## RaDaR Rare Disease Group Annual Report

### April 2015 – March 2016

<table>
<thead>
<tr>
<th>Rare Disease Group</th>
<th>HNF1b</th>
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<tr>
<td>Lead Clinician</td>
<td>Coralie Bingham</td>
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### Summary of RDG meetings
E-mail contact between members of the RDG

### Summary of patient events
A very successful patient information day was held in Manchester on 29th February 2016.

### Grant applications submitted
N/A

### Grants awarded
- MRC funded research fellow continues with her project in Exeter
- A PhD studentship in Manchester studying human kidney cells from induced pluripotent precursor cells with HNF1B mutations.

### Publications and presentations
- Dr Clissold: Poster at EDTA 2015, HNF1B whole gene deletions are associated with autistic traits.
- Dr Bingham: HNF1B Monogenic Diabetes Genetics Symposium, Exeter, February 2016
- Dr Sayer and Dr Montgomery: Poster at EDTA and ASN 2015. Phenotypic analysis of a cohort of patients with HNF1B mutations.
- Assessment of the HNF1B score as a tool to select patients for HNF1B genetic testing. Nephron 2015; 130:134-140
- HNF1B genetic testing in a Turkish Cypriot population with a high incidence of familial kidney disease. Journal of Nephrology and Therapeutics, 2016, 6:1

### Highlights
Patient day in Manchester
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<th>Problems</th>
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<td>Need to continue to encourage adult and paediatric units to register all HNF1B patients on RaDaR.</td>
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<td>Patients only under the care of diabetologists can not be recruited to RaDaR.</td>
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