■ Light micrograph of a section through a kidney affected by membranoproliferative glomerulonephritis, one of the first rare kidney diseases piloted on RaDaR

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What I tell my patients about the Registry for Rare Kidney Diseases

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Rare diseases are common. There are several thousand conditions overall, and it has been estimated that 5–10% of the population suffer from a rare disease. However, these are often complex in nature, as well as difficult to understand and treat, so the Renal Association has introduced a rare disease strategy, at the centre of which sits the Registry for Rare Kidney Diseases (RaDaR).

You have been diagnosed with a disease that is described as 'rare'. A rare disease is defined as having such a low incidence (rate of occurence) that it cannot be effectively managed on experience drawn from one, or even a few, medical centres. Most doctors are unlikely to have much experience of it, and will rely heavily on external information to offer advice or treatment. The problem is amplified because good-quality clinical and scientific information is difficult to capture, or may not exist. Many rare diseases are complex and have genetic or metabolic causes. Treatment is less likely to be developed and tested than in more common disorders.

Being diagnosed with a rare disease often brings a sense of isolation. Sometimes, the diagnosis is slow to be recognised, and this may reduce your confidence in the healthcare system. Standards of care can vary depending on local expertise. You may scan the internet for scraps of information or contact with other patients. This can be demoralising if the information is misleading, as is sometimes the case. There is a dearth of sound literature for specific patient groups, largely because of the scarcity of reliable clinical scientific publications on which to base it. For some of these conditions there are already patient support groups, but for many, there are not.

What is the solution? We need better co-ordination of rare diseases at a national level. As a principle, there should be integration of diagnostic and treatment services, audit and research, and patient information and empowerment. For individual patients, the best outcomes are obtained when you are well informed and take an active interest in your own management – in partnership with well-prepared, proactive teams of caregivers.

What is RaDaR?

An essential first step towards understanding and optimising management of any rare disease is the collection of informative clinical data. This has been difficult to do in the past in a co-ordinated manner across all renal centres in the UK and has, therefore, led to the creation of RaDaR (see Box 1).

RaDaR is a web-based portal that any healthcare professional or patient can access at any time, with centralised key information. The idea is that this benefits everyone involved in your care.

- **The patient** – you can see your clinical information, enter your own data (where permitted) and access information about your disease that is written by experts and kept constantly up to date.
- **The clinician** – can easily access and update information about any patient in their centre with a rare disease. They can access disease management protocols, diagnostic tests and the latest research.
- **The researcher** – has (anonymised) access to clinical and, where appropriate, biological data on all patients in the UK with the rare disease they specialise in. This allows sufficiently powered studies to be carried out, thereby maximising the potential of that information, and also gives the ability to identify and approach patients in the UK who may be suitable for future trials and studies.

RaDaR operates within the UK Renal Registry, a highly sophisticated database for patients with end-stage renal failure. Here, the existing technical expertise, management and governance structure will help to ensure success. Figure 1 shows the enrolment process for RaDaR.

How is RaDaR accessed?

The registry is organised into three websites, designed to be accessed by different stakeholders.

- Renal RaDaR (www.renalradar.org) is the site for professionals – clinicians and researchers. They obtain unique log-in details from this site and can then enter patient clinical data, as well as access disease-specific information.
- Renal PatientView (www.renalpatientview.org) is the site for consenting patients. You can view your clinical information, including your latest test results (if your renal unit system is linked up) and get information about your disease or diagnosis.
- Rare Renal (www.rarerenal.org) is the public face of the rare disease initiative. It provides information about all aspects of rare kidney diseases, including patient and clinician information, and details about rare disease groups (RDGs).

What are RDGs?

To bring together all the appropriate experts for each disease, the Renal Association commissioned RDGs. These are designed to ensure a holistic approach to clinical care and champion the cause of patients with a specific disorder. The groups are professionally led, but include patients and your representatives, and involve you in the process. For some professionals, their involvement will reflect a research interest, while others may bring expertise in different areas, such as patient education, clinical audit or specialised diagnostic services. The working groups are also ideally placed to design optimal patient information materials.

A list of current RDGs is provided in Box 2, and we encourage more professionals and patients to form RDGs for diseases that are not yet represented.

Box 1. Summary of RaDaR

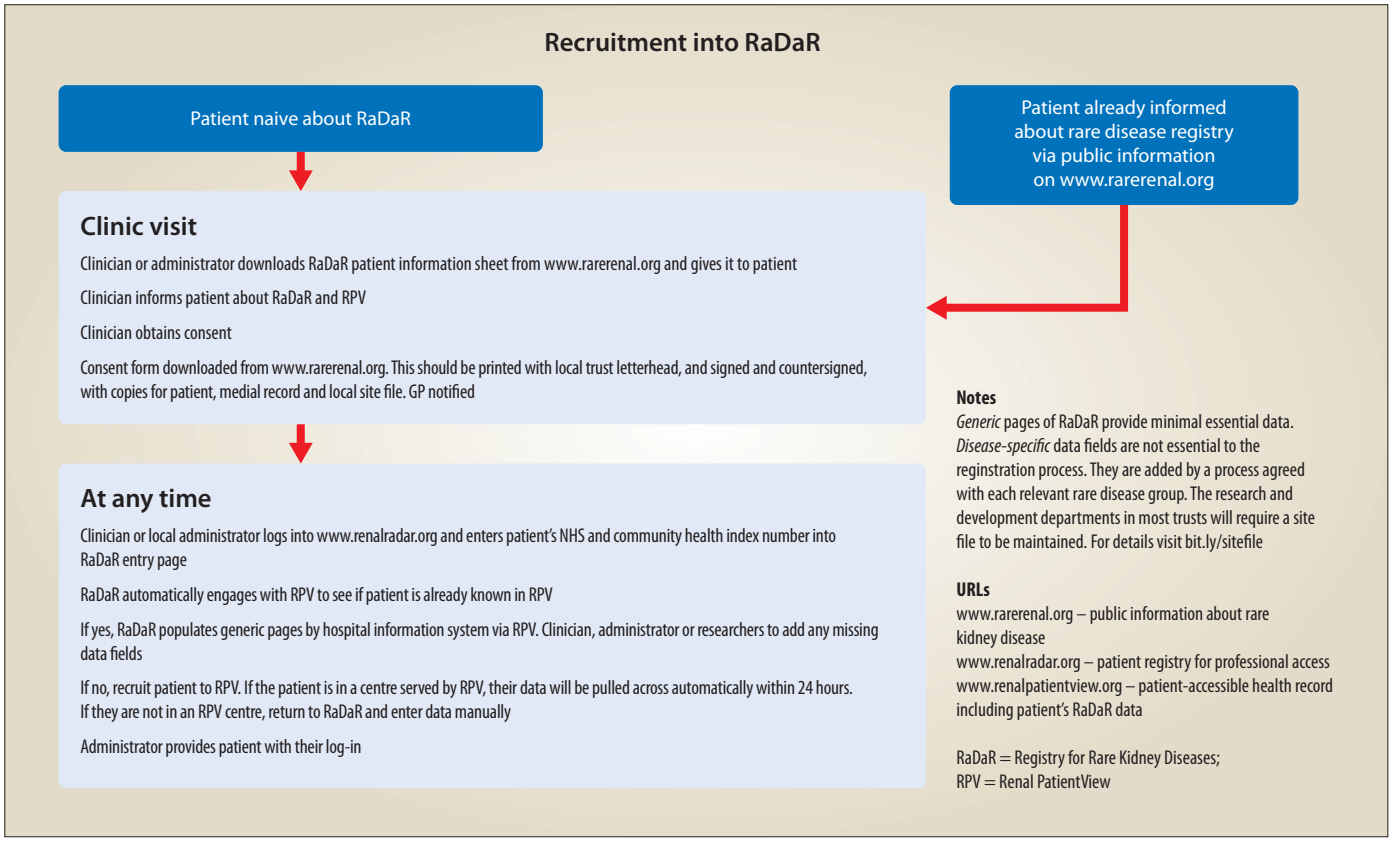
- The Registry for Rare Kidney Diseases (RaDaR) is a web-based national registry designed to collect and utilise patient information on renal rare diseases
- Rare disease groups are formed by enthusiastic experts and patients related to a specific diagnostic group
- Patients, clinicians and researchers are all key stakeholders in the information gathered
- Clinical information is uploaded by registered healthcare professionals and renal IT systems
- An important benefit is to facilitate patients' involvement in research studies by connecting you directly with research groups
- All renal units in the UK can participate in RaDaR

Box 2. Current renal rare disease groups

- Alport syndrome
- Adenine phosphoribosyltransferase deficiency
- Bardet–Biedel syndrome (UK nationally commissioned service)
- Bartter syndrome (Types 1, 2 and 4)
- Bartter syndrome (Type 3)
- Cystinosis
- Cystinuria
- Dense deposit disease
- Dent's disease
- EAST (epilepsy, ataxia, sensorineural deafness and tubulopathy) syndrome
- Familial uromodulin-associated nephropathy (hyperuricaemic nephropathy and medullary cystic kidney disease)
- Gitelman syndrome
- Haemolytic uraemic syndrome (atypical)
- Haemolytic uraemic syndrome (shiga toxin-associated)
- Hepatocyte nuclear factor-1-beta mutation
- Hyperoxaluria (primary hyperoxaluria, oxalosis)
- Liddle's syndrome
- Lowe syndrome
- Membranoproliferative glomerulonephritis
- Membranous nephropathy
- Nephrotic syndrome (steroid-resistant, congenital or associated with primary focal segmental glomerulosclerosis)
- Autosomal recessive polycystic kidney disease
- Pregnancy and chronic kidney disease
- Vasculitis

What is required of your clinician?

Your clinician is responsible for identifying patients with a rare disease (in conjunction with local research nurses if available) and providing you with the initial information (by post or during a routine clinical encounter). All patient information sheets and consent forms are available from the Rare Renal website (http://rarerenal.org/radar-registry/criteria-and-consent) and can be downloaded and printed during the clinic visit. You read and sign the consent form, which allows your data to be entered on to the website. Your clinician will file the consent form in your notes and give you a copy. At that time, they will



■ **Figure 1.** Enrolment process for RaDaR

also ask you for your email address, which is entered into the site before automatically sending you a log-in name and password for you to enter the site and see your details there yourself.

What will you be consenting to?

According to the disease group you conform to, you may be asked to sign more than one consent form. The first consent form, Part One Consent, allows any clinical data about you to be entered on to the website. This can be most easily defined as anything that might appear in your hospital notes, as well as results stored on your local hospital pathology database (such as blood or biopsy results). The next level, Part Two Consent, is requested if there is a specific research study under way into your disease (for example, genetic studies or a clinical trial).

Apart from you, only professionals who have obtained a RaDaR log-in from your hospital will be able to see Part One information. Researchers from your individual RDG will also be able to see information about all patients with your disease on RaDaR (Part One and Two), but will not be able to link this to your personal (identifiable) details. Thus, your personal confidentiality is protected at all times.

Are there any Part Two research studies ongoing?

The two pilot diseases in RaDaR were those for steroid-resistant nephrotic syndrome in childhood (SRNS) and membranoproliferative glomerulonephritis. Both were set up with Part Two studies from the outset. For SRNS, the research question was to collect DNA at the time of patient recruitment to analyse genetic risk of disease in detail, using the most up-to-date genetic sequencing technology – next generation sequencing (NGS). In addition, blood is collected during times of disease relapse to develop new laboratory tests that can predict disease activity and the risk of disease recurrence after kidney transplantation.

These studies illustrate the central importance of genetics in rare disease. Many rare renal disorders are primarily genetic in origin. Over the past decade, there has been remarkable expansion in the identification of specific DNA abnormalities involved in a wide variety of these conditions. This not only has the potential to lead to changes in the management of individual conditions, but also has major implications for preventive intervention for at-risk family members, whose needs in terms of preventive care have never been systematically addressed. It also prompts new research.

To illustrate the success of the strategy, the SRNS study has resulted in the first clinically approved NGS test for SRNS, to diagnose monogenic (single-gene mutation) forms of the disease by testing 37 genes simultaneously and rapidly in a single test (www.nbt.nhs.uk/genetics).[1] This has become available and can be commissioned for all patients in the UK, showing a diagnostic benefit to patients that stems directly from the RaDaR initiative.

This study began in children and has now been funded to extend to adult patients with SRNS, so that eventually all SRNS patients in the UK will be

recruited. This will allow for lifelong follow-up, adding considerable strength to our quest to fully understand this difficult disease.

What is the future of RaDaR?

RaDaR was commenced in all the paediatric renal units in the UK and is now in the process of being rolled out to all adult units as well. This means that any adult unit that has informed their local research and development department about RaDaR can start to recruit patients to any of the RDGs currently listed (see Box 2). This is for Part One Consent (the registry), and any RDGs that design Part Two research studies need to complete the additional steps of ethical approval and local site approvals, as with any research study.

The vision is for RaDaR to collate information about patients on a lifelong basis, with regular clinical updates. It will be up to individual RDGs to take this forward, with more sophisticated developments as funds and enthusiasm permit. For example, it may be possible for patients to enter your own data and information on to the site, such as home blood pressure recordings, or to complete questionnaires about your disease or lifestyle.

The rare disease strategy has the potential to transform the way complex rare conditions are approached. This presents a tremendous opportunity for all of us to learn about and manage rare diseases using an improved and co-ordinated approach from what has occurred to date. This will bring benefits at all levels if it can be broadly supported and integrated into patient care ■

Declaration of interest
None declared.

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Key points

- **Most doctors are unlikely to have much experience of managing rare diseases and treatments are often less developed.**
- **Patients may feel isolated and search the internet for what is frequently misleading information.**
- **RaDaR is a web-based portal which collects clinical information and can be accessed by patients, clinicians and researchers.**

■ *What I tell my patients about* ... is a patient information service specifically designed for renal units to use with their patients. You can now view this, and all of the previous *What I tell my patients about* ... articles, online and download them free of charge at **www.bjrm.co.uk/patient-information**

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Most people would agree that publicity for organ donation is a good thing, especially when you learn that some parts of our community lag far behind the rest in signing up to the organ donor register. Black, Asian and other minority ethnic groups are a prime example. Those groups are four times more likely to suffer renal failure, yet four times less likely to sign up to the register.

What then do we make of the recent episode of the BBC TV series, Holby City, aired on 2 August 2013? The story cut through all the checks and balances so carefully put in place by the service to ensure that the system of transplantation is not abused – checks and balances which had been explained carefully to the programme's researchers beforehand by NHS Blood and Transplant (NHSBT).

The episode portrayed a doctor so determined to go ahead with the transplant for her patient that she ignored every rule in the book. I am led to believe that, following the broadcast, many people withdrew their names from the register. NHSBT thought that immense harm had been done and said so publically to the BBC.[1]

Now we have a transplant story in EastEnders, running since 20 September 2013, but this time the BBC seems to have learnt its lesson. Well before the first episode was filmed, the programme's researchers contacted the NKF to check the potential storyline – episode by episode and scene by scene.

This was very gratifying, as was the opportunity afforded to the NKF to take an active part in the social networking websites and blogs that began discussing this story almost immediately. Currently, these forums show the interest in transplantation among the public to be huge.

The NKF felt that it would have been helpful to viewers to broadcast the NKF helpline number at the end of the episode; however, this is not currently BBC policy and so the request was declined ■

Timothy F Statham OBE,
Chief Executive

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