

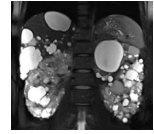
## Diagnosing PKD

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October 2014



## PKD – 8-10% of ESRF



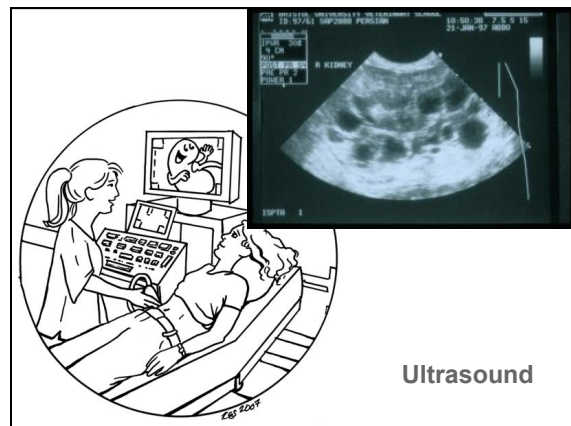
In South East Scotland:

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

**200** CKD

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

## Diagnosis



## Criteria for diagnosing PKD

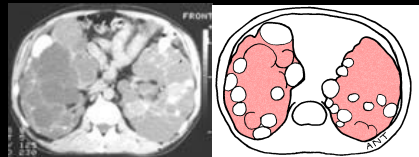
If the disease is in your family:

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

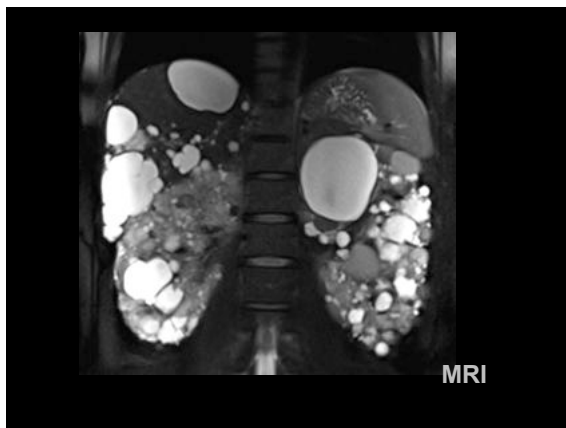
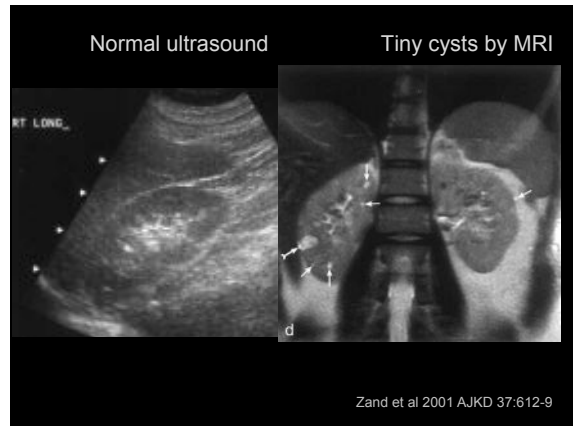
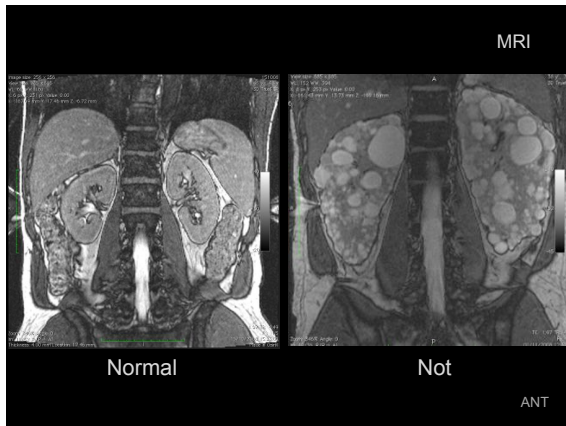
MRI is more sensitive, but some false positives

**Ravine criteria**

## Diagnosis

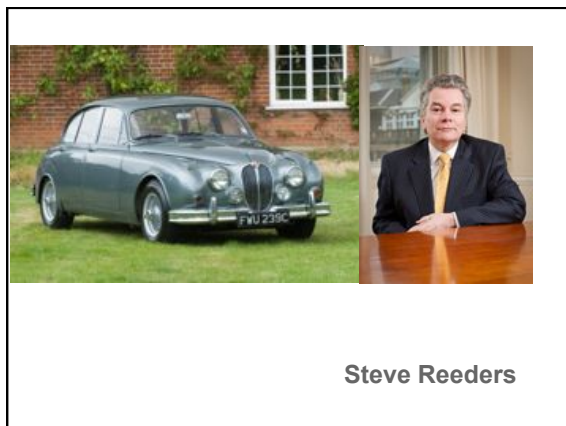


CT



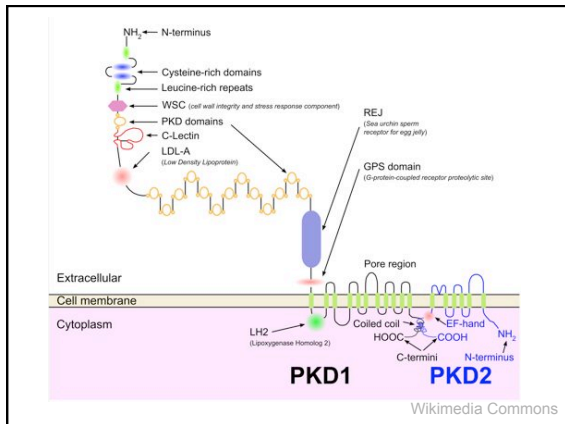
**PKD Genetics**

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*



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## 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](http://patientview.org)

[edren.org/info](http://edren.org/info)

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