HNF1B research update

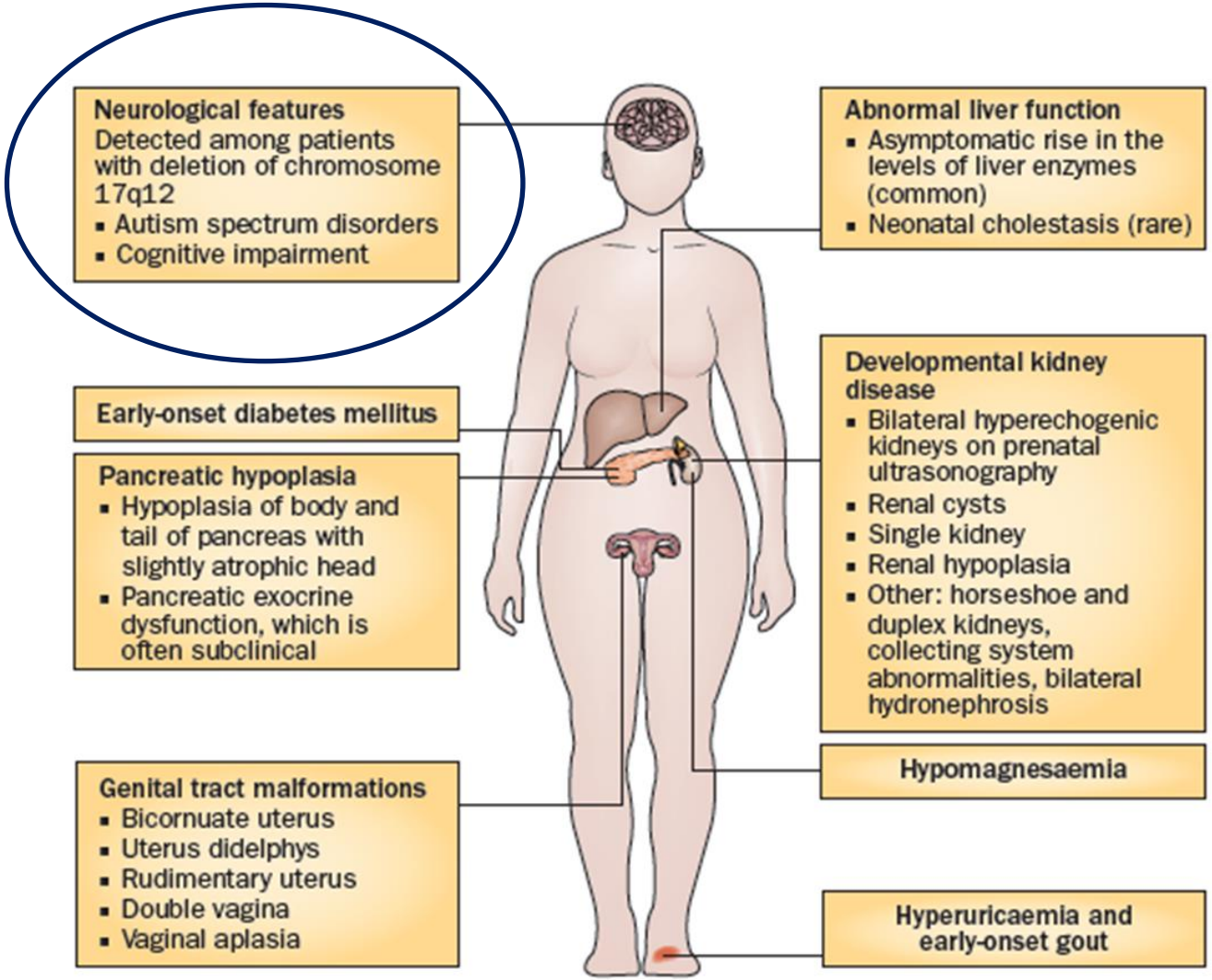
Dr Rhian Clissold

HNF1B support day, Feb 2016

UNIVERSITY OF EXETER | MEDICAL SCHOOL

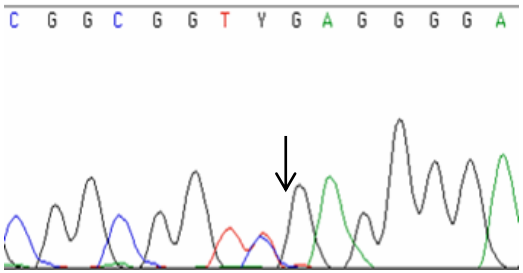
MRC Medical Research Council

HNF1B-associated disease is a multi-system disorder

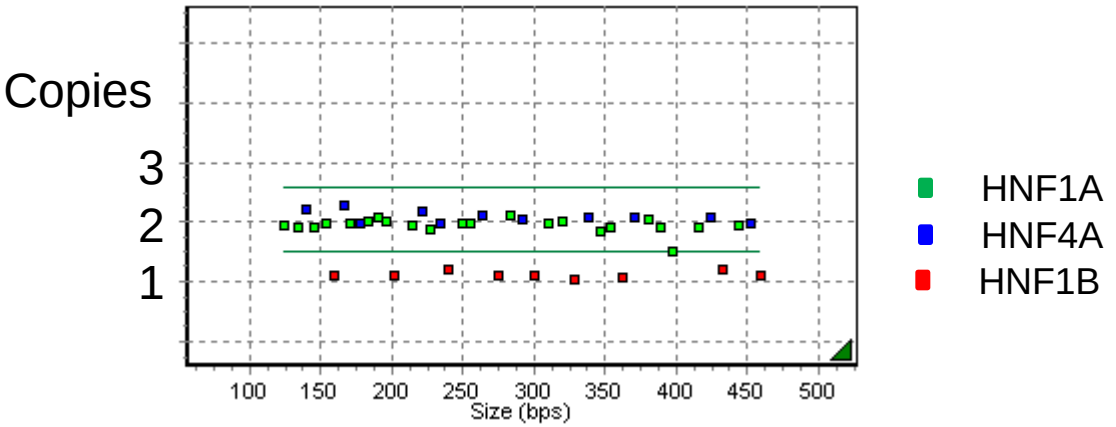


Genetics

- Genetic “spelling mistake” = mutation



- Gene is missing = deletion



HNF1B and the brain

- Approx. 15,000 people referred to clinical genetics with autism, delayed development or learning difficulties

- Large deletion including *HNF1B* found in 18 individuals

What about people in the renal clinic?

- 39 children with HNF1B renal disease
  - 26 with deletions, 13 with mutations

- No significant differences between deletion and mutation groups with regards learning abilities and schooling

Aim of our study

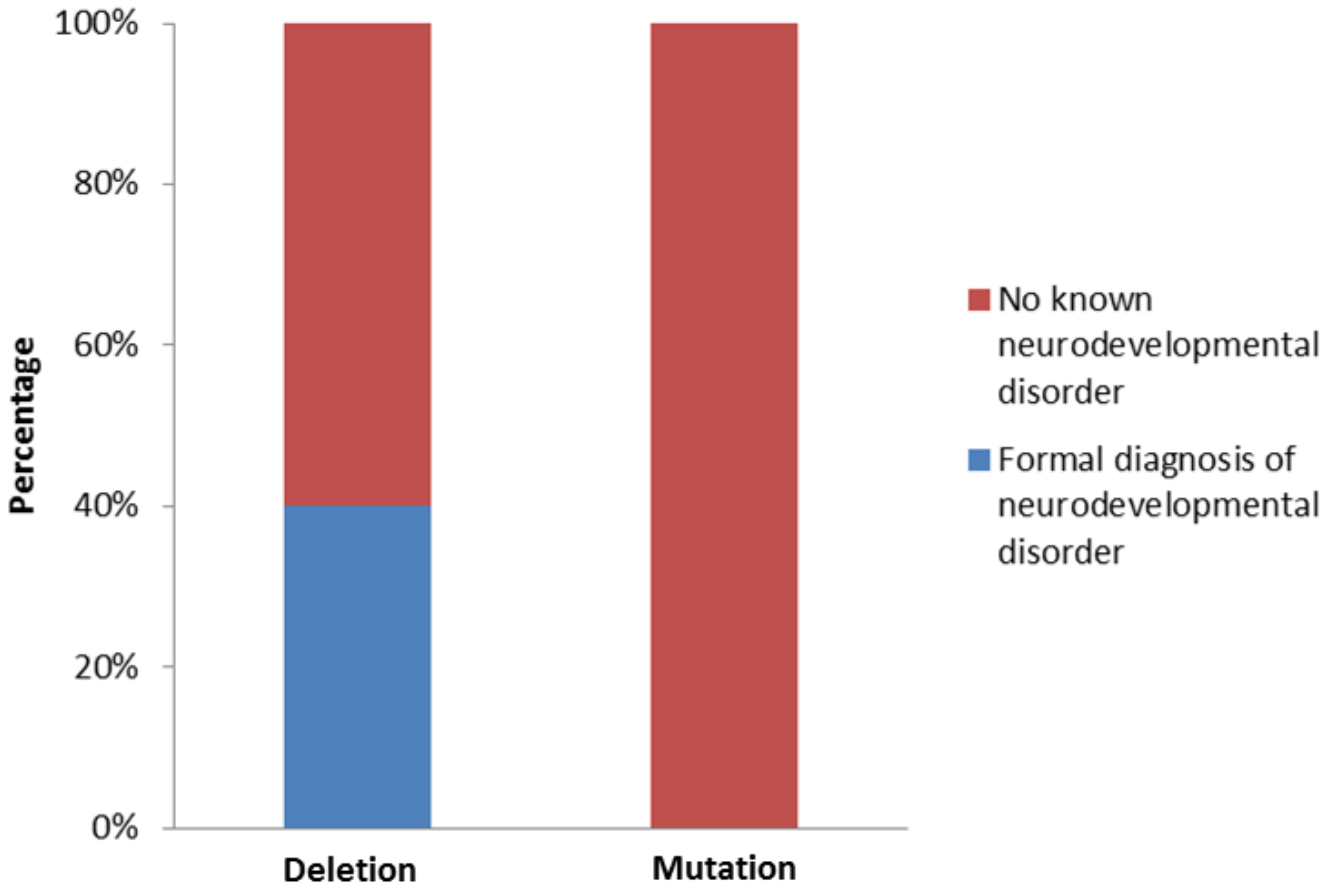
- To assess neuropsychological disorders in people with *HNF1B* mutations or deletions under follow-up with renal or diabetes services

What did we do?

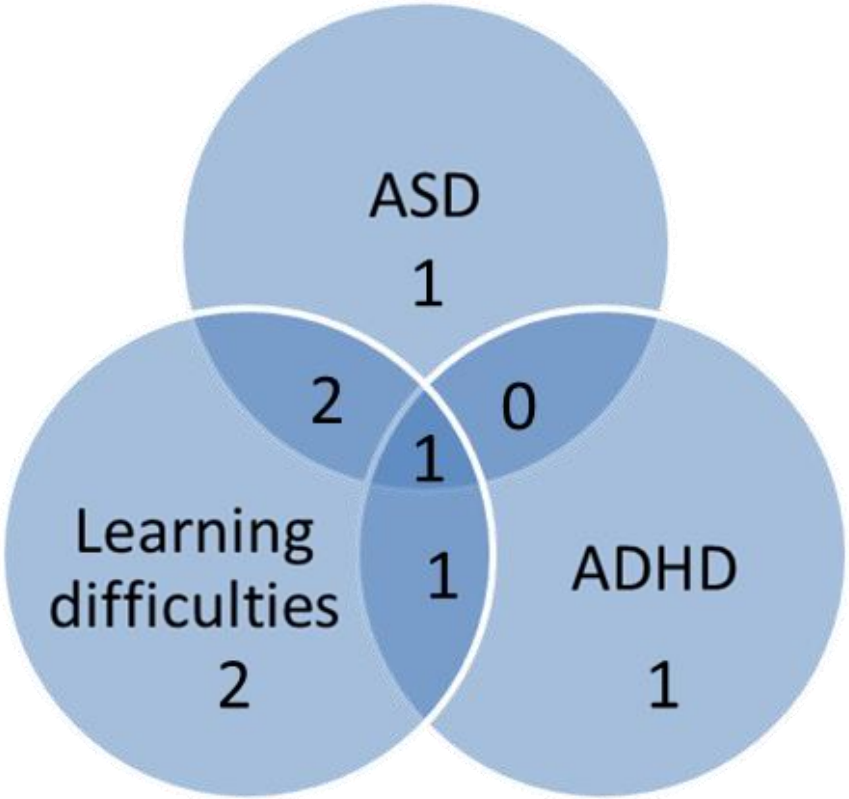
- 38 children and adults from 4 UK sites visited
  - 20 with deletion
  - 18 with mutation

Assessments:
Brief behavioural screening (SDQ)
Clinical diagnosis (review of medical notes)
Autistic traits (AQ)
IQ (KBIT2)

8/20 individuals with a deletion had a clinical diagnosis



Overlap of diagnoses



What did we conclude?

- Neurodevelopmental features only seen in people with a deletion
- *HNF1B* gene unlikely to be the cause
- Important for renal/diabetes teams to be aware of this increased risk

Limitations of work

- Not everyone who was eligible took part
  - Prevalence and spectrum of these conditions in HNF1B renal disease remains unknown
- Full diagnostic tests for autism and ADHD not used
  - Less severe disease may have been missed
- Genetic screening for other causes not done

Questions?

- A big thank you to everyone that took part in my study

- www.rarerenal.org for future opportunities for research and development