

Table 2. Characteristics of the 20 patients identified with monogenic diabetes

Study ID	Gene	Mutation	Protein effect	Gender	Age at diagnosis (yrs)	Diabetes duration (yrs)*	Initial treatment	Current treatment	BMI centile	Affected parent	UCPCR nmol/mmol	GAD	IA-2	Notes
211	<i>GCK</i>	c.97_117dup	p.(Val33_Lys39dup)	M	3	13	Insulin	None	99th	Mother	3.57	N/A	N/A	Known MODY
537	<i>GCK</i>	c.683C>T	p.(Thr228Met)	M	11	2	Diet	None	N/A	Mother	1.94	N/A	N/A	Known MODY Sibling of 538
538	<i>GCK</i>	c.683C>T	p.(Thr228Met)	M	9	1	Diet	None	N/A	Mother	1.73	N/A	N/A	Known MODY Sibling of 537
543	<i>GCK</i>	c.184G>A	p.(Val62Met)	M	4	0.2	Diet	None	N/A	Mother	N/A	N/A	N/A	Known MODY Sibling of 544
544	<i>GCK</i>	c.184G>A	p.(Val62Met)	M	3	2	Diet	None	N/A	Mother	N/A	N/A	N/A	Known MODY Sibling of 543
1396	<i>GCK</i>	c.1209del	p.(Ile404fs)	M	14	0.3	Diet	None	71st	Mother	N/A	N/A	N/A	Known MODY
8002095	<i>GCK</i>	c.1019G>T	p.(Ser340Ile)	M	9	5	Diet	None	88th	Father	0.79	Neg	N/A	Known MODY
8002372	<i>GCK</i>	c.1340G>A	p.(Arg447Gln)	M	18	0.6	Diet	None	90th	Neither	Not tested	Neg	Not tested	Newly identified MODY
599	<i>HNF1A</i>	c.608G>A	p.(Arg203His)	F	14	0.5	OHA	OHA	99th	Both parents	3.08	Neg	Neg	Known MODY
1012	<i>HNF1A</i>	c.872del	p.(Pro291fs)	F	10	0.7	Diet	Diet	99 th	Mother	5.6	Neg	Neg	Known MODY Sibling of 395
395	<i>HNF1A</i>	c.872del	p.(Pro291fs)	F	14	0.1	OHA	OHA	95th	Mother	5.8	Neg	Neg	Known MODY Sibling of 1012
455	<i>HNF1A</i>	c.872dup	p.(Gly292fs)	F	12	3	OHA	OHA	57th	Father	0.86	Neg	Neg	Known MODY
567	<i>HNF1A</i>	c.872dup	p.(Gly292fs)	M	8	2	Diet	OHA	94th	Mother	1.73	Neg	Neg	Known MODY
686	<i>HNF4A</i>	c.749T>C	p.(Leu250Pro)	M	16	0.7	Diet	Diet	99th	Father	4.74	N/A	N/A	Known MODY
1348	<i>HNF4A</i>	c.340C>T	p.(Arg114Trp)	F	15	0.2	Insulin	OHA	86th	Father	3.00	Neg	Neg	Newly identified MODY
1203	<i>HNF4A</i>	c.340C>T	p.(Arg114Trp)	M	7	2	Insulin	Insulin	39th	Neither	0.21	Neg*	Neg	Dual diagnosis: Newly identified HNF4A / known Type 1
377	<i>HNF4A</i>	c.-12G>A	p.(?)	F	11	2	Insulin	Insulin	99th	Mother	0.28	Neg	Neg	Newly identified MODY
854	<i>HNF1B</i>	c.1- ?_*151+?del	p.(0?) (whole gene deletion)	M	11	2	Insulin	Insulin	9th	Father	0.71	Neg	Neg	Newly identified MODY
555	<i>ABCC8</i>	c.4139G>A	p.(Arg1380His)	F	11	8	OHA	OHA	4 th	Father	3.00	Neg	Neg	Known MODY
758	<i>INSR</i>	c.3706C>G	p.(Pro1236Ala)	F	12	3	OHA	Diet	55th	Mother	9.07	N/A	N/A	Known MODY

*Diabetes duration at time of study

*GAD negative as defined in this study as <99th centile, but GAD 25.9 (97.5th centile)

N/A : Not applicable, genetic diagnosis made prior to study

Mutations described using the Human Genome Variation Society (HGVS) nomenclature guidelines according to the following reference sequences: *GCK* NM_000162.3; *HNF1A* NM_000545.6; *HNF4A* NM_175914.4; *ABCC8* NM_001287174.1; *INSR* NM_000208.2