

Supplementary Materials for A Comprehensive Analysis of Hungarian MODY Patients–Part I: Gene Panel Sequencing Reveals Pathogenic Mutations in *HNF1A*, *HNF1B*, *HNF4A*, *ABCC8* and *INS* Genes

Table S1. The list of genes examined with the different library preparation kits.

MODY MASTR kit (Multiplicom, Niel, Belgium)	7 genes: <i>ABCC8</i> , <i>GCK</i> , <i>HNF1A</i> , <i>HNF1B</i> , <i>HNF4A</i> , <i>INS</i> , <i>KCNJ11</i>
custom DNA library preparation kit (Qiagen, GmbH, Hilden, Germany)	17 genes: <i>ABCC8</i> , <i>GCK</i> , <i>HNF1A</i> , <i>HNF1B</i> , <i>HNF4A</i> , <i>INS</i> , <i>KCNJ11</i> , <i>SLC16A1</i> , <i>GLUD1</i> , <i>PDX1</i> , <i>INSR</i> , <i>KLF11</i> , <i>NEUROD1</i> , <i>APPL1</i> , <i>HADH</i> , <i>PAX4</i> , <i>BLK</i>
custom-designed gene panel (Twist Bioscience, South San Francisco, CA, USA)	18 genes: <i>ABCC8</i> , <i>GCK</i> , <i>HNF1A</i> , <i>HNF1B</i> , <i>HNF4A</i> , <i>INS</i> , <i>KCNJ11</i> , <i>SLC16A1</i> , <i>GLUD1</i> , <i>PDX1</i> , <i>INSR</i> , <i>KLF11</i> , <i>NEUROD1</i> , <i>APPL1</i> , <i>HADH</i> , <i>PAX4</i> , <i>BLK</i> , <i>RFX6</i> 20 genes: <i>ABCC8</i> , <i>GCK</i> , <i>HNF1A</i> , <i>HNF1B</i> , <i>HNF4A</i> , <i>INS</i> , <i>KCNJ11</i> , <i>SLC16A1</i> , <i>GLUD1</i> , <i>PDX1</i> , <i>INSR</i> , <i>KLF11</i> , <i>NEUROD1</i> , <i>APPL1</i> , <i>HADH</i> , <i>PAX4</i> , <i>BLK</i> , <i>RFX6</i> , <i>CEL</i> , <i>WFS1</i>

Table S2. Methods used for testing the index patients.

Sample ID	Result	Method
P001	Negative	Sanger: <i>HNF1A</i> ; Sanger: <i>HNF4A</i>
P002	Positive	Sanger: <i>GCK</i>
P003	Negative	Sanger: <i>HNF1A</i>
P004	Negative	Sanger: <i>HNF1A</i>
P015	Positive	Sanger: <i>GCK</i>
P020	Positive	Sanger: <i>HNF1A</i>
P024	Negative	Sanger: <i>HNF1A</i>
P025	Negative	Sanger: <i>HNF1A</i>
P028	Positive	Sanger: <i>GCK</i>
P032	Positive	Sanger: <i>HNF1A</i>
P034	Negative	Sanger: <i>HNF1A</i>
P035	Negative	Sanger: <i>HNF1A</i>
P036	Negative	Sanger: <i>HNF1A</i>
P037	Positive	Sanger: <i>GCK</i>
P046	Negative	Sanger: <i>HNF1A</i>
P047	Positive	Sanger: <i>GCK</i>
P049	Positive	Sanger: <i>HNF1A</i>
P056	Negative	Sanger: <i>GCK</i>
P057	Negative	Sanger: <i>GCK</i>
P058	Positive	Sanger: <i>GCK</i> ; Sanger: <i>HNF1A</i>
P062	Negative	Sanger: <i>HNF1A</i>
P063	Positive	Sanger: <i>GCK</i>
P064	Positive	Sanger: <i>GCK</i>
P068	Negative	Sanger: <i>GCK</i>
P069	Positive	Sanger: <i>GCK</i>
P071	Negative	Sanger: <i>GCK</i> ; Sanger: <i>HNF1A</i>
P072	Negative	Sanger: <i>HNF1A</i>
P073	Negative	Sanger: <i>HNF1A</i>

P074	Positive	Sanger: <i>GCK</i> ; Sanger: <i>HNFB1A</i>
P076	Negative	Sanger: <i>HNFB1A</i> ; Sanger: <i>HNFB4A</i>
P077	Negative	Sanger: <i>GCK</i>
P078	Positive	Sanger: <i>GCK</i>
P080	Negative	Sanger: <i>HNFB1A</i>
P081	Negative	Sanger: <i>HNFB1A</i>
P082	Positive	Sanger: <i>GCK</i>
P085	Positive	Sanger: <i>GCK</i>
P088	Positive	Sanger: <i>GCK</i>
P089	Positive	Sanger: <i>GCK</i>
P101	Negative	Sanger: <i>GCK</i>
P104	Positive	Sanger: <i>GCK</i>
P095	Negative	NGS: TWIST (20 genes)
P099	Negative	NGS: TWIST (20 genes)
P119	Negative	Sanger: <i>GCK</i> ; Sanger: <i>HNFB1A</i>
P120	Negative	Sanger: <i>GCK</i> ; Sanger: <i>HNFB1A</i>
P110	Negative	NGS: TWIST (20 genes)
P112	Negative	NGS: TWIST (20 genes)
P125	Negative	Sanger: <i>HNFB1A</i>
P126	Negative	Sanger: <i>GCK</i>
P127	Negative	Sanger: <i>GCK</i>
P128	Positive	Sanger: <i>GCK</i>
P131	Negative	Sanger: <i>HNFB1A</i> ; Sanger: <i>HNFB4A</i>
P132	Negative	Sanger: <i>HNFB1A</i>
P133	Positive	Sanger: <i>GCK</i>
P140	Negative	Sanger: <i>HNFB1A</i>
P141	Positive	Sanger: <i>GCK</i>
P144	Positive	NGS: Multiplicom (7 genes)
P145	Negative	Sanger: <i>GCK</i> ; Sanger: <i>HNFB1A</i>
P146	Negative	Sanger: <i>GCK</i>
P147	Negative	Sanger: <i>GCK</i>
P149	Negative	MLPA
P150	Positive	MLPA
P138	Positive	NGS: TWIST (20 genes)
P154	Negative	Sanger: <i>GCK</i>
P155	Negative	Sanger: <i>HNFB1A</i>
P157	Positive	Sanger: <i>GCK</i>
P159	Negative	Sanger: <i>GCK</i>
P160	Positive	Sanger: <i>GCK</i> ; Sanger: <i>HNFB1A</i>
P163	Negative	NGS: Multiplicom (7 genes)
P164	Negative	Sanger: <i>HNFB1A</i> ; Sanger: <i>HNFB4A</i> ; MLPA
P166	Negative	MLPA
P168	Positive	Sanger: <i>HNFB4A</i> ; MLPA
P170	Positive	Sanger: <i>GCK</i>
P171	Negative	Sanger: <i>HNFB1A</i> ; Sanger: <i>HNFB4A</i> ; MLPA
P172	Positive	Sanger: <i>GCK</i>
P174	Positive	Sanger: <i>HNFB1A</i>
P177	Positive	Sanger: <i>GCK</i> ; Sanger: <i>HNFB1A</i>
P178	Positive	Sanger: <i>GCK</i> ; Sanger: <i>HNFB1A</i> ; Sanger: <i>HNFB4A</i> ; MLPA

P186	Positive	Sanger: <i>HN1A</i>
P191	Negative	Sanger: <i>HN1A</i>
P192	Positive	Sanger: <i>GCK</i>
P195	Negative	Sanger: <i>HN1A</i> ; Sanger: <i>HN4A</i> ; MLPA
P196	Positive	Sanger: <i>GCK</i> ; Sanger: <i>HN1A</i>
P198	Positive	Sanger: <i>HN1A</i>
P199	Positive	Sanger: <i>GCK</i>
P203	Negative	Sanger: <i>GCK</i>
P204	Positive	Sanger: <i>GCK</i>
P205	Positive	Sanger: <i>GCK</i> ; Sanger: <i>HN1A</i> ; Sanger: <i>HN4A</i>
P207	Negative	Sanger: <i>GCK</i>
P208	Negative	Sanger: <i>GCK</i>
P209	Negative	Sanger: <i>HN1A</i> ; Sanger: <i>HN4A</i> ; MLPA
P210	Positive	Sanger: <i>GCK</i>
P213	Negative	NGS: Qiagen (17 genes)
P214	Positive	Sanger: <i>GCK</i> ; Sanger: <i>HN1A</i> ; Sanger: <i>HN4A</i>
P216	Negative	NGS: Junior
P218	Negative	Sanger: <i>GCK</i> ; Sanger: <i>HN1A</i>
P219	Negative	Sanger: <i>GCK</i> ; Sanger: <i>HN1A</i>
P220	Negative	Sanger: <i>GCK</i> ; Sanger: <i>HN1A</i>
P221	Negative	NGS: Multiplicom (7 genes); MLPA
P222	Positive	Sanger: <i>GCK</i>
P224	Negative	Sanger: <i>GCK</i> ; Sanger: <i>HN1A</i> ; Sanger: <i>HN4A</i> ; MLPA
P225	Negative	Sanger: <i>GCK</i> ; Sanger: <i>HN1A</i> ; Sanger: <i>HN4A</i>
P227	Positive	NGS: Multiplicom (7 genes); MLPA
P228	Negative	Sanger: <i>GCK</i>
P229	Positive	Sanger: <i>HN1A</i>
P230	Negative	NGS: Junior; MLPA
P231	Negative	Sanger: <i>HN1A</i> ; Sanger: <i>HN4A</i>
P232	Positive	Sanger: <i>GCK</i>
P236	Positive	Sanger: <i>GCK</i>
P243	Negative	Sanger: <i>GCK</i> ; Sanger: <i>HN1A</i> ; Sanger: <i>HN4A</i> ; Sanger: <i>KCNJ11</i> ; MLPA
P244	Negative	Sanger: <i>GCK</i> ; MLPA
P245	Negative	Sanger: <i>GCK</i>
P246	Negative	Sanger: <i>HN1A</i> ; Sanger: <i>HN4A</i>
P247	Negative	NGS: Junior
P248	Negative	Sanger: <i>GCK</i> ; Sanger: <i>HN1A</i>
P249	Negative	Sanger: <i>GCK</i>
P250	Positive	NGS: Junior
P254	Negative	Sanger: <i>GCK</i> ; Sanger: <i>HN1A</i> ; MLPA
P255	Negative	Sanger: <i>GCK</i> ; Sanger: <i>HN1A</i>
P256	Negative	NGS: Junior
P257	Positive	NGS: Junior
P262	Negative	NGS: Junior
P263	Negative	NGS: Junior
P264	Negative	NGS: Junior
P265	Negative	NGS: Junior
P266	Positive	NGS: Junior
P269	Negative	NGS: Junior

P271	Positive	NGS: Junior
P272	Negative	NGS: Junior
P273	Positive	NGS: Junior
P274	Negative	NGS: Junior
P275	Negative	NGS: Junior
P276	Negative	NGS: Junior
P277	Positive	NGS: Junior
P278	Negative	NGS: Junior
P279	Negative	NGS: Junior
P280	Positive	NGS: Junior
P285	Positive	NGS: Junior
P287	Negative	MLPA
P288	Positive	NGS: Junior
P289	Negative	NGS: Junior
P290	Positive	NGS: Junior
P294	Negative	NGS: Junior; MLPA
P295	Negative	NGS: Junior
P296	Negative	NGS: Junior
P297	Positive	NGS: Junior; MLPA
P298	Positive	Sanger: GCK
P300	Negative	NGS: Junior
P301	Negative	NGS: Junior; MLPA
P316	Negative	NGS: Junior
P317	Negative	NGS: Multiplicom (7 genes)
P318	Negative	NGS: Multiplicom (7 genes)
P319	Positive	NGS: Multiplicom (7 genes)
P320	Negative	NGS: Multiplicom (7 genes)
P321	Negative	NGS: Qiagen (17 genes)
P322	Negative	NGS: Multiplicom (7 genes)
P323	Positive	NGS: Multiplicom (7 genes)
P324	Positive	NGS: Multiplicom (7 genes)
P325	Negative	NGS: Qiagen (17 genes)
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P336	Negative	NGS: Multiplicom (7 genes)
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P338	Positive	NGS: Multiplicom (7 genes)
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P343	Positive	NGS: Multiplicom (7 genes)
P345	Negative	NGS: Multiplicom (7 genes)
P346	Positive	NGS: Multiplicom (7 genes)
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P351	Negative	NGS: Multiplicom (7 genes)
P352	Negative	NGS: Multiplicom (7 genes)
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P354	Negative	NGS: Multiplicom (7 genes)

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P357	Positive	NGS: Multiplicom (7 genes)
P359	Negative	NGS: Multiplicom (7 genes)
P360	Negative	NGS: Multiplicom (7 genes)
P361	Positive	NGS: Multiplicom (7 genes)
P362	Negative	NGS: Multiplicom (7 genes)
P363	Negative	NGS: Multiplicom (7 genes); MLPA
P364	Positive	NGS: Multiplicom (7 genes)
P366	Negative	NGS: Multiplicom (7 genes)
P367	Negative	NGS: Qiagen (17 genes)
P368	Negative	NGS: Multiplicom (7 genes)
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P384	Positive	NGS: Multiplicom (7 genes)
P387	Negative	NGS: Multiplicom (7 genes)
P388	Negative	NGS: Multiplicom (7 genes)
P389	Negative	NGS: TWIST (18 genes); MLPA
P390	Negative	NGS: Multiplicom (7 genes)
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P395	Negative	NGS: Multiplicom (7 genes)
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P406	Negative	NGS: Multiplicom (7 genes)
P413	Negative	NGS: Multiplicom (7 genes); MLPA
P414	Negative	NGS: Multiplicom (7 genes)
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P430	Negative	NGS: Multiplicom (7 genes)
P431	Negative	NGS: Multiplicom (7 genes); MLPA
P432	Negative	NGS: Multiplicom (7 genes)
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P434	Negative	NGS: Qiagen (17 genes)
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P436	Positive	NGS: Qiagen (17 genes); MLPA

P437	Positive	NGS: Qiagen (17 genes)
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P522	Negative	NGS: Qiagen (17 genes)
P523	Negative	NGS: Qiagen (17 genes); MLPA
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P554	Negative	NGS: Qiagen (17 genes)
P555	Negative	NGS: Qiagen (17 genes)
P556	Positive	NGS: Qiagen (17 genes); MLPA
P557	Positive	NGS: Qiagen (17 genes)
P558	Positive	NGS: Qiagen (17 genes)

P559	Positive	NGS: Qiagen (17 genes)
P564	Negative	NGS: Qiagen (17 genes)
P565	Negative	NGS: Qiagen (17 genes)
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P583	Negative	NGS: Qiagen (17 genes)
P584	Negative	NGS: Qiagen (17 genes)
P585	Positive	NGS: Qiagen (17 genes); MLPA
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P637	Negative	NGS: Qiagen (17 genes)
P638	Negative	NGS: Qiagen (17 genes)
P639	Positive	NGS: Qiagen (17 genes)
P640	Positive	NGS: Qiagen (17 genes)
P641	Positive	NGS: Qiagen (17 genes)
P643	Negative	NGS: TWIST (18 genes)
P644	Negative	NGS: TWIST (18 genes)
P647	Negative	NGS: TWIST (18 genes)
P648	Negative	NGS: TWIST (18 genes)
P649	Negative	NGS: TWIST (18 genes)
P650	Negative	NGS: TWIST (18 genes)
P651	Negative	NGS: TWIST (18 genes)
P652	Negative	NGS: TWIST (18 genes)
P656	Negative	NGS: TWIST (18 genes)
P657	Negative	NGS: TWIST (18 genes)
P658	Negative	NGS: TWIST (18 genes)
P659	Negative	NGS: TWIST (18 genes)
P660	Negative	NGS: TWIST (18 genes)
P661	Negative	NGS: TWIST (18 genes)
P662	Negative	NGS: TWIST (18 genes)
P663	Positive	NGS: TWIST (18 genes)
P664	Negative	NGS: TWIST (18 genes)
P665	Positive	NGS: TWIST (18 genes)
P666	Positive	NGS: TWIST (18 genes)
P669	Negative	NGS: TWIST (18 genes)
P670	Negative	NGS: TWIST (18 genes)
P671	Negative	NGS: TWIST (18 genes)
P672	Negative	NGS: TWIST (18 genes)
P673	Negative	NGS: TWIST (18 genes)
P674	Negative	NGS: TWIST (18 genes)
P675	Positive	NGS: TWIST (18 genes)
P676	Negative	NGS: TWIST (18 genes)
P677	Negative	NGS: TWIST (18 genes)
P678	Negative	NGS: TWIST (18 genes)
P679	Negative	NGS: TWIST (20 genes)
P680	Negative	NGS: TWIST (18 genes)
P681	Negative	NGS: TWIST (18 genes)
P682	Negative	NGS: TWIST (18 genes)
P683	Negative	NGS: TWIST (18 genes)
P684	Positive	NGS: TWIST (18 genes)
P685	Negative	NGS: TWIST (18 genes)
P686	Positive	NGS: TWIST (18 genes)
P689	Negative	NGS: TWIST (18 genes)
P690	Negative	NGS: TWIST (18 genes)
P691	Negative	NGS: TWIST (18 genes)

P692	Negative	NGS: TWIST (18 genes)
P693	Negative	NGS: TWIST (18 genes)
P694	Negative	NGS: TWIST (18 genes)
P695	Negative	NGS: TWIST (18 genes)
P696	Positive	NGS: TWIST (18 genes)
P697	Negative	NGS: TWIST (18 genes)
P698	Negative	NGS: TWIST (18 genes)
P699	Negative	NGS: TWIST (18 genes)
P700	Negative	NGS: TWIST (18 genes)
P701	Negative	NGS: TWIST (18 genes)
P702	Negative	NGS: TWIST (18 genes)
P703	Negative	NGS: TWIST (18 genes)
P704	Negative	NGS: TWIST (18 genes)
P706	Negative	NGS: TWIST (18 genes)
P707	Negative	NGS: TWIST (18 genes)
P708	Negative	NGS: TWIST (18 genes)
P709	Negative	NGS: TWIST (18 genes)
P710	Negative	NGS: TWIST (18 genes)
P711	Negative	NGS: TWIST (18 genes)
P712	Negative	NGS: TWIST (18 genes)
P713	Positive	NGS: TWIST (18 genes)
P715	Negative	NGS: TWIST (18 genes)
P717	Negative	NGS: TWIST (18 genes)
P718	Negative	NGS: Qiagen (17 genes)
P719	Negative	NGS: TWIST (18 genes)
P720	Negative	NGS: TWIST (18 genes)
P721	Negative	NGS: TWIST (18 genes)
P722	Positive	NGS: TWIST (18 genes)
P724	Negative	NGS: TWIST (18 genes)
P725	Negative	NGS: TWIST (18 genes)

Table S3. Clinical data of patients with *HNF1A* mutation.

Family ID	Sample ID	Age at Diagnosis of Diabetes	Age at Receiving Genetic dg	BMI*	Obesity	Complications	Therapy BEFORE Genetic Diagnosis	FPG (0') (mmol/L)	PPG (120') (mmol/L)	HbA1c % (mmol/mol)	MODY Calculator (%)	Family Screening
F007	P020	11	29	22.3	no	N/A	insulin	N/A	N/A	7.8 (61.7)	75.5	multiple generations affected
F007	P021	no diabetes	28	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	multiple generations affected
F007	P022	no diabetes	4	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	multiple generations affected
F007	P023	no diabetes	1	17	no	none	none	N/A	N/A	N/A	N/A	multiple generations affected
F012	P032	N/A	35	25	no	retinopathy, IHD	insulin	8.6	12	13.4 (123.0)	N/A	multiple generations affected
F012	P033	no diabetes	7	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	multiple generations affected
F020	P049	25	47	25.4	no	none	insulin	7.9	9.9	8.2 (66.1)	45.5	multiple generations affected
F020	P050	19	24	27.8	no	none	OAD - metformin	6.8	9.1	6.2 (43.3)	75.5	multiple generations affected
F020	P051	39	69	32.4	yes	retinopathy	OAD - metformin + insulin	7.7	9.8	7.5 (58.5)	N/A	multiple generations affected
F020	P053	no diabetes	24	24.5	no	none	N/A	N/A	N/A	N/A	N/A	multiple generations affected
F025	P058	11	37	23	no	proteinuria, retinopathy	insulin	N/A	N/A	N/A	N/A	multiple generations affected
F025	P059	13	16	27.6	yes	none	insulin	6.0	N/A	7.0 (53.0)	8.2	multiple generations affected
F095	P174	15	44	26	no	none	insulin	N/A	N/A	7.1 (54.1)	6.4	multiple generations affected
F095	P175	no diabetes	27	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	multiple generations affected
F095	P176	no diabetes	24	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	multiple generations affected
F096	P177	12	12	16.9	no	none	insulin	14.8	N/A	N/A	N/A	no family members tested
F098	P186	12	12	19.4	no	none	insulin	6.7	13.6	6.3 (45.4)	49.4	multiple generations affected
F098	P189	30	54	22.5	no	none	insulin	11.3	N/A	7.4 (57.4)	4.6	multiple generations affected

Family ID	Sample ID	Age at Diagnosis of Diabetes	Age at Receiving Genetic dg	BMI*	Obesity	Complications	Therapy BEFORE Genetic Diagnosis	FPG (0') (mmol/L)	PPG (120') (mmol/L)	HbA1c % (mmol/mol)	MODY Calculator (%)	Family Screening
F104	P198	32	32	20.6	no	none	diet	7.81	11.86	6.2 (44.3)	75.5	no family members tested
F128	P229	13	17	24.1	no	none	insulin	8.8	N/A	8.7 (71.6)	1.9	no family members tested
F141	P250	33	37	N/A	N/A	N/A	diet	N/A	N/A	8.5 (69.4)	N/A	multiple generations affected
F141	P252	16	39	N/A	N/A	N/A	OAD – sulphonylurea	N/A	N/A	N/A	N/A	multiple generations affected
F141	P253	42	79	N/A	N/A	N/A	OAD	N/A	N/A	N/A	N/A	multiple generations affected
F155	P273	14	17	27.5	no	none	insulin	N/A	N/A	7.0 (53.0)	1.9	no family members tested
F159	P277	36	44	24.5	no	none	OAD – sulphonylurea	8.2	13.5	8.1 (65.0)	N/A	no family members tested
F204	P346	N/A	42	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	parents not tested
F228	P378	13	15	N/A	N/A	none	insulin	10.9	N/A	7.8 (61.7)	N/A	no family members tested
F234	P384	35	50	36.1	yes	none	OAD - metformin + sulphonylurea	7.0	N/A	7.1 (54.1)	4.6	no family members tested
F259	P418	13	13	23.6	no	N/A	N/A	6.1	15.9	6.5 (47.5)	N/A	multiple generations affected
F259	P422	4	41	N/A	N/A	none	insulin	N/A	N/A	12.0 (107.7)	N/A	multiple generations affected
F283	P447	N/A	13	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	no family members tested
F287	P451	N/A	31	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	no family members tested
F297	P461	12	17	31	yes	none	insulin	15.2	N/A	10.6 (92.4)	6.4	no family members tested
F302	P466	15	28	20.7	no	none	insulin	7.0	14.9	6.0 (42.1)	49.4	no family members tested
F333	P512	17	19	25.3	no	none	diet	4.6	8.9	5.2 (33.3)	75.5	multiple generations affected
F333	P514	45	47	30.8	yes	acanthosis nigricans	OAD - metformin	10.2	N/A	6.9 (51.9)	N/A	multiple generations affected
F361	P545	23	37	22.2	no	none	insulin	4.4	14.8	5.8 (39.9)	75.5	parents not tested
F366	P550	15	26	20.3	no	none	OAD – sulphonylurea	4.2	N/A	5.7 (38.8)	75.5	no family members tested

Family ID	Sample ID	Age at Diagnosis of Diabetes	Age at Receiving Genetic dg	BMI*	Obesity	Complications	Therapy BEFORE Genetic Diagnosis	FPG (0') (mmol/L)	PPG (120') (mmol/L)	HbA1c % (mmol/mol)	MODY Calculator (%)	Family Screening
F380	P567	17	20	24	no	N/A	insulin	N/A	N/A	5.4 (35.5)	49.4	multiple generations affected
F380	P569	27	56	24	no	N/A	insulin	N/A	N/A	7.2 (55.2)	35.8	multiple generations affected
F394	P591	13	32	19.7	no	retinopathy	OAD – sulphonylurea	6.71	N/A	N/A	N/A	no family members tested
F423	P629	N/A	16	N/A	N/A	N/A	OAD - metformin	N/A	N/A	N/A	N/A	parents not tested
F423	P726	no diabetes	14	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	parents not tested
F430	P636	10	10	12.9	no	N/A	diet	6.5	15.5	5.3 (34.4)	75.5	no family members tested
F452	P663	16	18	25.1	no	none	insulin	20	N/A	9.6 (81.4)	75.5	no family members tested
F474	P686	N/A	15	obese	yes	N/A	N/A	7.5	N/A	N/A	N/A	multiple generations affected
F474	P688	no diabetes	37	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	multiple generations affected
F508	P722	N/A	18	N/A	N/A	N/A	insulin	N/A	N/A	8.0 (63.9)	N/A	no family members tested

Index patients are shown in bold. Age at diabetes: N/A—the patient has diabetes, but no information was received regarding the age of diagnosis; no diabetes - no information was given in the application form that the patient shows any signs of diabetes. dg—diagnosis; OAD—oral antidiabetic drug; IHD—Ischemic Heart Disease.

* BMI data refers to the time of referral for genetic testing.

Table S4. Clinical data of patients having a mutation in other MODY-causing genes.

Family ID	Sample ID	Age at Diagnosis of Diabetes	Age at Receiving Genetic dg	BMI*	Obesity	Complications	Therapy BEFORE dg	FPG (0') (mmol/L)	PPG (120') (mmol/L)	HbA1c % (mmol/mol)	MODY Calculator %	Family Screening
F196	P331	32	39	20.3	no	none	OAD - metformin	8.8	N/A	8.2 (66.1)	35.8	multiple generations affected
F196	P332	35	67	28.7	no	retinopathy, nephropathy, uraemia, PAD, proteinuria, IHD, CHF	insulin	6.9	N/A	5.8 (39.9)	4.6	multiple generations affected
F196	P334	17	42	26.8	no	retinopathy	insulin	9.2	N/A	6.7 (49.7)	8.2	multiple generations affected
F305	P469	15	45	30.7	yes	retinopathia, proteinuria	insulin	N/A	N/A	11.0 (96.7)	58	no family members tested
F076	P150	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	no family members tested

Family ID	Sample ID	Age at Diagnosis of Diabetes	Age at Receiving Genetic dg	BMI*	Obesity	Complications	Therapy BEFORE dg	FPG (0') (mmol/L)	PPG (120') (mmol/L)	HbA1c % (mmol/mol)	MODY Calculator %	Family Screening
F091	P168	15	16	27.9	no	renal cysts	OAD - metformin	N/A	N/A	8.7 (71.6)	75.5	siblings positive, parents not tested
F091	P169	no diabetes	19	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	siblings positive, parents not tested
F172	P297	15	17	29.5	yes	N/A	diet	6.1	13.9	6.7 (49.7)	75.5	no family members tested
F372	P556	31	33	23.9	no	proteinuria, renal cysts	insulin	8.2	N/A	8.6 (70.5)	21	no family members tested
F388	P585	27	38	N/A	N/A	none	OAD - sulphonylurea	6.9	11	7.7 (60.7)	N/A	no family members tested
F097	P178	26	42	22	no	none	insulin	N/A	N/A	about 6	49.4	multiple generations affected
F097	P179	10	17	19	no	none	insulin	N/A	N/A	8 - 9	75.5	multiple generations affected
F192	P324	10	16	28	yes	none	diet	8.2	13.3	6.6 (48.6)	75.5	no family members tested
F284	P448	5	8	N/A	N/A	N/A	N/A	N/A	N/A	7.6 (59.6)	N/A	no family members tested
F315	P483	13	16	18.4	no	polyuria, polydipsia	none	6.7	9.4	6.6 (48.6)	75.5	multiple generations affected
F315	P485	43	44	23.7	no	none	diet	6 -7	7.9	7.1 (54.1)	N/A	multiple generations affected

Index patients are shown in bold. Age at diabetes: N/A—the patient has diabetes, but no information was received regarding the age of diagnosis; no diabetes—no information was given in the application form that the patient shows any signs of diabetes. dg—diagnosis; OAD—oral antidiabetic drug; PAD—Peripheral Arterial Disease; IHD—Ischemic Heart Disease; CHF—Congestive Heart Failure. * BMI data refers to the time of referral for genetic testing.