## Summary of RDG meetings

<table>
<thead>
<tr>
<th>Date</th>
<th>Event Description</th>
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<tbody>
<tr>
<td>16&lt;sup&gt;th&lt;/sup&gt; February 2016</td>
<td>Teleconference on application to NHS England as Highly Specialised Ciliopathy Service</td>
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<tr>
<td>17&lt;sup&gt;th&lt;/sup&gt; July 2016</td>
<td>Kingston at Patient Support Day</td>
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<tr>
<td>16&lt;sup&gt;th&lt;/sup&gt; December 2016</td>
<td>Strategy meeting at ICH</td>
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## Summary of patient events

We held the 5<sup>th</sup> National ARPKD Patient Support Day on 17<sup>th</sup> July 2016 at Kingston University organised by Dr Evi Goggolidou – see attached Programme at end of report

## Grant applications submitted

<table>
<thead>
<tr>
<th>Grant Application</th>
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<tr>
<td>Sparks Charity for Video for Patient Support Day</td>
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<tr>
<td>Kidney Research UK Grant for Patient Support Day</td>
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## Grants awarded

<table>
<thead>
<tr>
<th>Grant Award</th>
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<tbody>
<tr>
<td>Sparks Charity for Video for Patient Support Day (Dr Goggolidou)</td>
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<tr>
<td>Kidney Research UK Grant for Patient Support Day (Tess Harris)</td>
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## Publications and presentations

| IPNA Brazil 2016 – see attached. |
| The National Registry of Rare Kidney Diseases (RaDaR) at Birmingham Children’s Hospital |
| Autosomal Recessive Polycystic Kidney Disease (ARPKD) in the National Registry of Rare Kidney Diseases (RaDaR) |
| Also, Submitted to Renal Association 2017 |
| New Patient Information Leaflet: |
| “Autosomal recessive polycystic kidney disease (ARPKD) - A guide for parents 2017” |

## Highlights

Patient Support Meeting and Presentation at International Conferences

## Problems

No admin support and European and US members not able to join in meetings.
5th ARPKD Family Day 17th July 2016

9.30am - 10am  Coffee and registration

10am - 10.15am  Welcome and Introduction – Dr Evi Goggolidou and Tess Harris

10.15am – 10.30 am  Genetics of ARPKD and an Introduction to the Physiology of the Kidney and Liver – Dr Evi Goggolidou

10.30 am – 11.00am  ARPKD Basics and Clinical Aspects of ARPKD – Dr Larissa Kerecuk

11.00 – 11.15am  Coffee break

11.15 am – 11.45am  The Liver in ARPKD - Dr Marianne Samyn

11.45am – 12.00pm  Patient Story

12.00 - 12.30pm  Research Update - Dr Evi Goggolidou

12.30 – 12.35pm  RaDaR – Dr Larissa Kerecuk

12.35 pm – 2.00pm  Lunch

1.45pm – 2.15pm  Sources of information and how to use your CAB - Geoff Pope

2.15 pm – 3.45pm  World Café Style Workshop Sessions

  Enjoy afternoon tea, coffee and biscuits whilst discussing ARPKD and research etc

4.00pm – 4.15pm  Feedback and close
Autosomal Recessive Polycystic Kidney Disease (ARPKD) in the National Registry of Rare Kidney Diseases (RaDaR)

M. Kokocinska, L. Kerecuk, D. Milford, S. Hulton, M. Muorah, Birmingham Children’s Hospital; M. Dillon, UK Renal Registry

Background
Autosomal recessive polycystic kidney disease (ARPKD) is a rare genetic condition that causes cysts to develop in the liver and kidneys. It is usually first diagnosed in infancy and affects approximately 1 in 20,000 live births. As the condition has multisystem effects, a comprehensive care strategy requires a multidisciplinary team and detailed data collection.

This poster describes data collected by the ARPKD Rare Disease Group via the UK’s National Registry of Rare Kidney Diseases (RaDaR).

Objectives
The ARPKD rare disease group use RaDaR to identify eligible patients for family information slays and to study the progression of the condition. It is intended to be a ready cohort for research into this condition: from observational to interventional studies.

Methods
The RaDaR dataset is defined by the UK Renal Registry in association with over 20 Rare Disease Groups, made up of experts in each eligible condition. Data fields include demographics, blood and urine results, medications, transplant and dialysis history, genetic and co-morbidities. Data is entered retrospectively from the patient’s medical records following consent.

Results
88 ARPKD patients from 29 UK renal units have been consented to date, with an age range of 3 weeks to 65 years. There are 51 (58%) paediatric (under 16) patients with an average age of 3 years 6 months and 37 (42%) adult patients with an average age of 31 years. There are 42 females (48%) and 46 males (52%) males. The first paediatric patient was recruited in October 2012 and the first adult patient in August 2013.

Conclusion
RaDaR provides important epidemiology data on ARPKD patients which is shared amongst the members of the Rare Disease Group to develop further research into this condition and improve the quality of care for these patients.
Overview of Rare Renal Diseases at a UK Paediatric Renal Centre through the National Registry of Rare Kidney Diseases (RaDaR)

M. Kokocinska, L. Kerecuk, D. Milford, S. Hulton, M. Muorah Birmingham Children’s Hospital; M. Dillon, UK Renal Registry

Background
The National Registry of Rare Kidney Diseases (RaDaR) is a UK Renal Association initiative designed to gather information from patients with rare kidney diseases. Recruitment began in 2010 and now covers over 30 conditions. There are over 7,000 recruits from 70 renal units.

This poster describes the range of conditions and patient numbers recruited to RaDaR from Birmingham Children’s Hospital (BCH), a national tertiary renal referral hospital and the leading paediatric recruiter in the UK.

Objectives
BCH use RaDaR to identify eligible patients for family information days and to tailor clinical interventions and access genetic testing services. In the future it is intended to be used to design clinical trials to improve diagnosis and treatment.

Methods
The RaDaR dataset is defined by the UK Renal Registry in association with over 20 Rare Disease Groups, made up of experts in each eligible condition. Data fields include demographics, blood and urine results, medications, transplant and dialysis history, genetics and co-morbidities. Data is entered retrospectively by the patient’s medical records following consent.

Results
289 patients have been consented at BCH to date. The age range is from birth to 16 years with mean of 4.9 years. The male to female ratio is 55%:45%.

The most common condition is Idiopathic Nephrotic Syndrome (n=119; 39%), followed by Alport Syndrome (n=32; 12%), ARPKD (n=24; 9%), Hyperoxaluria (n=23; 9%) and Streptococcal Glomerulonephritis (n=21; 8%).

Conclusion
RaDaR provides important epidemiology data which is shared amongst the renal team to facilitate further research into rare kidney diseases, develop evidence-based clinical guidelines and improve the quality of care for these patients.

RaDaR is supported by:

If you have any questions in regards to this review or the results please contact: Maria.Kokocinska@bch.nhs.uk
Kidney Versus Combined Kidney and Liver Transplantation in Young People With Autosomal Recessive Polycystic Kidney Disease: Data From the European Society for Pediatric Nephrology/European Renal Association-European Dialysis and Transplant (ESPN/ERA-EDTA) Registry

Djaëffa Megahfi, MD, PhD,1,2 Karlijn J. van Straalen, PhD,3 Marjolein Borthuis, PhD,3 Kitty J. Jager, MD, PhD,3 Ayse Balat, MD,4 Elisa Benetti, MD,5 Nathalie Godefroid, MD,6 Vidar O. Edvardsson, MD,7,8 James G. Heaf, MD, DMSc,9 Augustina Jankauskiene, MD,10 Larissa Kerecuk, MBBS, BSc, MRCPCH, FRCPCH,11 Svetlana Marinova, MD,12 Flora Puteo, MD,12 Tomas Seeman, MD, PhD,14 Aleksandra Zuczkowska, MD, PhD,15 Jacques Pirenne, MD, PhD,16 Franz Schaefer, MD,17 and Jaap W. Groothoff, MD, PhD,18 on behalf of the ESPN/ERA-EDTA Registry

Background: The choice for either kidney or combined liver-kidney transplantation in young people with kidney failure and liver fibrosis due to autosomal recessive polycystic kidney disease (ARPKD) can be challenging. We aimed to analyze the characteristics and outcomes of transplantation type in these children, adolescents, and young adults.

Study Design: Cohort study.

Setting & Participants: We derived data for children, adolescents, and young adults with ARPKD with either kidney or combined liver-kidney transplants for 1995 to 2012 from the ESPN/ERA-EDTA Registry, a European pediatric renal registry collecting data from 36 European countries.

Factor: Liver transplantation.

Outcomes & Measurements: Transplantation and patient survival.

Results: 202 patients with ARPKD aged 19 years or younger underwent transplantation after a median of 0.4 (IQR, 0.0-1.4) years of dialysis therapy at a median age of 8.0 (IQR, 4.1-13.7) years. 32 (15.8%) underwent combined liver-kidney transplantation, 163 (80.7%) underwent kidney transplantation, and 7 (3.5%) were excluded because transplantation type was unknown. Age- and sex-adjusted 5-year patient survival posttransplantation was 95.5% (95% CI, 92.4%-98.8%) overall. 97.4% (95% CI, 94.9%-100.0%) for patients with kidney transplantation in contrast to 87.0% (95% CI, 79.8%-93.8%) with combined liver-kidney transplantation. The age- and sex-adjusted risk for death after combined liver-kidney transplantation was 6.7-fold (95% CI, 1.6- to 25.4-fold) greater than after kidney transplantation (P = 0.005). Five-year death-censored kidney transplant survival following combined liver-kidney and kidney transplantation was similar (92.1% vs 85.9%; P = 0.4).

Limitations: No data for liver disease of kidney therapy recipients.

Conclusions: Combined liver-kidney transplantation in ARPKD is associated with increased mortality.
Autosomal recessive polycystic kidney disease (ARPKD) - A guide for parents

This information sheet is about autosomal recessive polycystic kidney disease (ARPKD). It is intended as a general guide for:

- Expectant parents who have been told that their unborn child has or may have ARPKD
- Parents of young babies and children who have been diagnosed with the disease
- Couples who are thinking about having a baby and have been told any children they have could have ARPKD.

It explains the causes of ARPKD, how it is diagnosed, its symptoms and how they are treated, and how the disease might progress.

What is ARPKD?

ARPKD is a rare disease that affects the kidneys and liver [Guay-Woodford 2014]. It is usually diagnosed in babies and young children and occurs in roughly one in every 20,000 live births in the UK [Guay-Woodford 2014]. Sometimes only the liver is affected; this condition is called congenital hepatic fibrosis (CHF) [Guay-Woodford 2014]. This information sheet, however, is about ARPKD when both the kidneys and liver are affected.

ARPKD causes cysts - sacs filled with fluid - to develop in the small tubes of the kidneys [Guay-Woodford 2014; Buscher 2014]. These tubes produce and transport urine. ARPKD also causes problems with liver, including the formation of cysts, scar tissue (called fibrosis), and a swollen bile duct [Guay-Woodford 2014; Buscher 2014; Chandler 2015]. The bile duct is a tube involved in producing and transporting bile, a fluid that helps in digestion.

Although ARPKD affects the kidneys and liver, the immediate risk to about a third of babies born with the disease is lung under-development [Buscher 2014]. This can make it difficult for the baby to breathe and mean they need emergency breathing support from a machine (ventilation) [Bergmann 2015]. Over time causes damage to the kidneys, stopping them working properly and eventually leading to kidney failure, often in childhood or young adulthood [Hartung 2014]. It can also cause complications in the liver and bile duct, such as infections [Buscher 2014].

ARPKD symptoms, severity of disease and the age that problems occur vary.