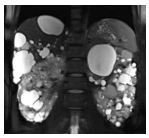
Diagnosing PKD

Neil Turner

January 2015

PKD – 8-10% of ESRF



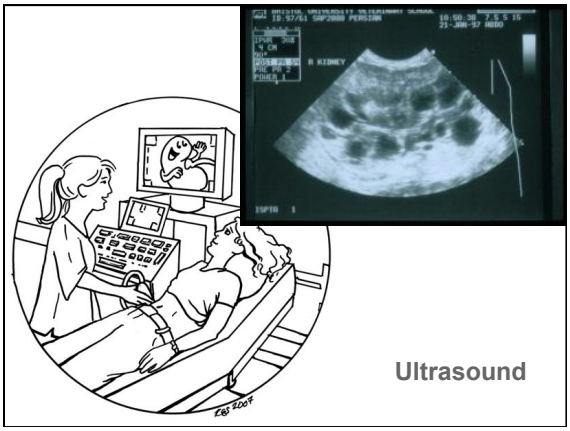
In South East Scotland:

290 patients in Lothian and Borders
10-20 have had genetic testing?

200 CKD

90 RRT (12.5%): 35 dial + 55 transplant

Diagnosis



Ultrasound

Criteria for diagnosing PKD

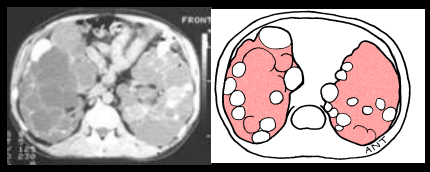
If the disease is in your family:

Diagnosis		Exclusion	
Age	**Number of cysts**	**Age**	**Number of cysts**
>60	4+	>40	Maximum 1
<60	3+	<40	Cannot exclude

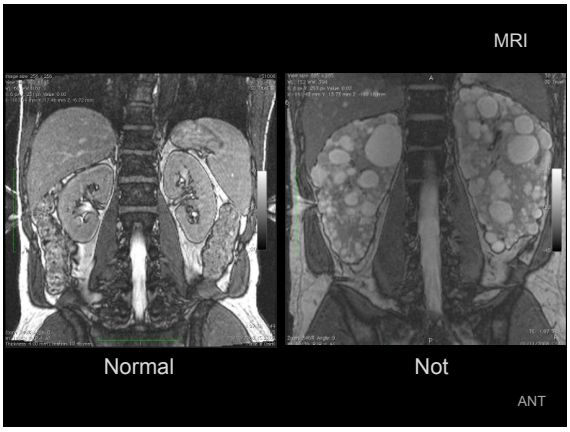
MRI is more sensitive, but some false positives

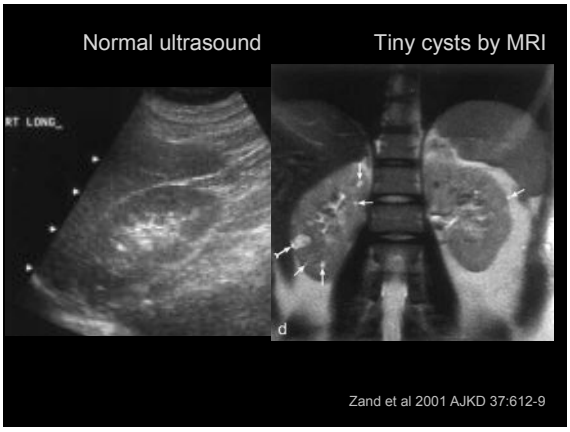
Ravine criteria

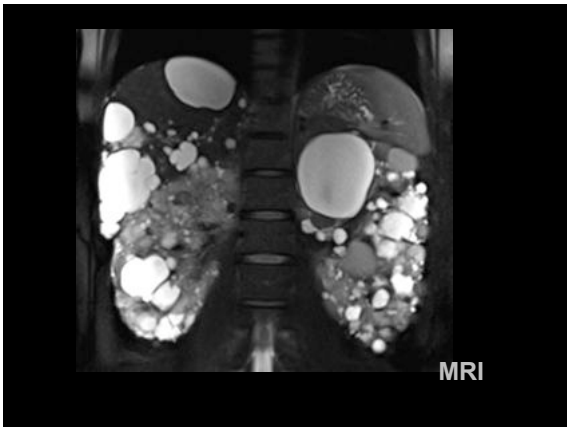
Diagnosis



CT







PKD Genetics

1985 – PKD1 tracked down to Chr 16

1994 – PKD1 gene sequenced

1999 – PKD2 sequenced

2002 – complete mutation screen of PKD1

2011 – PKD1+2 sequence analysis
in UK Genetic Testing Network

There are some other rare causes

Steve Reeders

PKD Genetics

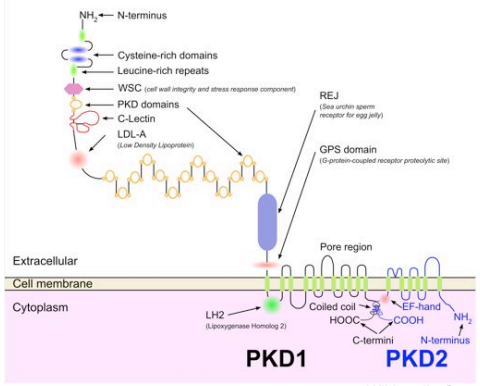
1985 – PKD1 tracked down to Chr 16

1994 – PKD1 gene sequenced

1999 – PKD2 sequenced

2002 – complete mutation screen of PKD1

2011 – PKD1+2 sequence analysis
in UK Genetic Testing Network



Wikimedia Commons

What genetics has taught so far

- PKD1 mutations are worse
- Not everyone who carries the gene gets into trouble
- Sometimes, another gene makes it worse

Problems with genetic testing

- Large, complex gene
- Unique mutations
- Imperfect sensitivity
- Expensive!

The 'new genetics'

So who should have a genetic test?

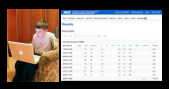
Most people don't need a test to make the diagnosis. It adds extra if:

- The diagnosis is probable but not certain
- Your related donor might have PKD

In the future, everyone will want one

- For early detection
- For other genes

More info



patientview.org

edren.org/info

neil.turner@ed.ac.uk