

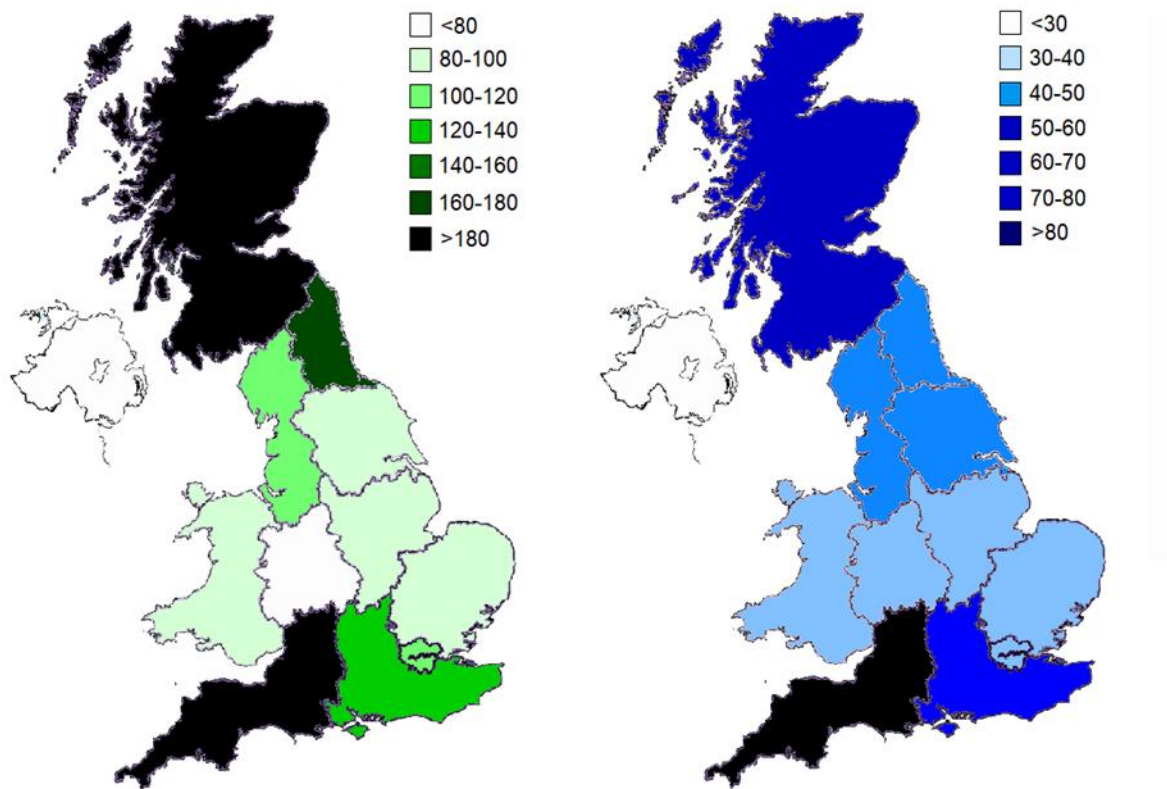
Supplementary material to accompany

Improvements in awareness and testing has led to a three-fold increase over 10 years in the identification of monogenic diabetes in the UK

Authors: Lewis Pang MSc, Kevin C Colclough DClSci, Maggie H Shepherd PhD, Joanne McLean BSc, Ewan R Pearson PhD, Sian Ellard PhD, Andrew T Hattersley DM, Beverley M Shields PhD

Supplementary figure 1:

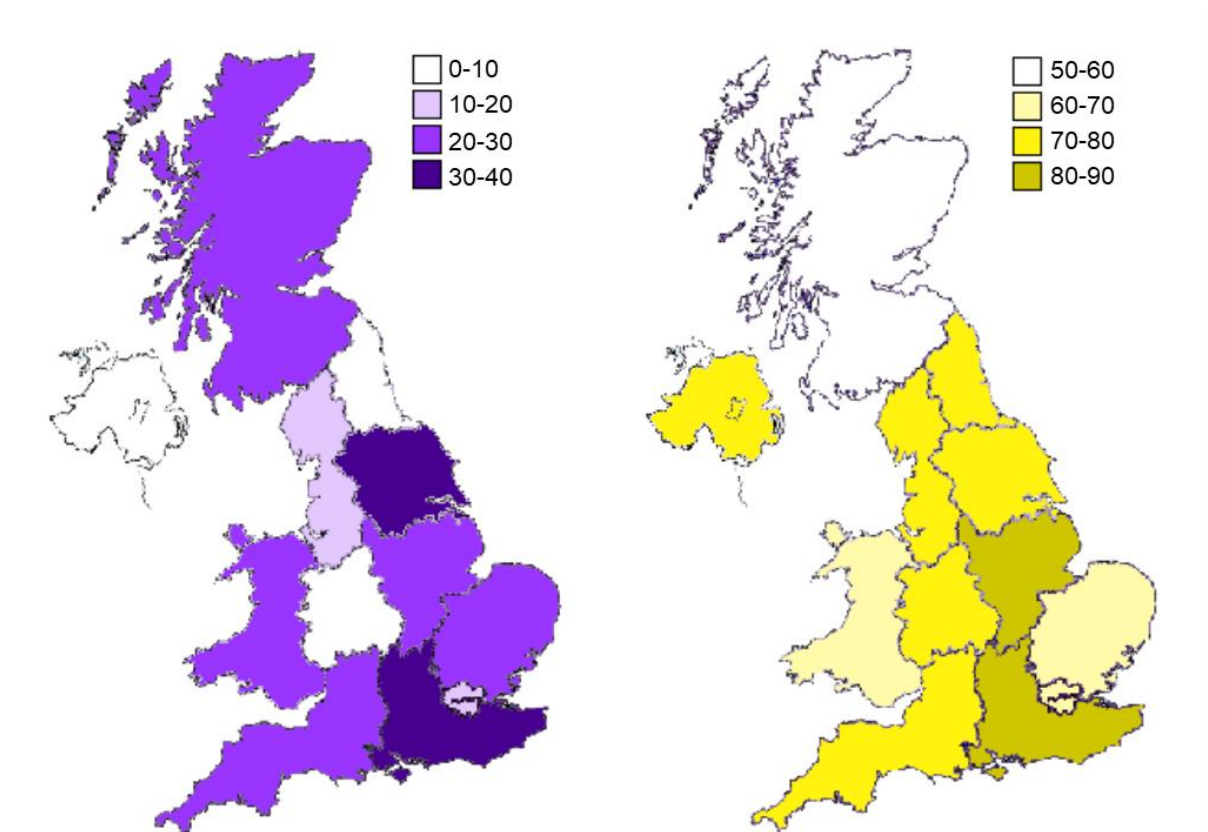
Map showing variability in a) referral rates and b) confirmed MODY cases across regions of the UK from 1st January 1996 to 31st December 2019. Figures shown are referrals/cases per million population.



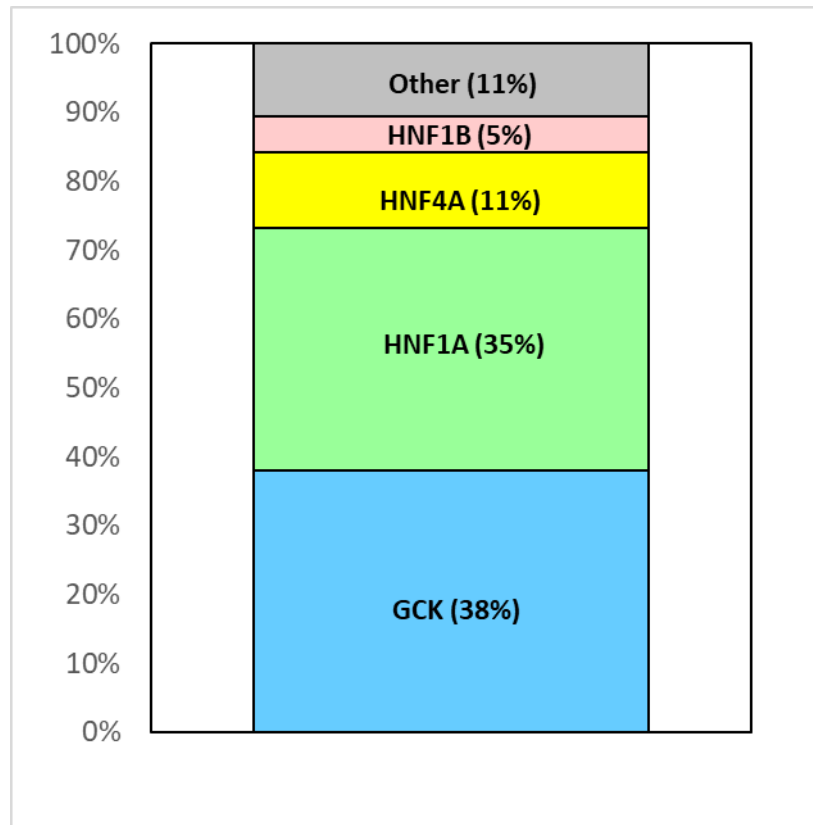
Supplementary Table 1 – Involvement of Genetic Diabetes Nurse (GDN) or the MODY calculator in referrals, split by region for the Exeter Laboratory (data not collected in the Scotland Laboratory; testing Scotland from 2016). Data presented as percentage of referrals where GDN/MODY calculator involvement reported.
 *MODY calculator involvement only assessed in referrals using the Exeter diagnostic request form since 2014 (when MODY calculator usage first recorded) and those eligible for the MODY calculator (White European, diagnosed aged ≤35).

Region Code	GDN person-time (months)	GDN involved in referral n (%)	MODY calculator involved in referral n (%)*
Scotland	227	196/813 (24%)	40/69 (58%)
Wales	95	69/255 (27%)	41/59 (69%)
Northern Ireland	0	1/127 (8%)	23/30 (77%)
England	2237	1555/6786 (23%)	1120/1499 (75%)
English regions			
East	333	147/571 (26%)	101/151 (67%)
South East	346	361/1163 (31%)	250/309 (81%)
South West	635	394/1402 (28%)	201/269 (75%)
London	96	130/1019 (13%)	120/191 (63%)
West Midlands	144	47/461 (10%)	64/80 (80%)
East Midlands	160	94/393 (24%)	90/103 (87%)
Yorkshire/Humber	195	206/543 (38%)	101/131 (77%)
North East	93	43/448 (10%)	59/83 (71%)
North West	235	133/781 (17%)	127/171 (74%)
Unknown	N/A	0/5 (0%)	7/11 (64%)

Supplementary Figure 2: a) Map showing variability in percentage of referrals associated with genetic diabetes nurses (GDNs) (in purple); b) Map showing variability in percentage of referrals reporting usage of the MODY calculator (in yellow). Note that the Scotland laboratory (testing Scottish referrals since 2016) does not record GDN or MODY calculator involvement. MODY calculator involvement only assessed in referrals using the Exeter diagnostic request form since 2014 (when MODY calculator usage first recorded) and those eligible for the MODY calculator (White European, diagnosed with diabetes aged 1-35).



Supplementary Figure 3: Stacked bar chart to show distribution of genetic causes for the 3860 confirmed MODY cases. Full breakdown of genetic causes (including those in “Other”) is in Supplementary Table 1:



Supplementary Table 2: Full breakdown of the genetic causes for the 3860 cases diagnosed with monogenic diabetes in the UK

Gene	Number of cases	Proportion of Total
<i>GCK</i>	1465	37.95%
<i>HNF1A</i>	1361	35.26%
<i>HNF4A</i>	423	10.96%
<i>HNF1B</i>	201	5.21%
<i>m.3243A>G</i>	172	4.46%
<i>ABCC8</i>	70	1.81%
<i>RFX6</i>	29	0.75%
<i>INS</i>	21	0.54%
<i>LMNA</i>	20	0.52%
<i>INSR</i>	13	0.34%
<i>WFS1</i>	13	0.34%
<i>KCNJ11</i>	12	0.31%
<i>PPARG</i>	11	0.28%
<i>NEUROD1</i>	10	0.26%
<i>SLC19A2</i>	8	0.21%
<i>POLD1</i>	7	0.18%
<i>PDX1</i>	5	0.13%
<i>GATA6</i>	3	0.08%
<i>HNF1A</i> & <i>HNF4A</i>	3	0.08%
<i>SLC29A3</i>	3	0.08%
<i>TRMT10A</i>	3	0.08%
<i>MAFA</i>	2	0.05%
<i>PAX6</i>	2	0.05%
<i>6Q24</i>	1	0.03%
<i>HNF1A</i> & <i>GCK</i>	1	0.03%
<i>ZBTB20</i>	1	0.03%
Total	3860	