RaDaR
National Registry of Rare Kidney Diseases

UKKA
UK Kidney Association
Rare Renal

Welcome to the Winter 2024 edition of the RaDaR patient newsletter!
We are thankful for your feedback to date. As this is a new format we would also appreciate your feedback on this issue through our three-question feedback form.
We will then incorporate what we can in our next issue.

Contents:

RaDaR facts and figures

34,846 RaDaR patients in the database - up **3,055** in the last year

254 new patients recruited per month (on average)

40,641,851 result and observation records **- 450,000** per month

Top Three recruiting sites:
London - Guy's and St. Thomas' Hospital
Oxford - Churchill
London - Royal Free Hospital

Results are automatically received for **75%** of RaDaR patients

394 primary diagnoses and **176** rare conditions

200 active users on RaDaR

Activity and outputs

Research Publications

Throughout the life of RaDaR, **more than 30** academic papers have been published using its data. 5 have been published this year so far.

We continue to be very grateful to all of you for supporting so much high quality research.

A 2023 paper on IgA Nephropathy, co-authored by several members of the RaDaR team, has been cited **more than 100 times** to date!

IgA nephropathy is a rare kidney disease. Little is known about how likely someone living with IgA nephropathy is to develop kidney failure over their whole lifetime. This study found that people living with IgA nephropathy who agreed to join RaDaR had a high risk of reaching kidney failure in their lifetime, and this was higher in patients with higher levels of protein in their urine (proteinuria). The findings in this paper suggest that more needs to be done to diagnose and treat patients with IgA nephropathy earlier.

Links to RaDaR papers and posters can be found on the **Research page of the RaDaR website**.

Patient surveys

Thank you so much to everyone who responded to one or both of our recent patient surveys. We were overwhelmed by the number of people who so generously took the time to fill them out.

We had **3,867 responses** to the survey on loin pain. The initial results will be presented at the 2025 World Congress of Nephrology.

We found that loin pain is common – it was reported by **62% of respondents**. We found that people who are female, have a higher BMI, are from more socially deprived areas, and have metabolic or cystic kidney disease are more likely to report having experienced loin pain. Most people described their pain as aching. We will be doing more in depth investigations into the data over the next few months.

Our survey on reproductive health and kidney function has just closed with nearly **2,100 responses** – another fantastic response rate! We are excited to see what the data shows us.

Upcoming events

- **UK Kidney Week** - 10-12 June 2025. Registration is open now and concessions are available for patients. There will be a session on rare kidney diseases, and other short talks and posters on RaDaR research.
- Check out the **Rare Renal Events calendar** on the RaDaR website for more upcoming events.

Ways to get involved

- Next year we want to create a survey that asks about your experiences of kidney disease and how it affects your life. We will consult with patients when developing the survey. We will be in touch with opportunities to participate in this.
- **Nephrotic Syndrome (UK) App** - An app designed to help you manage your condition, share your test results with medical staff, share information if you take part in research, and keep you up to date with news and events. Available for Android & iOS.

RaDaR at UK Kidney Week - 11-13th June 2024

UK Kidney week saw **three** abstracts presented by members of the UKKA RaDaR team:

How the UK National Registry of Rare Kidney Diseases (RaDaR) can inform clinical care via interventional studies
Danny Gale

Natural History of Idiopathic Nephrotic Syndrome: The UK National RaDaR Idiopathic Nephrotic Syndrome Cohort
David Pitcher

Clinical characteristics and long-term outcomes of 287 C3 glomerulopathy and immune complex MPGN patients from the UK National Registry of Rare Kidney Diseases (RaDaR)
Sherry Masoud

UKKW 2024 abstracts can be found here
Full poster information can be found here

Update from the RaDaR Patient Representatives Day - 12th July 2024

The day started with a presentation from **RaDaR Director Danny Gale**, highlighting the challenges in studying and developing new treatments for rare diseases. RaDaR helps address some of these challenges by bringing together everyone with a rare disease into one database. RaDaR has information about how diseases affect people over their whole lives, which is important in understanding whether a treatment is working.

This led into the next talk, by **Dr Hannah Beckwith**, about the **FREDA study**. The FREDA study is looking into reproductive health and kidney disease. It focuses on two areas - the effects of menopause on kidney disease, and the health of children born to mothers with kidney disease. Very little is known about either of these areas. The researchers are using a survey - recently sent to RaDaR participants - to help them to understand more.

The third presentation was about the **Kidney PREM** - an annual, national survey of the experiences of people with kidney disease. This survey has run for a number of years. We want to learn from the PREM and develop a survey for RaDaR that captures the experiences of people with rare diseases.

Finally we had a talk from an organisation called **Beacon for rare diseases**. Beacon supports people who want to set up rare disease patient groups. They offer support, training and mentorship, which many of the patient representatives in the audience were very interested in. Most patient groups are made up of volunteers, who are doing the best they can in their spare time with no funding.

It was a very interesting day and we look forward to doing it again next year.
You can see the PDFs of the presentations on the RaDaR Events page.

Rare disease spotlight - Alport Syndrome

Alport Syndrome is a rare genetic disorder of the glomerular basement membrane (part of the kidney's filtration system). It is the **second most common** cause of inherited chronic kidney disease.

People with severe Alport syndrome are born healthy but can develop deafness and kidney failure in their teens to twenties. Other symptoms include high blood pressure and minor eye problems in later life.

Alport Syndrome can be diagnosed by blood and urine tests (a kidney biopsy is sometimes also required). If the condition is detected at an early stage it may be possible to **slow down your rate of kidney disease** by using drugs called ACE Inhibitors to lower blood pressure. Transplanted patients usually do very well.

Updates from the RDG leads - Susie Gear and Neil Turner

Alport was one of the first Rare Disease Groups in 2011. The combined group of patients and clinicians established Alport UK in 2012. In a deliberate early collaborative leap, the charity designed and ran an international symposium in Oxford in 2014 bringing together patients, doctors, and researchers. The workshop atmosphere was powerful, and following it, research competitors became collaborators, and there have been 7 more workshops.

We have worked together so closely that there is no real separation between the RDG and the charity. We also play a leading role in the international Alport Alliance that now organises international workshops (Cyprus 2024, Beijing 2025). The leaders of the charity and the RDG have written this together:

Susie Gear comes from a multi-generation Alport family. Two of her sons have recently had kidney transplants. She has a background in business consultancy and is CEO of Alport UK.
Neil Turner is professor of nephrology in Edinburgh with special interests in basement membranes, young people moving into adult services, and genetic and glomerular diseases.

Major achievements of the group:

- Valuable results are now emerging from the data on over 1,000 Alport patients recruited to RaDaR – an achievement that is envied internationally.
- We have shown that we can use RaDaR to identify and contact people who might participate in tightly targeted research opportunities, for instance with a particular type of mutation, and a particular stage of kidney disease. This ability is almost unique.
- Massively enriched collaboration between researchers, clinicians, and patients have led to greatly increased interest from other researchers, research funders, and drug companies. Many have been drawn in by both the collaborative energy in the community, and the opportunities that RaDaR and the group creates to scope and test new treatments.
- The Stoneygate Trust-Kidney Research UK 'Alport Hub' attracted over £3M funding across a wide range of collaborative projects. This would not have been possible without the collaboration we have developed. It is being looked at as a model for other rare diseases.

Our aims for the next 2-3 years:

- Enrol more participants, and enrich the data we have on each. We expect the first Alport scientific publications using RaDaR to come out soon.
- We are focusing on identifying those at high risk of worsening so that we can target treatments better.
- Find out why only a small proportion of people with just one copy of a defective Alport gene develop serious kidney disease.
- Education of everyone about the spectrum of Alport, so that people aren't unduly worried, but get the monitoring they need.

Events planned:

- Patient day in Nottingham 26 April 2025
- Alport Research Hub Symposium July 2025
- International workshop in Beijing, 4-7 Sep 2025
- Additional events to enrol patients in a big genome sequencing project coming in 2025

Further info:

- Alport UK provides support and info on events
- Understanding the Alport spectrum
- Finding out you have severe Alport Syndrome – video
- Please do what you can to encourage further enrolment into RaDaR at your centre!

Rare disease spotlight - ADTKD

ADTKD is a group of rare genetic diseases, caused by different abnormalities or mutations, that causes damage to the tiny tubes of the kidneys. The most frequent of these is a mutation on the Uromodulin (UMOD) gene.

ADTKD affects about **1 in 65,000 people** in the UK. In cases where UMOD mutations are the cause of ADTKD, there is a **one in two chance** that a child born to an affected person will develop ADTKD themselves.

ADTKD is considered to be a 'silent' condition. The damage it causes to the kidneys is unlikely to have any obvious symptoms until it reaches a severe level. ADTKD is usually diagnosed between the **ages of 30 and 60**. It is usually diagnosed by chance in blood tests. Gout occurs in **around half** of people with ADTKD.

There is currently no direct cure for ADTKD so treatment focuses on managing the symptoms. Transplantation is very successful in people with ADTKD and the condition does not reoccur in the transplanted kidney.

Updates from the RDG leads - Holly Mabillard and John Sayer

I am **Dr Holly Mabillard**, a kidney registrar, in Newcastle upon Tyne and my PhD, supervised by **Professor John Sayer**, is focused on better understanding how ADTKD-UMOD progresses and identifying factors that might influence this process. By studying both the clinical aspects and the genetic architecture of the condition, we hope to find clues that could lead to new treatment options. My PhD project uses patient DNA, kidney cells from patient urine samples, and blood samples to measure the Uromodulin protein to model ADTKD-UMOD and establish disease markers and treatment targets.

Professor Sayer and I have developed a very close working relationship with ADTKD researchers around the world and we are working together to get closer to treatments for all types of ADTKD.

In July 2024, we held an innovative UK ADTKD-UMOD patient day where people living with ADTKD shared their stories through art and narrative. This was facilitated by an illustrator and creative writing artists thanks to a Tilly-Hale award from Newcastle University and Kidney Research UK. This event generated valuable insights into the patient experience and inspired new research ideas. A patient day report has been generated and is available on the RaDaR website summarising all the educational sessions from this day for those who were unable to attend.

We have published a patient-friendly paper, "UMOD and you! Explaining a rare disease diagnosis", aimed at both patients and healthcare professionals to improve disease understanding and diagnosis. Download it here.

Over the next 2 years our plans include:

- Working with collaborators to establish an international ADTKD registry and biobank (ADTKD-NET)
- Developing a patient network
- Improving UK diagnostics for ADTKD-MUC1
- International dissemination of the patient story to drive research
- Raising awareness of this condition through seminars/webinars/educational tools
- Furthering our research on disease modifiers, treatment targets and disease biology

A poem written by a member at the ADTKD-UMOD patient day:

Dan was born, such a joy
He was perfect in every way,
Such a good young boy,
Until a urine infection was noticed one day.

We had him tested,
Reflux was detected.
Years of managed care
Until he was transplanted.

A UMOD was discovered
The family was all tested.
Angela, Laura, Joe, Dan.
Hoping the disease will be bested.

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For more information about this newsletter or the RaDaR National Registry of Rare Kidney Diseases, please contact:

radar@ukidney.org

A reminder to please complete our three-question feedback form so that we can improve future editions of this newsletter. Thank you!