disease. This gene provides instructions for making a protein called renin that is produced in the kidneys. Renin plays an important role in regulating blood pressure and water levels in the body.

Mutations in the *REN* gene that cause *REN*-related kidney disease result in the production of an abnormal protein that is toxic to the cells that normally produce renin. These kidney cells gradually die off, which causes progressive kidney disease.

Learn more about the gene associated with REN-related kidney disease

- REN

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

- Familial juvenile hyperuricemic nephropathy 2

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Familial juvenile hyperuricemic nephropathy type 2 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C2751310/)

Genetic and Rare Diseases Information Center

- REN-related autosomal dominant tubulointerstitial kidney disease (https://rarediseases.info.nih.gov/diseases/13461/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, 4; ADTKD4 (https://omim.org/entry/613092)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28familial+juvenile+hyperuricemic+nephropathy+2%29+OR+%28%28REN%29+AND+%28gene%29%29+AND+%28renin%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2160+days%22%5Bdp%5D)

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