

Supplement 2: HNF4A polymorphisms observed in Norwegian MODY patients

<i>HNF4A</i> gene location	Codon or relative nucleotide position	Nucleotide change	Designation at protein level	Frequency
5' UTR	nt -67	G to C	-	G – 0.97, C – 0.03
Intron 1a	nt +33	G to A	-	G – 0.99, A – 0.01
Exon 1c	49	GTG to ATG	V49M	G – 0.82, A – 0.18
Intron 1b	nt -38	C to T	-	C – 0.47, T – 0.53
Intron 1b	nt -5	C to T	-	C – 0.84, T – 0.16
Exon 2	58	GCC to GCT	A58A	C – 0.97, T – 0.03
Exon 4	130	ACT to ATT	T130I	C – 0.97, T – 0.03
Intron 4	nt +140	C to T	-	C – 0.99, T – 0.01
Intron 4	nt -52	G to A	-	G – 0.99, A – 0.01
Exon 5	160	GTC to ATC	V160I	G – 0.99, A – 0.01
Exon 7	255	GTG to ATG	V255M	G – 0.99, A – 0.01
Intron 7	nt -88	C to T	-	C – 0.59, T – 0.41

Exon 9

392

AAC to AAT

-

C – 0.99, T – 0.01

Allele frequencies were calculated based on 95 diabetic MODY probands. Previously, V49M, A58A, T130I and V255M have all been reported as polymorphisms. The variant V160I, found in a single subject, is unlikely to be pathogenic since it occurs in an unconserved amino acid residue and since the rat HNF4A gene contains isoleucine in this position.