



The PKD Patient Pathway within the NHS: from referral to treatment

Dr Richard Sandford

Wellcome Trust Senior Fellow in Clinical Research &
Consultant in Medical Genetics,

Addenbrooke's Hospital, Cambridge

PKD Information Weekend

Birmingham 2007

‘Individuals and families living with PKD require ‘flexible’ access to many different health services’

Summary of talk

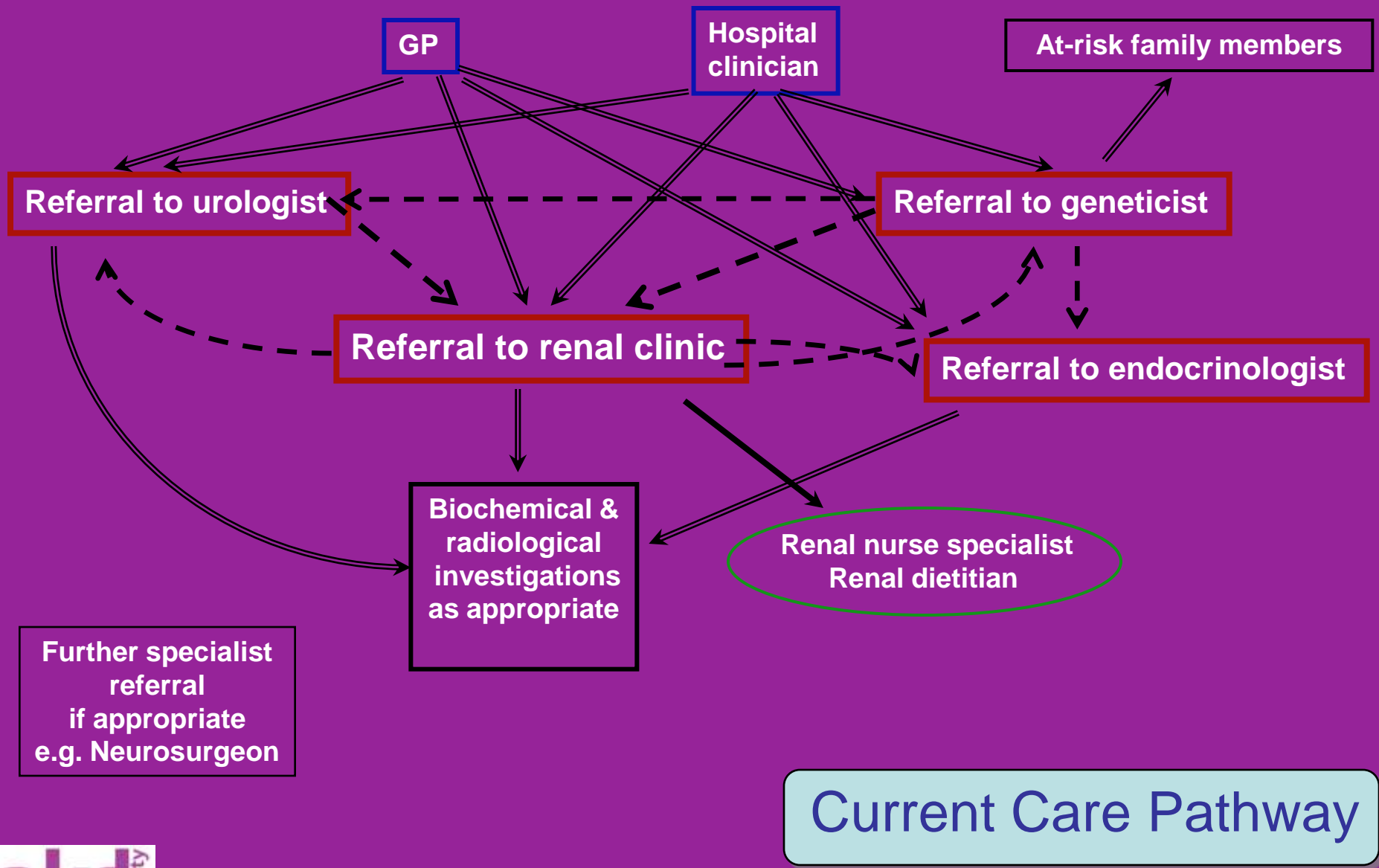
- Discussion points!
- How are individuals and families with PKD currently managed within the NHS
- What improvements can we make?
- The Cambridge RGTD Clinic
- What are the challenges?



‘At Addenbrooke’s Hospital ~ 12% of all patients attending nephrology outpatient clinics have a primary renal genetic disorder’

'No national guidelines or data on the management or use of genetic services or screening for inherited renal disease'

- White paper “Our inheritance, our future – realising the potential of genetics in the NHS”(2000)
- Renal NSF “to provide our patients with tailored information that will empower them to make informed decisions about their care”
- Single gene disorders account for over 6% of adult patients requiring renal replacement therapy, but currently there is no specialist provision for this group of patients or their families



Current Care Pathway





Department of Health: 'Bringing Genetics into Mainstream Medicine'

**2 year pilot for multidisciplinary clinic at Addenbrooke's
External evaluation (Nottingham University and DoH)
Specialist commissioner approval required**

**Clinic
Care pathway
Genetic Tests
Educational material - patients and GPs**



- **Clinic opened on 12th January 2005**
- **To date >250 patients seen**
- **Patients are allocated to RGTD1 or RGTD2**
 - **RGTD1: PKD**
Familial hypertension
Gitelman / Bartter syndrome
Other tubular disorders
'Funny' potassium
 - **RGTD2: inherited stone forming disorders (eg cystinuria)**
recurrent nephrolithiasis
nephrocalcinosis

Anthony Norden

Clinical Biochem

Dick Sandford

Medical Genetics

Nimish Shah

Urology



Sarah Waller

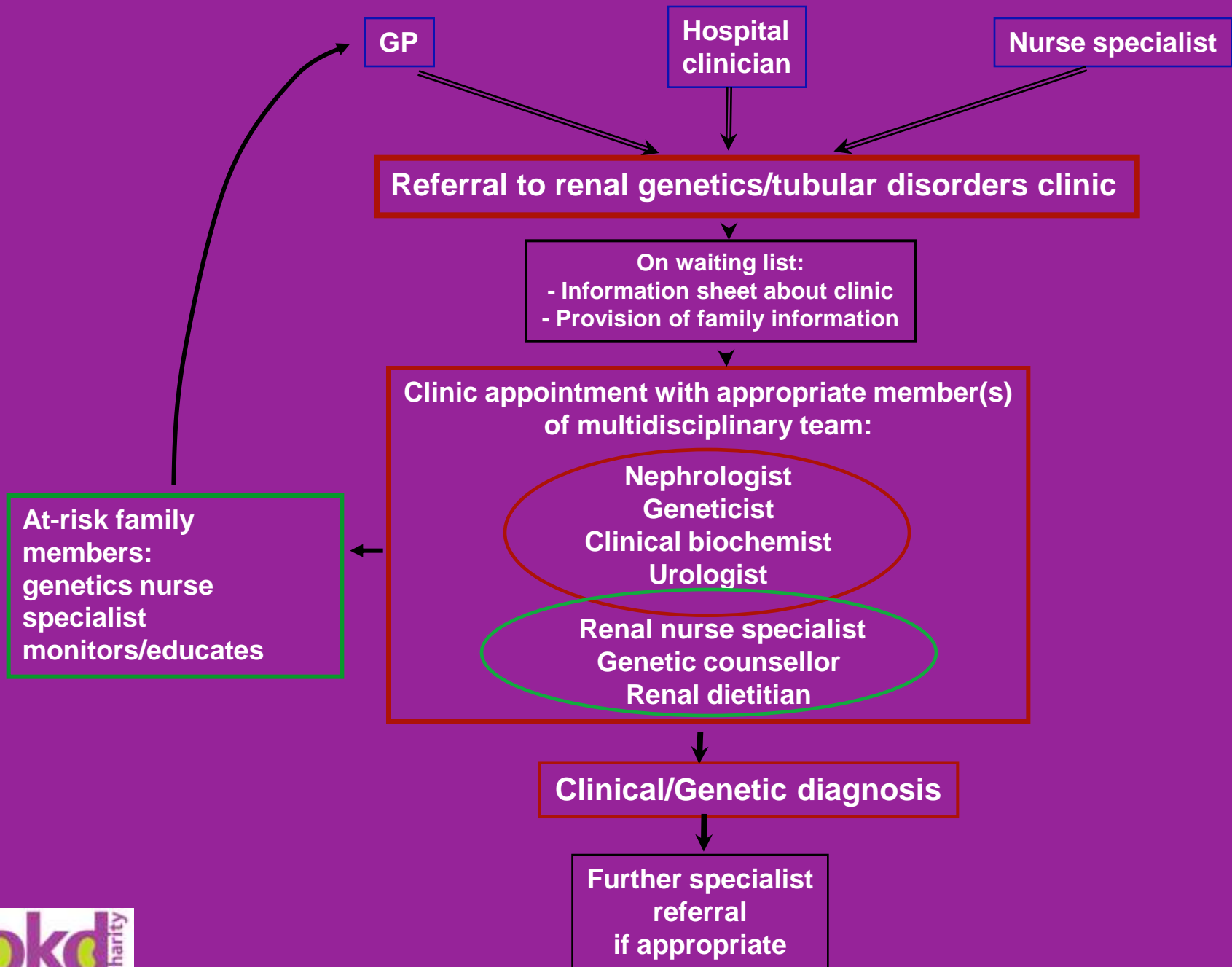
Molecular Genetics

Fiona Karet

Nephrology

Deborah Spencer

Renal Nurse Specialist





Communications

Web-based clinic access to patient data

Web pages for patients and GPs
(www.addenbrookes.nhs.uk/serv/clin/med/renalgenetics_clinic.html)

Patient information leaflets

The screenshot displays the 'RENAL GENETICS CLINIC' web interface. At the top, there is a navigation menu with links for CLINICS, GENETICS, PATHOLOGY, REPORTS, CONTACTS, and INFORMATION. Below the menu is a search section with input fields for 'firstname', 'lastname', 'dd', 'mm', 'yyyy', 'hosp_ID', and 'gen_ID'. A large empty text box is provided for additional search criteria. Below the search section are four data entry fields, each with a label and a small icon:

- GENETICS: A text input field with a '0' in a small box to the right.
- RENAL: A text input field with the label 'CRN : 0'.
- CLINICS: A text input field with the label 'CRN : 0'.
- LETTERS: A text input field with the label 'EDMS'.



Cambridge University Hospitals **NHS**
NIH Foundation Trust

Cambridge | Home

About us Patients Visitors Press Services Research Teaching Careers A to Z

Medical Services

Renal Genetics and Tubular Disorders Clinic

Tel: 118
 Tel: 01223 256308

About the clinic

This Clinic brings together staff from Addenbrooke's Hospital: Nephrology, Medical Genetics, Urology and Clinical Biochemistry to see patients whose disease usually needs to be looked after by one or more of these specialities.

The Clinic was set up in February 2003 with initial funding for two years from the Department of Health as part of its Mainstreaming Genetics programme.

Patients may be referred by their general practitioners (GPs) or by hospital doctors from Addenbrooke's, from local hospitals or from further away if there is no local centre.

What does the clinic offer?

- The clinic has a specialist nephrologist, medical geneticist, urologist and chemical pathologist with a specialist renal nurse.
- We have the expertise in the clinic at a single visit to evaluate fully and care for the patient, which should lead to better and more efficient care.
- We have access to genetic counselling and can arrange diagnosis, mutation screening for selected conditions where the causative gene is known.

Which patients are referred to us?

We can provide services to:

- Adults who have all types of renal tubular diseases including polycystic kidney disease: the cause may be acquired, hereditary or unknown.
- Patients who have recurrent kidney stones under a family history of kidney stones.
- Patients who are likely to get kidney problems in future.

Typical diagnoses or problems include:-

- Autosomal dominant polycystic kidney disease
- Bilateral hydronephrosis
- Cystinosis
- CHST3 disease
- Family hypercalcaemia
- Sjögren syndrome
- Hypercalcaemia (if unknown cause)
- Hyperphosphataemia (if unknown cause)
- Renal tubular acidosis disease
- Nephrocalcinosis
- Renal tubular acidosis or hyperkalaemia (if unknown cause)
- Recurrent kidney stones
- Nephrocalcinosis
- Renal tubular acidosis (distal or proximal)
- Patients with renal impairment

Key contacts

Clinic PA Tel: 01223 256 318; Fax: 01223 286 506

Consultants:

- Prof Frank Kurland (Nephrology) Tel: 01223 262 917
- Dr Andrew Nordin (Clinical Biochemistry) Tel: 01223 286 820
- Dr Robert Sandford (Clinical Genetics) Tel: 01223 262 915
- Dr Martin Shah (Urology) Tel: 01223 267 123

Renal Nurse Specialist: Sr Deborah Spencer Tel: 01223 257 184

For referring doctors

To make referrals, please contact the Clinic PA, ROTO Clinic, Box 118, Addenbrooke's Hospital, Hills Road, Cambridge CB2 2QQ; Tel: 01223 256 318; Fax: 01223 286 506.

If you are a referring doctor, please feel free to discuss patients with the clinic staff listed above.

Patient involvement

We are developing patient information about some disorders of the kidney and are including patients' views and experiences. Contact the clinic for further information.

Useful links

We have compiled a [list of links to other websites](#) which we hope you will find helpful.

Addenbrooke's Online: <http://www.addenbrookes.nhs.uk> - only accessible within NHS systems.

Addenbrooke's Online is our internet website on NHSnet. It was launched in September 2003 and includes comprehensive information about Addenbrooke's for all NHS staff including referring general practitioners (GPs).

It replaces Intranet (our 'GP website'). For further information contact the [GP Liaison Office](#) or [PB & Communications](#).

For further information contact: The Renal Services Manager, or Clinical Director, The Dialysis Centre, Box 118, Addenbrooke's Hospital, Hills Road, Cambridge CB2 2QQ; Tel: 01223 217 828 (general enquiries); Fax: 01223 286 506.

Last updated: 17 October 2009

Addenbrooke's Hospital, Cambridge University Hospital NHS Foundation Trust,
 Hills Rd, Cambridge CB2 2QQ
 Switchboard Tel: 01223 247 151; webmaster@addenbrookes.nhs.uk | www.addenbrookes.nhs.uk

NHS Direct



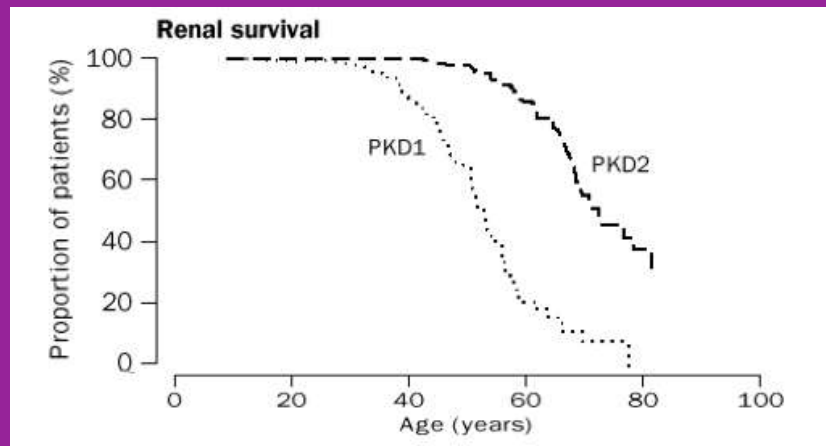


‘Gene dossiers’: almost no genetic tests for inherited kidney diseases currently available in the UK

- **Advantages of genetic tests:**
 - diagnostic/predictive testing
 - optimizing therapy
 - exclusion from unnecessary follow up
 - consideration for living related transplantation
 - patient knowledge
 - Prenatal diagnosis

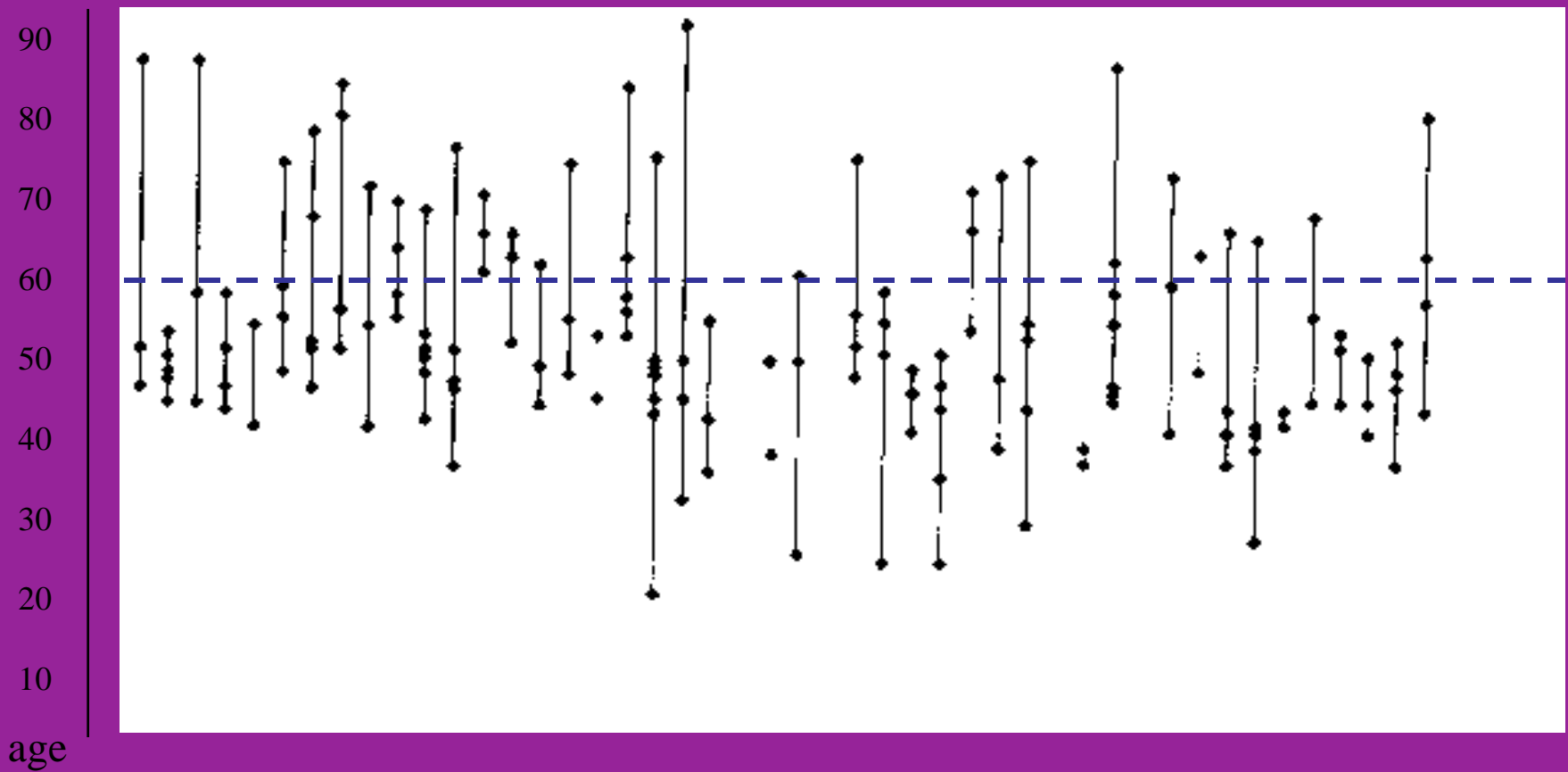
Genetic testing: *PKD2*

- ADPKD is caused by mutations in *PKD1* and *PKD2*
- 85% have a mutation in *PKD1*
- A *PKD2* mutation predicts 'milder disease'.



PKD2 mutation testing in Cambridge

- RGTD1 clinic
- All ADPKD patients with Cr < 400
- Patients > 60
 - 25/50 (50%) have *PKD2*
- Implications for
 - Management
 - Follow-up
 - Family screening



Variation in age at renal failure in ADPKD between families

(Ritz, 1996)

Ultrasound screening in ADPKD: Effect of genotype

sensitivity	PKD1	PKD2
<30	95%	67%
>30	100%	100%

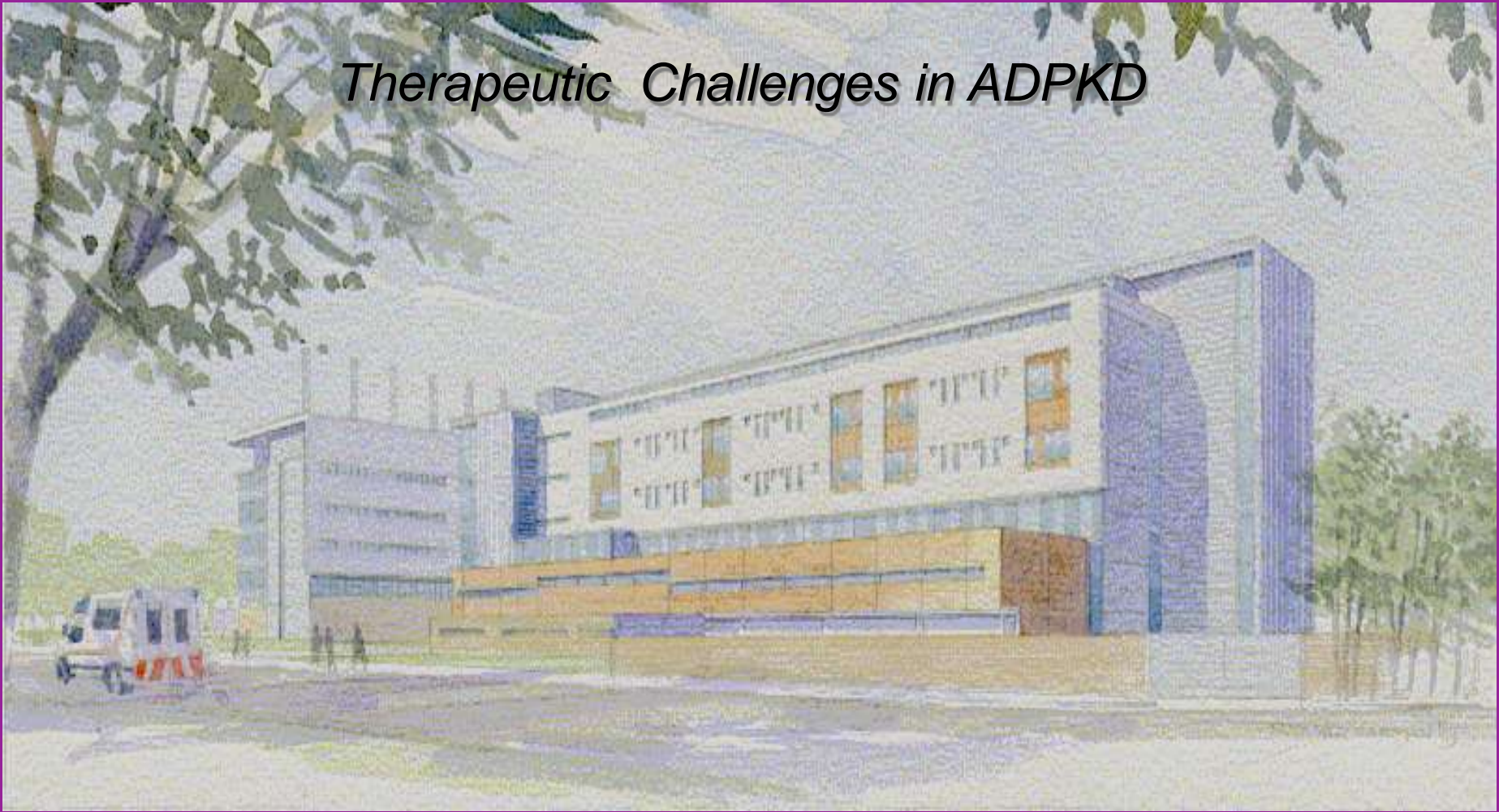
Management of ADPKD:

- Specialist referral when appropriate (CKD guidelines)
- CKD management
- Genetic counselling
- Genetic testing
- Monitoring disease progression
- Disease specific treatment
- Family screening
- Flexible follow-up

Current Challenges

- How best to provide services for individuals and families living with PKD
- Access to and provision of screening services (in-clinic)
- Role of genetic testing
- Equality of Management
- Current best treatments for complications and CVS risk
- Clinical trials

Therapeutic Challenges in ADPKD



The next 5 years.....

Acknowledgements

**Prof. Fiona Karet
Dr. Jo Whittaker**

**Mr. Simon Griffith
Ms. Karan Hotchkin
Ms. Miranda Fyfe**

**Dr. John Bradley
Mr. Nigel Bullock
Prof. Martin Bobrow**

Department of Health