Autosomal Recessive Polycystic Kidney Disease (ARPKD) RaDaR Registry and Rare Diseases Group

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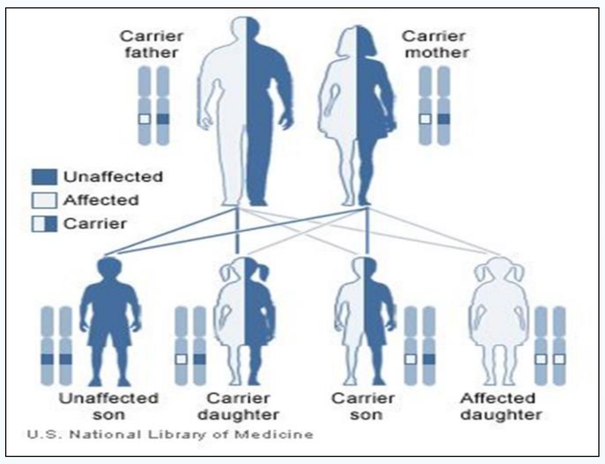
Background

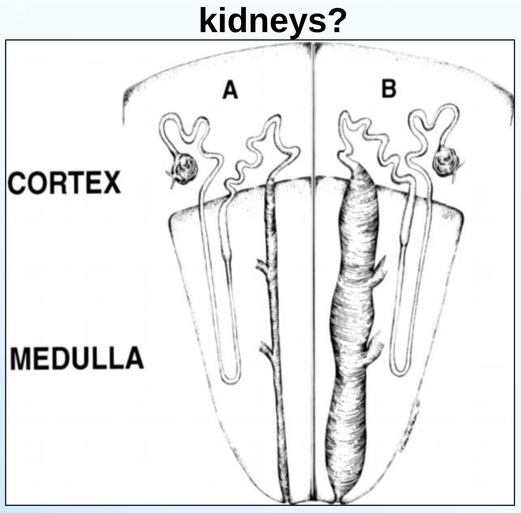
Autosomal recessive polycystic kidney disease (ARPKD) is a rare genetic condition that causes cysts to develop in the liver and kidneys leading to progressive liver fibrosis, portal hypertension and renal failure. At the severe end of the spectrum, patients may require combined liver kidney transplant. 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 approach.

How is ARPKD inherited?



The ARPKD RaDaR Rare Disease Renal Group was formed in 2012 and brought together patients, PKD Charity and international experts clinicians and scientists.

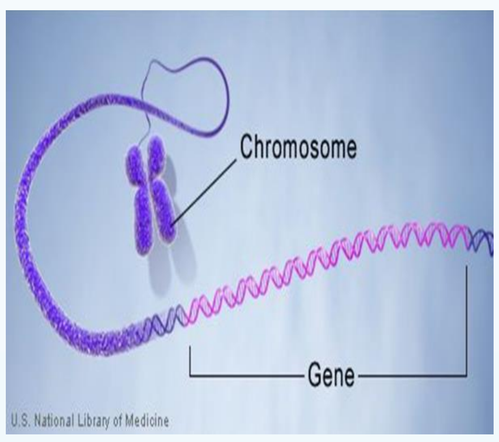
How does it go wrong in the kidneys?

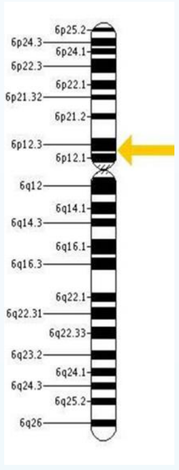


Lonergan G J et al. Radiographics 2000;20:837-855

Collecting duct dilation in ARPKD

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





Initiatives

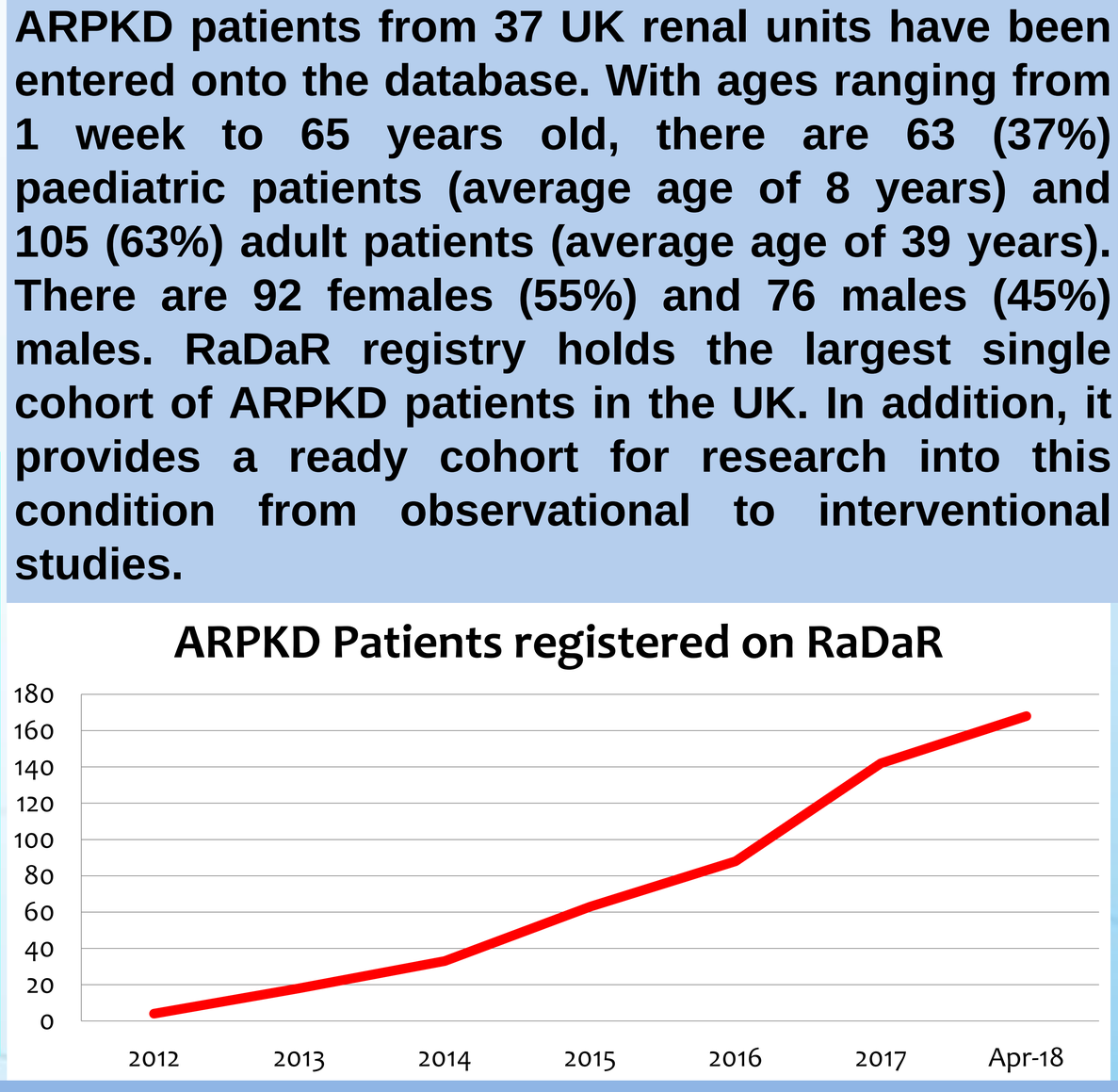
Family Support Days with experts speaking on various aspects of the condition to support families. Since 2012, we have had six Family Days held in Birmingham, London, Leeds and Kingston with different topics all based on feedback and requests from families. During these events childcare is provided to allow parents to concentrate on the information being given. We have had fantastic feedback and have families travelling from Norway and Hong Kong to attend.

Registry

The registry called RaDaR collects data, following consent, including demographics, blood and urine results, medications, transplant and dialysis history, genetics and co-morbidities from the patient's medical records and entered onto a database where it is made available to researchers working on this condition. To date, data from 168 ARPKD patients from 37 UK renal units have been entered onto the database. With ages ranging from 1 week to 65 years old, there are 63 (37%) paediatric patients (average age of 8 years) and 105 (63%) adult patients (average age of 39 years). There are 92 females (55%) and 76 males (45%) males. RaDaR registry holds the largest single cohort of ARPKD patients in the UK. In addition, it provides a ready cohort for research into this condition from observational to interventional studies.

Objectives

- To provide information and support for patients and families affected by ARPKD
- To collect detailed epidemiology data in the RaDaR registry
- To develop future research project and best practice guidelines.



Collaborations

We have links with international registries from North America (Prof Guay-Woodford, University of Alabama Hepatorenal Fibrocystic Registry) and European (Prof Carsten Bergman and Dr Max Liebau, Germany).

Conclusions

International experts took part in a consensus conference in Washington in 2013 and this lead to the publication of the first international guidelines for the management of this complex condition:
http://rarerenal.org/wp-content/uploads/2013/06/ARPKD-Int-Consensus-2014.pdf

This has highlighted areas that require more evidence in the form of research. Currently patients with ARPKD are also being offered a chance of taking part in the UK's 100 000 Genome Project and having their full genome sequenced. It is hoped that the data from the project will increase our understanding of this highly variable condition and will eventually lead to the development of new medications and treatments.

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@nhs.net