Hepatocyte nuclear factor 1 regulates the expression of the organic cation transporter OCT1 via binding to an evolutionary conserved region in intron 1 of the *OCT1* gene

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Supplementary *(O'Brien et al.)*

HNF 1A Genotyping Method

OCT1 polymorphisms were genotyped using singe-base primer extension reaction as described previously (Tzvetkov et al., 2009). *HNF1A* polymorphisms were genotyped by multiplex single-base primer extension as follows: QIAGEN Maultiplex PCR Kit (Qiagen, Hilden, Germany) was used to pre-amplify the five DNA regions harboring the analyzed polymorphisms. The amplification conditions were denaturation for 15 min at 95°C, followed by 35 amplification cycles of 30 s at 94°C,
90 s at 64°C and 1 min at 72°C, and a final elongation of 10 min at 72°C. The sequences and the concentrations of the primers used are given in supplementary table 1. The PCR products were purified by incubation with 2.5 U alkaline phosphatase and 3 U exonuclease I (both from MBI-Fermentas) for 1 hour at 37°C followed by an inactivation of the enzymes for 15 min at 80°C. The purified PCR products were used as templates for the single-base primer extension reaction. The reaction was performed using SNaPshot^{TM} multiplex kit (Applied Biosystems) according to the manufacturer's instructions. The sequences and the concentrations of the primers used are given in supplementary table 1. The samples were analyzed on a 3130xl Genetic Analyses capillary sequencer and the genotype calling was performed using GeneMapper version 3.7 (both from Applied Biosystems). All samples were genotyped in duplicate with 100% concordance in the obtained results.

Supplementary table 1. Sequences of the primer used in the multiplex single-base primer extension genotyping of HNF1A SNPs

	Primer name	Sequence	PCR amplicon length	Length of the extended primer	Concentration in the SNaPshot primer mix
PCR pre-amplification	HNF1_PCR1_f	5'-GCCGAGCCATGGTTTCTAAACTGAG-3'	266 bp.		
	HNF1_PCR1_r	5'-GAGGATGGGTGGCGTGAAGTCTTCC-3'			
	HNF1_PCR2_f	5'-CCCCACAGGGAGACCCACAGCAGAG-3'	189 bp		
	HNF1_PCR2_r	5'-CGGCCGCGGCTGGTTTTTCAACAGA-3'	.		
	HNF1_PCR3_f	5'-TGGCCTCCACGCAGGCACAGAGTGT-3'	193 bp.		
	HNF1_PCR3_r	5'-TCACCGTGGGGGCTCTGCAGCTGAG-3'			
	HNF1_PCR4_f	5'-CCTCCTCCCCATCTCTGAAGTCTGA-3'	136 bp.		
	HNF1_PCR4_r	5'-TCTGGAGGGGAGCTGACTTCTCAGT-3'			
	HNF1_PCR5_f	5'-TCTGGCCCAAGACAGGTGCTCAAAA-3'	230 bp		
	HNF1_PCR5_r	5'-AGCCCTCCAGCTCCAGGTGAGAACT-3'	.		
Primer extention reaction (SNaPshot™)	S487N	5'-CCATGGTGGCCATGAAGGGG- 3'		21 bp.	1 µM
	rs7310409	5'- gatcgatCATGACTCACAGGTGGCATC- 3'		29 bp.	0.5 µM
	I27L	5'- $(gact)_3$GGCTGAGCAAAGAGGCACTG- 3'		33 bp.	2 µM
	rs1183910	5'- $(gact)_4$gaATGCGGAAACTGAGGCACAC- 3'		39 bp.	2 µM
	rs1169313	5'- $(gact)_6$gaGCCGCATCTACCCCATGGCC- 3'		43 bp.	2 µM

Supplementary table 2 HNF1A genotypes and HNF1A and OCT1 expression levels in the set of 40 human liver samples

Sample ID	Gender	Age	Ile27Leu rs1169288	rs1183910	rs7310409	Ser487Asn rs2464196	rs1169313	**OCT1 expression** [transcripts / transcript of TPB]
156	male	20	Ile/Ile	C/C	G/G	Ser/Ser	T/T	2.9
162	male	18	Ile/Ile	C/C	G/A	Ser/Asn	T/C	37.3
174	female	23	Ile/Ile	C/C	G/G	Ser/Ser	T/T	120.3
175	male	32	Ile/Ile	C/C	G/G	Ser/Ser	T/C	60.2
200	female	2	Ile/Ile	C/C	G/G	Ser/Ser	T/T	34.6
203	male	13	**Leu/Leu**	T/T	A/A	**Asn/Asn**	C/C	**24.0**
204	female	8	Ile/Leu	C/T	G/A	Ser/Asn	T/C	73.0
217	male	20	Ile/Ile	C/C	G/G	Ser/Ser	T/T	13.9
220	male	36	Ile/Leu	C/T	G/A	Ser/Asn	T/C	9.5
223	male	14	Ile/Leu	C/T	G/A	Ser/Asn	T/C	15.3
251	male	7	**Leu/Leu**	T/T	A/A	**Asn/Asn**	C/C	**8.9**
325	male	60	Ile/Ile	C/C	G/A	Ser/Ser	T/C	26.6
331	female	62	Ile/Ile	C/C	G/G	Ser/Ser	T/T	118.6
332	female	65	Ile/Leu	C/T	G/A	Ser/Asn	T/C	188.4
333	male	59	Ile/Ile	C/C	G/A	Ser/Ser	T/C	41.4
334	male	63	Ile/Ile	C/C	G/G	Ser/Asn	T/C	114.2
335	male	36	Ile/Ile	C/C	G/G	Ser/Ser	T/T	84.7
336	male	70	Ile/Ile	C/C	G/A	Ser/Ser	T/C	11.4
337	male	34	Ile/Ile	C/C	G/G	Ser/Ser	T/T	45.2
340	male	52	Ile/Leu	C/T	G/A	Ser/Asn	T/C	183.3
341	male	2	Ile/Ile	C/C	G/A	Ser/Ser	T/T	8.8
342	male	43	Ile/Leu	C/T	G/A	Ser/Asn	T/C	206.3
343	male	35	Ile/Leu	C/T	A/A	Ser/Ser	T/C	17.2
344	male	63	Ile/Ile	C/C	G/G	Ser/Ser	T/T	65.8
346	male	24	Ile/Leu	C/T	A/A	Ser/Asn	C/C	77.7
347	female	4	Ile/Leu	C/T	G/A	Ser/Asn	T/C	10.8
349	male	2	Ile/Ile	C/C	G/G	Ser/Ser	T/T	2.1
352	male	72	Ile/Ile	C/T	G/A	Ser/Asn	T/C	8.9
353	female	68	Ile/Leu	C/T	A/A	Ser/Asn	C/C	103.3
356	male	37	Ile/Leu	C/T	G/A	Ser/Asn	T/C	102.2
358	male	53	Ile/Ile	C/C	G/G	Ser/Ser	T/T	101.2
360	male	54	Ile/Ile	C/C	G/G	Ser/Ser	T/T	15.2
837	female	40	Ile/Leu	C/T	G/A	Ser/Asn	T/C	107.5
845	female	10	Ile/Ile	C/C	G/G	Ser/Ser	T/T	229.0
849	female	46	Ile/Ile	C/C	G/G	Ser/Ser	T/T	227.1
852	male	69	Ile/Ile	C/C	G/A	Ser/Ser	T/C	249.0
856	male	67	Ile/Leu	C/T	G/A	Ser/Asn	T/C	64.8
860	male	47	Ile/Ile	C/C	G/G	Ser/Ser	T/T	138.8
865	male	34	Ile/Ile	C/C	G/G	Ser/Ser	T/T	127.7
867	male	30	Ile/Leu	C/T	G/A	Ser/Asn	T/C	79.3

TBP, TATA-box binding protein.