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Familial Juvenile Nephronophthisis

National Cancer Institute

Source

National Cancer Institute. *Familial Juvenile Nephronophthisis*. NCI Thesaurus. Code C74998.

Progressive tubulointerstitial nephritis inherited in an autosomal recessive manner. It is caused by mutations in the NPHP1 gene. Patients present with anemia, polyuria, and polydipsia during childhood. The progressive bilateral kidney damage results in renal failure.