

HNF1B research and you

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Where?

- The NIHR Exeter Clinical Research Facility oversees a wide range of research projects
- Internationally recognised for its research into diabetes and genetic misprints linked to the condition



Why?

- Volunteers are essential to our research
- You can help us to better understand the condition and contribute to improvements in future diagnosis and treatment



Research success so far



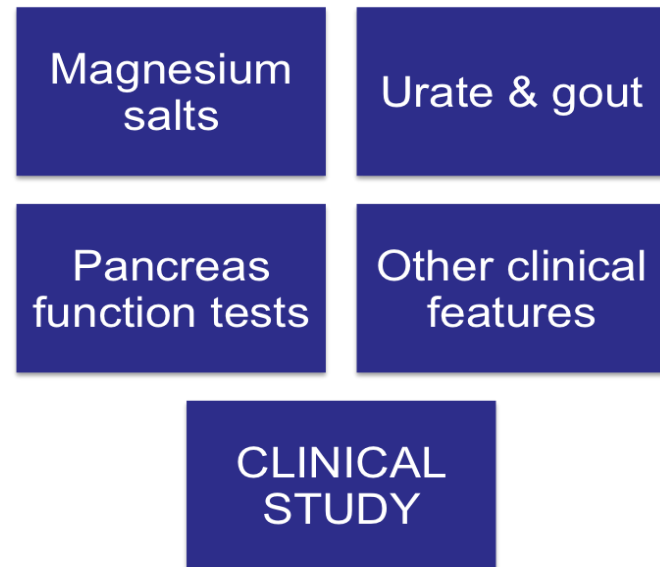
- The major causes of kidney failure in children and young adults are problems in the development of the kidney
- Work so far has shown that the commonest genetic cause of this is due to a change in the *HNF1B* gene
- Other organ development is also affected

New projects 1

- **Problem:** people with a change in the *HNF1B* gene can have a variety of different clinical features
- This can make it difficult for healthcare professionals to recognise *HNF1B* disease



- **Plan:** to design a “calculator” that works out the likelihood of someone having a change in the *HNF1B* gene
- **Outcome:** this should lead to increased recognition and diagnosis of *HNF1B* disease

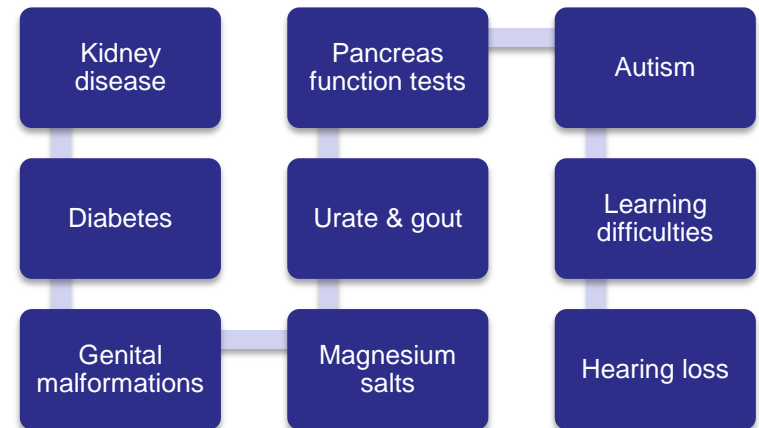


New projects 2

- **Problem:** it is not clear why people with different changes in the *HNF1B* gene have different clinical features



- **Plan:** to compare a large group of individuals with different changes in the *HNF1B* gene
- **Outcome:** this should lead to a better understanding of the role of *HNF1B*



What will it involve for you?



Use of existing medical information
Interview
Height, weight & blood pressure measurements
Simple screening tests for learning disability & autism
Blood test
Urine & stool samples
(MRI scan of abdomen & hearing test in some cases)



Other research

PROBLEM

- Testing of *HNF1B* gene confined to “coding” regions of DNA

PLAN

- Use new lab techniques to test the “non-coding” regions

OUTCOME

- Improvements in genetic testing



Other research

PROBLEM

- Many people have features of RCAD but no cause is found

PLAN

- Use new sequencing techniques to look for changes in other genes

OUTCOME

- Design of new genetic tests for developmental kidney disease



Is there any risk to volunteering?

- All clinical research studies in the UK must be approved by a formal committee
 - This ensures the research is ethically and scientifically sound
- Potential volunteers will be provided with information about the study, including any risks
- You can withdraw at any point





What next?

- Completion of declaration of interest leaflet
- By signing up you are agreeing for us to stay in contact with you and invite you to take part in studies
 - This is purely voluntary and you may withdraw at any time



Any questions?

