UMOD patient information day summary (22nd September 2018, Wellcome Collection, London)

What's in a name?

Names of this condition discussed

New names discussed: Autosomal Dominant Tubulointerstitial Kidney Disease (ADTKD)

Varieties of ADTKD: ADTKD-UMOD; ADTKD-MUC1, ADTKD-HNF1B, ADTKD-NOS

These varieties are dependent upon knowing the gene

All types are autosomal dominant – so a 50% risk of passing the gene on with each child. Does not skip generations. If a person doesn't have the gene, they can't pass it on.

What are the complications?

Chronic kidney disease in adults –a slow decline

High blood pressure (BP) (also called Hypertension)

There may be cystic kidney disease – usually medullary cysts (in the middle of the kidney) which are shown up on an ultrasound of the kidneys. Sometimes the kidneys are just a bit smaller, sometimes the kidneys look normal.

Gout – may predate chronic kidney disease

Usually treated with allopurinol or feboxustat. Losartan is a useful BP tablet as it lowers uric acid as well.

Low purine diet is advised

Family presentations of UMOD disease

UMOD can start "out of the blue" so called de novo mutation but usually there is a family history of gout and chronic kidney disease.

Each family can have different severities of disease and even within each family the disease can be quite different. Control of Blood pressure is important. Treat gout when it occurs.

Clinical trials

A definitive clinical trial to show if allopurinol (or any other drug) has not been performed due to its rarity. It would need a European or worldwide collaboration to perform this study properly. Newcastle and Cambridge performed a pilot study which showed that UMOD patients were very compliant and took part in the study readily. The variable progression of disease makes a short term study difficult

Diet

Information about Gout and diet is readily available on the web. See

www.ukgoutsociety.org/docs/goutsociety-allaboutgoutanddiet-0113.pdf

Transplantation and UMOD

Disease does not recur in transplant. Knowing gene defect may make it easier for other family members to donate (they can be checked to ensure they are mutation negative). Preparation for transplant should be done as chronic kidney disease progresses.

Renal RaDaR

The Renal community is trying to establish a map and database of all patients with ADTKD and UMOD. There is lots of good information here: http://rarerenal.org/patient-information/adtkd-patient-information/

Please ask your doctor to sign you up for RaDaR if you have not done so already. RaDaR is linked to PatientView where you can see all your blood test results. Follow doctors and patients interested in ADTKD and UMOD on twitter @UMOD_ADTKD