UKKA
UK Kidney Association

RaDaR
Registry of Rare Kidney Diseases

# Training

## 2. Rare Renal Website

# Information portal for patients, clinicians and researchers

- Patient Information
- Clinician Information
- Rare Disease Groups
- Recruitment Resources for sites

- RaDaR Research Papers and Posters
- Metadata / Data Dictionary / Application for RaDaR Analysis
- Patient Newsletters
- Rare Renal Events
- Glossary

UK Kidney Association

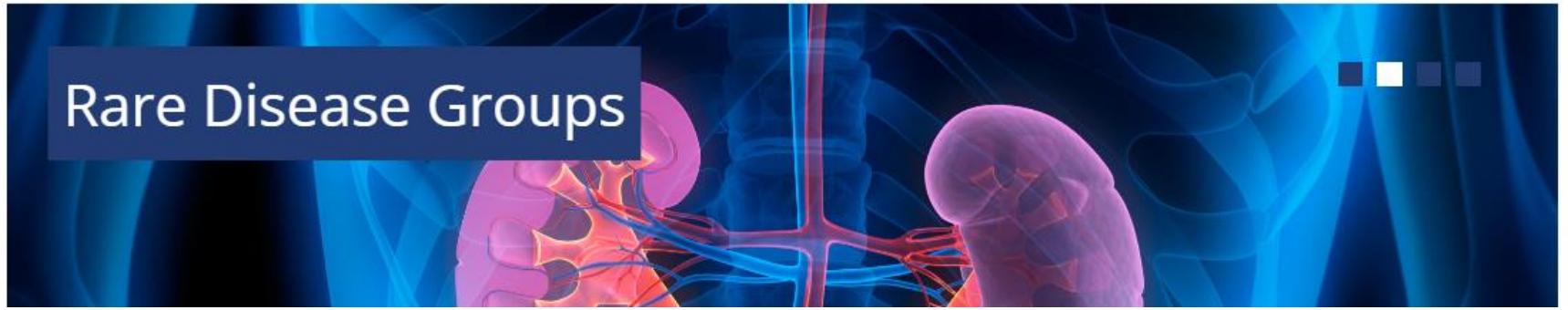
RaDaR
National Registry of Rare Kidney Diseases

Home | Patient Information | Clinician Information | Rare Disease Groups | RaDaR Research | Registry (RaDaR)

News | Glossary | Contact

# Rare Renal

## Information on rare kidney diseases



UK Kidney Association

RaDaR
National Registry of Rare Kidney Diseases



# Rare Renal

## Information on rare kidney diseases

**Alport Syndrome**

**APRT Deficiency**
(APRT-D)

**Atypical Haemolytic Uraemic Syndrome**
(aHUS)

**Autosomal Dominant Polycystic Kidney Disease**
(ADPKD)

**Autosomal Dominant Tubulointerstitial Kidney Disease**
(ADTKD)

**Autosomal Recessive Polycystic Kidney Disease**
(ARPKD)

**Bartter Syndrome**

**BK Nephropathy**

**Calciphylaxis**

**Congenital Anomalies of the Kidneys and Urinary Tracts**
(CAKUT)

**Cystinosis**

**Cystinuria**

**Dent Disease**

**EAST Syndrome**
(Epilepsy, Ataxia, Sensorineural deafness, Tubulopathy Syndrome)

**Fabry Disease**

**Fibromuscular Dysplasia**

**Gitelman and Type 3 Bartter Syndromes**

**Haemolytic Uraemic Syndrome**
(Shia Toxin Associated: STEC-HUS)

**Hepatocyte Nuclear Factor 1B Mutation**
(HNF1b)

**Hyperoxaluria**
(Primary Hyperoxaluria, Oxalosis)

**IgA Nephropathy**

Hepatocyte Nuclear Factor 1B Mutation
(HNF1b)

Hyperoxaluria
(Primary Hyperoxaluria, Oxalosis)

IgA Nephropathy

Inherited Renal Cancer Syndromes

Liddle Syndrome

Lowe Syndrome

Lupus Nephritis

Membranous Nephropathy

Mitochondrial Disease affecting the kidney
(Mitochondrial)

Monoclonal Gammopathy of Renal Significance
(MGRS)

MPGN, DDD & C3 Glomerulopathy

Nephronophthisis

Nephrotic Syndrome

Pregnancy and Chronic Kidney Disease

Pure Red Cell Aplasia

Retroperitoneal Fibrosis

Tuberous Sclerosis

Vasculitis

# Patient info



## Alport Syndrome

### Patient Information

- How the illness affects people
- What can be done about it?
- Other peoples' experiences
- Patient support group
- How the disease works
- What's new? Opportunities for research and development
- Further Information

# Clinician Info



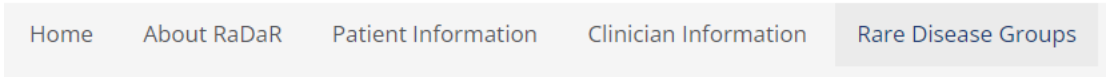

## Alport Syndrome

### Clinician Information

This page focuses on the diagnosis and management of Alport Syndrome in a UK context.

- Prevalence
- Clinical course and prognosis
- Diagnosis
- Management
- Carriers
- Organising local services
- Renal replacement therapy
- Contact us
- Further Information

# Rare Disease Group (RDG) / Cohort




## Alport Syndrome

### Rare Disease Group

- Aims of the Group
- Current Activities
- International Links
- Patient Support Groups
- Reports
- Group Members
- Disclosure of Conflicts of Interest

# Recruitment Resources

- Study Documents (intro letters, PIS and consent forms)
- RaDaR training guidance for sites
- Inclusion/Exclusion Criteria
- Electronic consent guidance

# Research

## RaDaR Research

## RaDaR Research Papers

1. **Effects of rare kidney diseases on kidney failure: a longitudinal analysis of the UK Natio**
   2024 https://www.thelancet.com/journals/lancet/article/PIIS0140-6736(23)02843-X/fulltext )
2. **Distribution of epidemiology and clinical characteristics of RaDaR participants** pre-print
   (https://www.medrxiv.org/content/10.1101/2023.09.24.23296009v2)
3. **Long term outcomes in IgA nephropathy Pitcher et al Clinical Journal of the American Sc**
   https://journals.lww.com/cjasn/fulltext/2023/06000/long_term_outcomes_in_iga_nephropathy.
4. **Clinical Predictors of Long-term Outcomes in C3 Glomerulopathy and Immune-Complex**
   Downward L, Pitcher D, Webb NJ, Proudfoot C, RaDaR Consortium, Wong EK, Gale DP. medRxi

## RaDaR Research Posters

# RaDaR Metadata

## Top level metadata in RaDaR, updated March 2024

| Rare Disease Cohort | Cohort Size | Patients with Lab Results via datafeed | Patients with a recording of RRT | Number of people with creatinine readings (any source) | Mean number of creatinine readings per person |
|---|---|---|---|---|---|
| Alport Syndrome | 1089 | 831 | 420 | 847 | 75 |
| APRT Deficiency | 10 | 6 | 2 | 8 | 80 |
| Atypical Haemolytic Uraemic Syndrome | 303 | 246 | 154 | 252 | 238 |
| Autosomal Dominant Polycystic Kidney Disease | 8614 | 6992 | 3626 | 7180 | 84 |
| Autosomal Dominant Tubulointerstitial Kidney Disease (FUAN) | 253 | 197 | 118 | 200 | 86 |
| Autosomal Recessive Polycystic Kidney Disease/Nephronophthisis | 261 | 207 | 118 | 226 | 103 |
| BK Nephropathy | 96 | 92 | 93 | 92 | 241 |
| Calciphylaxis | 63 | 49 | 57 | 48 | 236 |
| CKD due to Genetic Factors in people of African ancestry | 309 | 150 | 124 | 150 | 131 |
| Congenital Anomalies of the Kidneys and Urinary Tracts (new group) | 6 | 3 | 3 | 3 | 10 |
| Cystinosis | 187 | 156 | 100 | 158 | 139 |
| Cystinuria | 505 | 341 | 10 | 363 | 28 |
| Dent Disease and Lowe Syndrome | 68 | 39 | 17 | 43 | 77 |
| Fabry Disease | 58 | 48 | 26 | 47 | 83 |
| Fibromuscular Dysplasia | 56 | 42 | 0 | 41 | 19 |
| HNF1b Mutations | 107 | 66 | 15 | 70 | 47 |
| Hyperoxaluria | 136 | 108 | 43 | 113 | 80 |
| Idiopathic Nephrotic Syndrome | 4691 | 3464 | 1187 | 3955 | 119 |
| IgA Nephropathy | 4879 | 4144 | 2580 | 4218 | 107 |
| Inherited Renal Cancer Syndromes | 315 | 13 | 4 | 13 | 77 |
| Lupus Nephritis (new group) | 16 | 2 | 0 | 5 | 13 |
| Membranoproliferative Glomerulonephritis | 1216 | 941 | 629 | 1020 | 149 |

# Communications

ISSUE 2

View this email in your browser

RaDaR

National Registry of Rare Kidney Diseases

## RaDaR Newsletter

Welcome to the Autumn 2023 National Registry of Rare Kidney Diseases (RaDaR) newsletter!

We would like to thank you for taking part in RaDaR. The information that you have allowed us to use from your records is continuing to improve national understanding of rare kidney diseases.

### Current recruitment

# Events

## Rare Renal Events

- Patient Representative Meeting 2023
- Annual Rare Disease Group Leads Meeting 2024

## Calendar 2024

- **January**
- **February**
  - 2nd Feb Annual Rare Disease Group Lead Meeting 2024, London
- **March**
  - 2nd March **PKD** patient day, Newcastle
  - 23rd March **HNF1B** Support Day on-line via TEAMS for Patients, families, carers and interested clinicians:
    https://www.eventbrite.co.uk/e/hnf1b-support-day-registration-829918477507
- **April**
  - 27th April **Cystinuria** Patient Day, Newcastle
    https://www.cystinuriauk.co.uk/patient-day
- **June**
- **July**

# Rare Renal Glossary

**Glossary**

**Acute Kidney Injury (AKI)** - a sudden reduction in kidney function. AKI can occur due to s
result of the side effects of some drugs. Sometimes it's due to a combination of factors. Ac
called acute kidney injury it is not caused as a result of a physical blow to the body. Nor is
other organs, and cause you to be dehydrated.

**Amino Acid** - Many amino acids occur in nature. Only 22 are used as the building blocks o
cannot make them ourselves. They link to each other to create chains of amino acids (pep

The body's ability to line up amino acids in the right order to make a protein is governed b
lead to the production of a faulty protein. This in turn gives rise to a structural or functiona

Some amino acids are not used in protein production but have biological effects of their o

An amino acid is defined by its make up with an amine (-NH2) and a carboxylic acid (-COO

**Amniotic fluid** - Fluid that surrounds an unborn baby during pregnancy. It contains protei
protects it from injury by cushioning sudden movements. The amount of fluid in the womb
the lungs are not fully developed.