Autosomal Recessive Polycystic Kidney Disease (ARPKD) in the UK National Registry of Rare Kidney Diseases (RaDaR)

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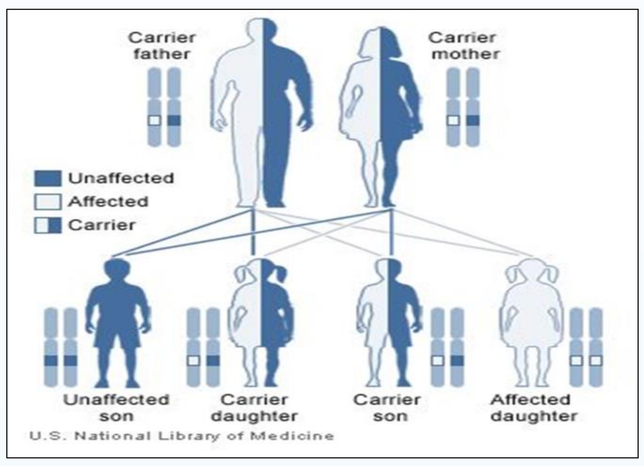
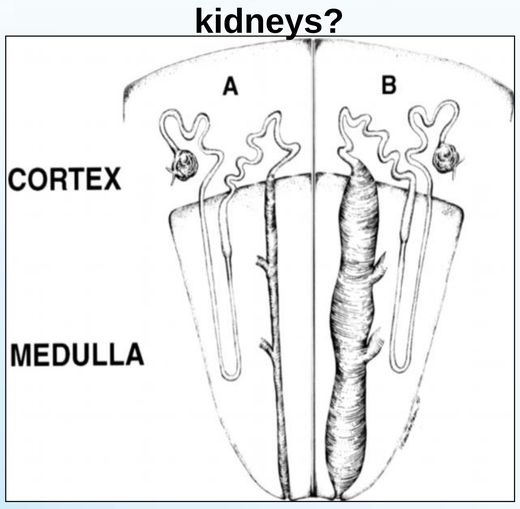
Background

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 (1). As the condition has multisystem effects, a comprehensive care strategy requires a multidisciplinary team and detailed data collection. We describe data collected by the ARPKD Rare Disease Group via the National Registry of Rare Kidney Diseases (RaDaR) in the UK.

How is ARPKD inherited?



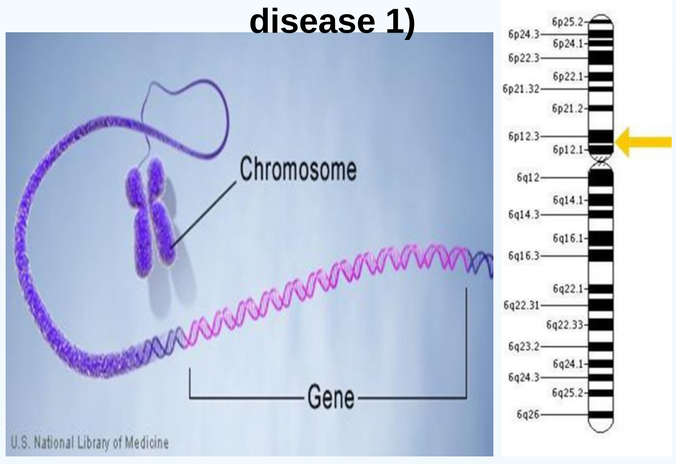
How does it go wrong in the kidneys?



Lonergan G J et al. Radiographics 2000;20:837-855

Collecting duct dilation in ARPKD

What causes ARPKD? Defect in a single gene PKHD1 (Polycystic kidney and hepatic disease 1)



Objective

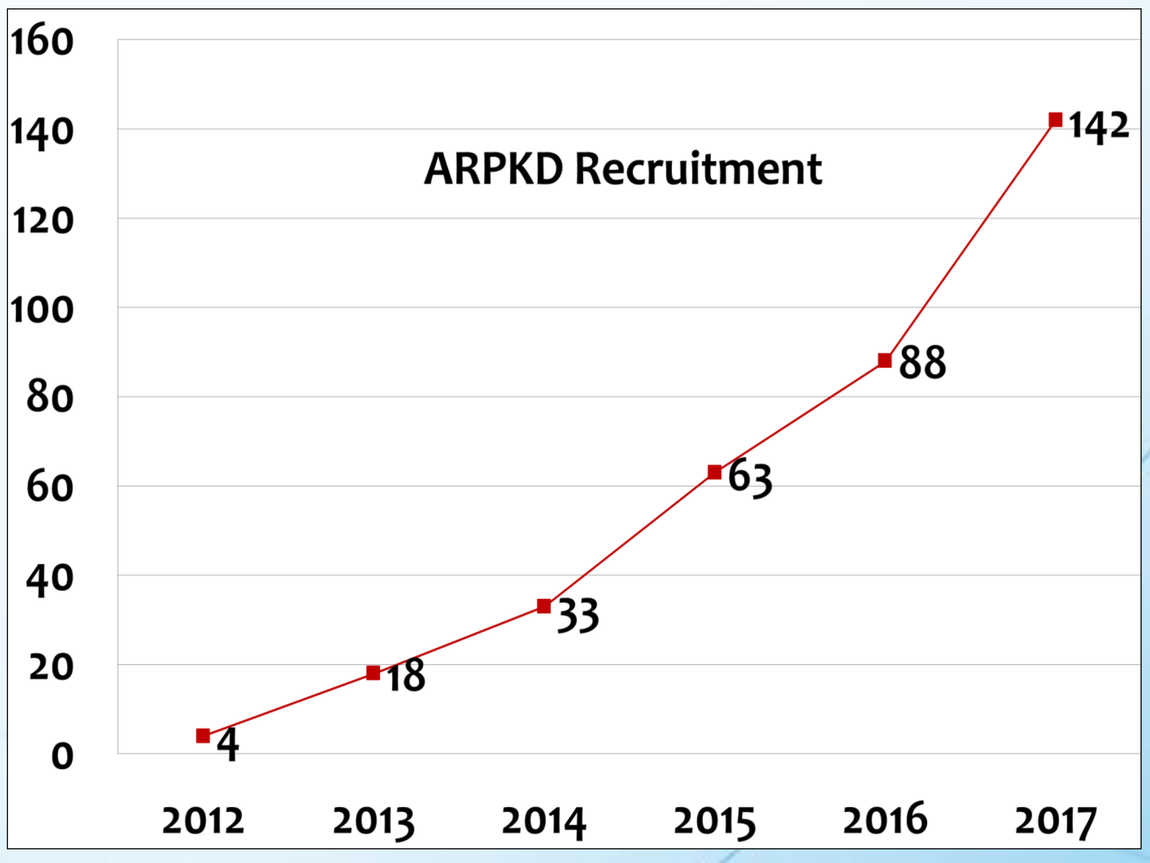
The ARPKD rare disease group use RaDaR to identify eligible patients for family information days and to study the progression of the condition. It is intended to be a ready cohort for research into this condition: from observational to interventional studies.

Methods

The RaDaR dataset is defined by the UK Renal Registry in association with over 25 Rare Disease Groups, made up of clinicians, scientists and patient's representatives 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 from the patient's medical records following consent.

Results

142 ARPKD patients from 35 UK renal units have been consented to date, with an age range of 5 months to 84 years. There are 58 (40.8%) paediatric (under 16) patients with an average age of 8 years 5 months and 84 (59.2%) adult patients with an average age of 40 years. There are 73 females (51.4%) and 69 males (48.6%) males. The first paediatric patient was recruited in October 2012 and the first adult patient in August 2013.



Conclusions

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. The fact that we have so many adult patients shows that this is no longer just a paediatric disease and with better paediatric care these patients are now becoming adults. We have well established links with the US based Hepato/Renal Fibrocystic Diseases Core Center **(UAB HRFDCC)**, the European Renal Reference Network ERKNET and the ARegPKD Registry (based in Cologne), in order to increase the international cohort of patients with ARPKD available for research.

RaDaR is supported by:

References:
(1) http://pkdcharity.org.uk/about-arpkd/diagnosis/arpkd-a-guide-for-parents?gclid=CK7W6_OOrNQCFQaNGwodtOoALQ

If you have any questions in regards to this review or the results please contact: Maria.Kokocinska@bch.nhs.uk