Table 2S: Summary of Single Nucleotide Polymorphisms and Insertion/Deletion Variants in the PKLR Region

GENE	LOCATION	RS NUMBER	UCSC Mar2006 build 126	VARIATION	EUROPEAN AMERICAN			AFRICAN AMERICAN		
					CASES	CONTROLS	P-VALUE	CASES	CONTROLS	P-VALUE
ASH1L	Intron 2	rs1360554	153738595	A/G	0.30	0.31	0.6	0.23	0.26	Pool
ASH1L	Intron 3	rs12724079	153700566	C/T	0.35	0.31	0.3	---	---	---
ASH1L	Intron 4	rs12730906	153681392	C/T	0.18	0.13	0.05	---	---	---
ASH1L	Intron 4	rs1325908	153679928	G/T	0.30	0.28	0.45	0.19	0.20	---
ASH1L	Intron 4	rs11264372	153679554	A/G	0.25	0.22	0.34	ND	ND	ND
ASH1L	Intron 25	rs12748814	153577067	C/T	0.09	0.06	0.17	ND	ND	ND
RUSC1	3'UTR	Novel	153567262	A/G	ND	ND	ND	<2%	<2%	<2%
RUSC1	3'UTR	Novel	153567180	A/C	ND	ND	ND	<2%	<2%	<2%
RUSC1	3'UTR	Novel	153567004	G/A	<2%	<2%	<2%	ND	ND	ND
RUSC1	Intron 7	Novel	153565160	13 bp deletion	ND	ND	ND	<2%	<2%	<2%
RUSC1	Intron 6	Novel	153563809	G/T	ND	ND	ND	<2%	<2%	<2%
RUSC1	Intron 5	Novel	153562969	G/A	ND	ND	ND	<2%	<2%	<2%
RUSC1	Intron 4	Novel	153562191	C deletion	ND	ND	ND	<4%	<4%	<4%
RUSC1	Exon 2	Novel	153561470	CCC(pro)20/CCT(Pro)	<2%	<2%	<2%	ND	ND	ND
RUSC1	Exon 2	rs35826120	153561538	TCT(Ser)8/CCT(Leu)	<2%	<2%	<2%	ND	ND	ND
RUSC1	Intron 1	rs4971075	153561396	A/G	LD*	LD*	LD*	LD*	LD*	LD*
c1orf104	Exon 2	Novel	153557674	GTG(Val)76/ATG(Met)	ND	ND	ND	<2%	<2%	<2%
c1orf104	Exon 2	rs16836822	153557211	GAG(Ser)231/GCG(Arg)	ND	ND	ND	0.08	0.03	0.0023 (not in HWE)
FDPS	Exon 11	rs41314549	153556855	GTG(Val)342/CGC(Ala)	<2%	<2%	<2%	ND	ND	ND
FDPS	Intron 9	rs11264361	153556169	G/T	0.28	0.20	0.014	0.22	0.27	0.039
FDPS	Exon 9	Novel	153556089	TTC(Leu)269/GTC(Val)	ND	ND	ND	<2%	<2%	<2%
FDPS	Intron 7	Novel	153555245	G deletion	ND	ND	ND	<2%	<2%	<2%
FDPS	Intron 6	rs34961293	153554808	C/T	ND	ND	ND	ND	ND	ND
FDPS	Intron 5	Novel	153554133	C/T	<2%	<2%	<2%	ND	ND	ND
FDPS	Intron 5	rs1409140	153554230	C/T	ND	ND	ND	ND	ND	ND
FDPS	Intron 4	rs17367421	153553865	C/G	ND	ND	ND	ND	ND	ND
FDPS	Intron 4	Novel	153550112	T/C	ND	ND	ND	<4%	<4%	<4%
FDPS	Intron 4	Novel	153549913	T deletion	ND	ND	ND	<2%	<2%	<2%
FDPS	intron 4	rs11264359	153549453	A/G	0.29	0.22	0.045	0.37	0.35	0.7
FDPS	Ex 3	Novel	153546470	CTC(Leu)63/TTC(Cys)	<2%	<2%	<2%	ND	ND	ND
FDPS	intron 1	rs2297480	153546106	A/C	0.2769	0.1989	0.01	0.2827	0.3343	0.09
PKLR	Promotor	rs12032720	153541584	C/G	0.29	0.22	0.03	0.37	0.42	0.036

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PKLR	Promotor	rs4971072	153540493	A/G	0.34	0.23	0.10	ND	ND	ND
PKLR	Intron 3	rs3020781	153536400	C/T	0.29	0.22	0.02	0.42	0.38	0.25
PKLR	Intron 5	rs2071053	153531801	C/T	0.30	0.22	0.01	0.41	0.37	0.21
PKLR	Intron 10	rs4620533	153529237	C/G	0.29	0.22	0.022	0.45	0.40	0.13
PKLR	Exon 12	rs1052176	153527007	C/T	0.30	0.21	0.007	0.32	0.29	Pool
PKLR	3' UTR	rs1052177	153526974	A/C	0.30	0.21	0.007	0.49	0.48	0.30
PKLR	3' UTR	rs932972	153526720	C/T	0.29	0.21	0.012	0.47	0.45	Pool
HCN3	3' UTR	novel	153525953	C/A	ND	ND	ND	<2%	<2%	<2%
HCN3	3' UTR	rs8847	153525947	C/T	0.28	0.22	0.08	0.46	0.46	0.9
HCN3	3' UTR	rs3814318	153525380	A/G	LD*	LD*	LD*	LD*	LD*	LD*
HCN3	3' UTR	rs3814319	153525376	C/T	0.34	0.28	0.06	0.35	0.32	0.5
HCN3	3' UTR	rs12118947	153525072	A/G	<2%	<2%	<2%	ND	ND	ND
HCN3	Exon 8	novel	153524232	GAG(Glu)560/AAG(Lys)	ND	ND	ND	<1%	<1%	<1%
HCN3	Intron 7	rs11264355	153524116	C/G	0.30	0.22	0.02	0.50	0.47	Pool
HCN3	Intron 6	novel	153524863	T/C	ND	ND	ND	<2%	<2%	<2%
HCN3	Intron 6	novel	153525265	G/C	ND	ND	ND	<2%	<2%	<2%
HCN3	Intron 6	rs11264353	113522530	G/C	0.30	0.19	0.004	0.49	0.53	0.10
HCN3	Intron 6	rs11264352	153522461	T/C	0.30	0.23	0.030	0.49	0.45	0.29
HCN3	Exon 6	novel	153522639	GUG(Val)461/AUG(Met)	ND	ND	ND	<2%	<2%	<2%
HCN3	Exon 6	novel	153522688	CTG(Leu)444/CTT(Leu)	ND	ND	ND	0.12	0.12	0.80
HCN3	Intron 5	novel	153521829	C/T	<2%	<2%	<2%	ND	ND	ND
HCN3	Exon 4	novel	153521083	GUA(Val)334/AUA(Ile)	ND	ND	ND	0.02	0.02	0.72
HCN3	Intron 2	novel	153520330	G/T	<2%	<2%	<2%	ND	ND	ND
HCN3	Intron 2	rs7520184	153520207	A/G	0.29	0.22	0.025	0.48	0.49	0.29
HCN3	Intron 2	rs12049375	153519457	A/G	LD*	LD*	LD*	LD*	LD*	LD*
HCN3	Intron 1	rs7551854	153515621	G/T	ND	ND	ND	0.10	0.09	Pool
HCN3	Intron 1	rs7549276	153515199	G/A	0.25	0.35	Pool	0.43	0.40	Pool
HCN3	Intron 1	rs11264349	153514737	A/T	0.30	0.22	0.014	0.30	0.35	0.07
HCN3	Intron 1	rs12749306	153514331	G/T	0.30	0.22	0.014	0.33	0.36	0.2
HCN3	Exon 1	novel	153514211	AAA(Lys)69/AGA(Arg)	0.02	0.02	0.7	ND	ND	ND
CLK2	Exon 1 (UTR)	novel	153514000	G/C	ND	ND	ND	<2%	<2%	<2%
CLK2	Exon 1 (UTR)	rs12563994	153510716	G/A	0.30	0.23	0.046	0.46	0.43	0.37
CLK2	Exon 1 (UTR)	novel	153509850	G/C	ND	ND	ND	<1%	<1%	<1%
CLK2	Intron 1	novel	153507978	T/G	ND	ND	ND	<1%	<1%	<1%
CLK2	Intron 1	rs2236863	153507985	C/T	0.30	0.22	0.013	0.45	0.43	0.51
CLK2	Exon 2	rs12749700	153507338	CGU(Arg)19/AGU(Ser)	<1%	<1%	<1%	ND	ND	ND

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CLK2	Intron 2	rs2361543	153506248	C/T	0.28	0.21	0.017	0.41	0.41	Pool
CLK2	Intron 5	novel	153504671	G/C	ND	ND	ND	<1%	<1%	<1%
CLK2	Intron 6	rs4971070	153503828	C/T	0.05	0.05	0.97	ND	ND	ND
CLK2	Intron 6	novel	153503494	G/A	<1%	<1%	<1%	ND	ND	ND
CLK2	Intron 6	novel	153503457	C/T	ND	ND	ND	0.11	0.11	0.76
CLK2	Intron 8	rs1078699	153502117	A/G	0.28	0.23	0.078	0.38	0.35	0.36
CLK2	Intron 8	rs1076556	153501445	C/T	0.10	0.08	0.35	0.40	0.37	0.32
CLK2	Intron 8	novel	153501241	T/A	ND	ND	ND	<1%	<1%	<1%
CLK2	Intron 11	novel	153500028	[AAAG] ins/del	ND	ND	ND	0.07	0.07	0.87
SCAMP3	5' Flanking	novel	153498979	[AG] ins/del	ND	ND	ND	<1%	<1%	<1%
SCAMP3	5' Flanking	rs1046188	153498698	A/G	0.30	0.23	0.035	0.47	0.48	0.79
SCAMP3	Intron 1	rs2242576	153498169	A/C	LD*	LD*	LD*	0.40	0.40	1.00
SCAMP3	Exon 3	novel	153494076	CCT(Pro)55/TCT(Ser)	0.02	0.01	0.10	ND	ND	ND
SCAMP3	Exon 4	rs3180018	153496775	GGA(Gly)126/GGG(Gly)	0.29	0.22	0.017	0.44	0.48	0.22
SCAMP3	3' UTR	novel	153489492	C/T	0.01	0.01	0.68	ND	ND	ND
SCAMP3	3' UTR	novel	153489193	A/G	ND	ND	ND	<2%	<2%	<2%
SCAMP3	3' UTR	novel	153488979	A/G	ND	ND	ND	<1%	<1%	<1%
C1ORF2	Intron 1	rs2242577	153490925	A/C	0.28	0.22	0.04	0.40	0.38	0.56
C1ORF2	Intron 5	rs2072647	153488461	G/T	0.28	0.22	0.0639	0.37	0.37	0.736

ND, not detected in this population; Pool, allele frequencies determined in pooled samples with no p value calculated; * = in LD with another typed SNPs and not individually typed. Frequencies shown as less than a percentage were observed in only 1 or two alleles in the screening set and not typed in the full set. P values represent allelic tests of association with type 2 diabetes.