RaDaR
National Registry of Rare Kidney Diseases

UKKA
UK Kidney Association
Rare Renal

Welcome to the Spring 2024 edition of the RaDaR patient newsletter! This is a new format and we would very much appreciate your feedback in our three-question feedback form.

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## RaDaR facts and figures

**33,503 patients** as at 1/5/24 - up 2,030 since Sep 2023

**250 new patients per month** (on average)

**33 rare disease groups** covering >120 diagnoses

**Three** new rare disease groups:

- **Lupus Nephritis** - open now
- **CAKUT** (Congenital Anomalies of the Kidneys and Urinary Tracts) - open now
- **CMV** (Post Transplant Cytomegalovirus) - opening soon

Results are **automatically received for 76%** of RaDaR patients

**192 results & observations,** and **40 million** separate result & observation records

**230 active researchers** across the UK adding data every month

## Activity and outputs

### Research publications

A paper on RaDaR has been published in The Lancet, one of the world's most prestigious journals.

The research found that, over 5 years, **people with rare diseases are 28 times more likely to face kidney failure** than those in the general population with chronic kidney disease, yet are less likely to die before needing dialysis or a kidney transplant.

This will raise the profile of rare kidney diseases internationally, and would not have been possible without the data that you have provided. Thank you so much for your participation.

### System developments

This year will see a complete rewrite of the RaDaR database to improve security, functionality, and maintainability.

### Patient surveys

We recently purchased a licence for Jisc online surveys. Jisc offers simple and safe collection of data and was created specifically for education and research institutions.

In March we launched **our first survey, on loin pain** - a symptom of some kidney diseases. We do not understand what causes this pain, how it impacts everyday life, nor how it can be managed. We have nearly 3,000 responses from patients in specific RaDaR groups so far and will roll this out in stages to all RaDaR patients.

Our **second survey** is about the health and development of children whose mothers had **kidney disease during pregnancy**. Current information suggests that these children do not have extra health problems, but we need more information. This study will help us to advise people with kidney disease who are thinking about pregnancy.

## Upcoming events

- **Patient representatives' day 2024** - We are busy planning this year's event. Invitations will be sent out to patient representatives soon.
- **UK Kidney Week** - 11-13 June 2024. Registration is open now and concessions are available for patients. There will be a session on rare kidney diseases, and two talks on RaDaR research.
- Check out the **Rare Renal Events calendar** on the RaDaR website for more upcoming events.

## Ways to get involved

- **Loin pain survey** - ADPKD, INS, Membranous Nephropathy and Vasculitis patients please check your inboxes. We will invite further groups in the next few weeks. **Please respond even if you don't have loin pain** - your response is just as important as the response of someone with loin pain.
- **Pregnancy and CKD survey** - Coming soon.
- **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.

## Update from the RDG Leads Day - 2 Feb 2024

**Over 40 leads and patient charity representatives** attended the 2024 Rare Disease Group Leads meeting.

There were presentations on current and upcoming research publications, an update on the success of a recent patient survey, and a patient representative's perspective.

We also reviewed the Rare Disease Strategy, published in 2010. The strategy originally outlined the need for two key developments:

1. Development of a rare disease registry.
2. Development of rare disease groups to bring together clinical expertise.

We took a moment to celebrate the huge success of RaDaR to date. **RaDaR is the largest rare kidney disease registry in the world** boasting **>33,000 patients recruited from 109 sites** incorporating **>120 rare diseases within >30 rare disease groups.**

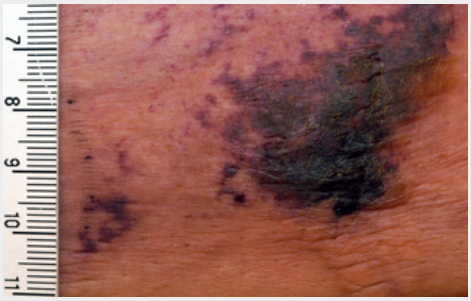
No registry can function without patient support and data provision – **it is because of every RaDaR participant that we have come this far and are making real changes for patients with rare kidney diseases** so from all of us at the UKKA – a huge thank you for your ongoing support!

We are writing a plan for the next 5 years that will include **two exciting new areas of focus for RaDaR:**

**1. Genomics**
Studying all of a person's genes - genomics - can be used to detect and diagnose disease early and tailor healthcare to individuals or groups. **We will bring already available genetic diagnoses into RaDaR** and collect more genome information from our rare kidney disease patients in the future.

**2. Supporting Clinical Trials**
Clinical trials are research studies in which people volunteer to help find answers to specific health questions. **£10m of funding has been invested to set up a new rare kidney disease research centre** and RaDaR will be a major collaborator. As part of this, several research nurses will be improving the data held in RaDaR. Having higher quality data will allow us to support clinical trials and identify areas where new treatments are needed.

## Rare disease spotlight - Calciphylaxis



**Calciphylaxis is most often seen in people receiving dialysis.** It sometimes occurs in people who do not have kidney disease. Small blood vessels in and around the skin become blocked by a build-up of calcium products. The blockage leads to a lack of blood supply. This causes **painful skin ulcers which can lead to serious infections** that require hospitalisation.

Image: Calciphylaxis on a patient's abdomen, by Niels Olson. Licence: Creative Commons Attribution-Share Alike 3.0 Unported.

**The cause of calciphylaxis is unknown**. Possible risk factors (things that may contribute to the development of calciphylaxis) include: warfarin use (a blood thinning medication); long-term dialysis; high blood levels of calcium and/or phosphate; and being overweight.

Calciphylaxis commonly affects the abdomen and legs but can affect any area of the body.

Some people make a good recovery, but **sadly around 5 out of 10 people die within a year of diagnosis**.

**There is currently no specific treatment**. A research study looking at the experiences of patients is underway, with the aim of finding better ways to support people with calciphylaxis. There are also plans for the UK to participate in an international study investigating which treatments have the best outcomes for people with calciphylaxis.

**There are 63 people with calciphylaxis in RaDaR** as at 1/4/24.

## Who are the RDG leads for calciphylaxis?

I'm **Smeeta Sinha**, a kidney consultant from Salford, and my research has focused on **trying to understand why some people develop calciphylaxis**. The UK kidney community, and particularly our patients, have contributed a lot to our understanding of the condition. I hope that we'll eventually find a treatment for the disease.

I'm **Sharon Huish**, kidney dietitian. **The many unknowns and the devastating impact of calciphylaxis** sparked my interest and led to me becoming RDG co-lead.

We support calciphylaxis research in the UK and have put together patient information for RaDaR, Kidney Research UK, Kidney Care UK and the National Kidney Federation. In 2023 we had a session at UK Kidney week to promote awareness and update health professionals' knowledge.

**Over the next 2 years** our plans include:

- Working towards an **international rare disease registry for calciphylaxis**.
- Bringing **calciphylaxis research trials** to the UK.
- Developing a **patient support group/network**.
- Exploring **kidney counselling/psychology services** (and providing education).
- Further developing the available **patient information**.
- **Raising awareness** of, and improving recruitment to, RaDaR.

**We're undertaking a research study** interviewing patients who have, or had, calciphylaxis (within the past 3 years); **if interested please email DRRC@nca.nhs.uk**.

## Patient perspective on calciphylaxis

When my partner, Russell, developed Kidney Disease in late January 2021, we had no experience or knowledge of kidney problems and there was a lot to take in.

He had started to develop major ulcers on his legs which grew and multiplied. Gradually, he became unable to lie down without extreme pain. No-one understood exactly why.

I now know that these are both key symptoms of Calciphylaxis but at the time, the hospital could not determine why he was not responding to any treatment and none of the painkillers worked.

Because of its rarity, diagnosis of Calciphylaxis was very late and he died 5 months later, aged 60.

We had both retired five years before and had moved out of London after many years of hard work and long hours. I had not expected to be on my own again so soon. I can't even put into words how it has changed my life.

I came across Professor Sinha and Sharon Huish's work when I was looking for a way to donate towards Calciphylaxis research. Sharon told me about RaDaR, and I attended a patient/carer representative meeting, where I learnt about many other rare kidney conditions and heard stories and experiences that echoed many of my experiences. Notably, that often a possible diagnosis had initially been as a result of a carer and/or loved one scouring the internet in desperation.

Thanks to those who set up RaDaR we have a national record of patients who have been diagnosed with these many rare kidney diseases. There is a lot of work still to do but at the most recent meeting I attended, it was clear that progress is continually being made to gather more patient data – both national and international. Without enough data, research is held back and the patient/carer experience, the late diagnosis, treatment and the mortality rate for those with Calciphylaxis will not improve. In hospital, no-one told us about RaDaR or asked Russell to give consent for his data to be used. This stage is vital in the collection of data for Calciphylaxis, and other rare kidney conditions, and RaDaR works to make that happen.

I miss Russell every day. I continue my bereavement counselling and am doing my best. But my life will never be the same without him in it.

Joanna Biggs

We are exceptionally grateful to Joanna Biggs for providing her and Russell's story, and for her openness and honesty about the impact that rare kidney disease continues to have on her life.

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

radar@ukidney.org

https://ukkidney.org/rare-renal/contact-us/form

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