

**CDKAL1 rs7756992 is associated with diabetic retinopathy in a Chinese population with type 2 diabetes**

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Supplementary Table 1 Distributions of SNPs among stage 1 samples

Chr.	SNP	Gene	Minor/major allele	Minor allele frequency			
				Case-control study (1)		Case-control study (2)	
				Controls	DR only	DN only	DR&DN
1	rs2641348	ADAM30	C/T	0.038	0.040	0.033	0.031
1	rs10923931	NOTCH2	T/G	0.040	0.040	0.034	0.032
1	rs340874	PROX1	G/A	0.393	0.440	0.378	0.377
2	rs7578597	THADA	C/T	0.007	0.005	0.006	0.008
2	rs243021	LOC105374756	C/T	0.328	0.315	0.312	0.310
2	rs7593730	RBMS1	T/C	0.145	0.133	0.151	0.152
2	rs3923113	GRB14	G/T	0.110	0.094	0.108	0.144
2	rs13389219	LOC101929615	T/C	0.079	0.078	0.081	0.097
2	rs16856187	G6PC2	C/A	0.274	0.242	0.282	0.252
2	rs7578326	LOC646736	G/A	0.147	0.138	0.158	0.131
2	rs2943641	LOC646736	T/C	0.051	0.072	0.053	0.065
3	rs1801282	PPARG	G/C	0.045	0.054	0.032	0.059
3	rs7612463	UBE2E2	A/C	0.195	0.196	0.200	0.185
3	rs831571	PSMD6	T/C	0.359	0.351	0.356	0.370
3	rs4607103	ADAMTS9-AS2	T/C	0.418	0.354	0.383	0.358
3	rs4402960	IGF2BP2	T/G	0.258	0.278	0.278	0.262
3	rs7651090	IGF2BP2	G/A	0.258	0.283	0.283	0.261
3	rs16861329	ST6GAL1	T/C	0.186	0.198	0.170	0.167
4	rs6815464	MAEA	G/C	0.421	0.413	0.390	0.381
4	rs10010131	WFS1	A/G	0.037	0.062	0.043	0.051
5	rs459193	C5orf67	T/C	0.426	0.409	0.438	0.451
5	rs4457053	ZBED3-AS1	G/A	0.053	0.051	0.049	0.048
6	rs7756992	CDKAL1	A/G	0.475	0.428	0.499	0.414
6	rs9470794	ZFAND3	C/T	0.306	0.328	0.314	0.340
6	rs1535500	KCNK16	T/G	0.434	0.466	0.459	0.495
7	rs2191349	GTF3AP5	G/T	0.316	0.355	0.350	0.348
7	rs864745	JAZF1	G/A	0.208	0.204	0.223	0.224
7	rs1799884	GCK	A/G	0.223	0.280	0.225	0.224

Supplementary Table 1 (Continued) Distributions of SNPs among stage 1 samples

Chr.	SNP	Gene	Minor/major allele	Minor allele frequency			
				Case-control study (1)		Case-control study (2)	
				Controls	DR only	DN only	DR&DN
7	rs917793	<i>YKT6</i>	A/T	0.227	0.271	0.218	0.220
7	rs6467136	<i>LOC105375490</i>	A/G	0.183	0.175	0.203	0.165
7	rs10229583	<i>PAX4</i>	A/G	0.138	0.152	0.173	0.167
7	rs791595	<i>LOC101928423</i>	A/G	0.156	0.163	0.132	0.158
7	rs972283	<i>LOC105375508</i>	A/G	0.246	0.257	0.295	0.265
8	rs516946	<i>ANK1</i>	A/G	0.084	0.109	0.101	0.112
8	rs515071	<i>ANK1</i>	T/C	0.122	0.163	0.150	0.143
8	rs896854	<i>TP53INP1</i>	A/G	0.338	0.307	0.314	0.306
8	rs13266634	<i>SLC30A8</i>	T/C	0.354	0.352	0.374	0.411
9	rs7041847	<i>GLIS3</i>	A/G	0.434	0.486	0.462	0.438
9	rs17584499	<i>PTPRD</i>	T/C	0.091	0.098	0.095	0.100
9	rs10811661	<i>CDKN2A/B</i>	C/T	0.370	0.385	0.410	0.409
9	rs13292136	<i>CHCHD2P9</i>	T/C	0.070	0.075	0.094	0.082
9	rs2796441	<i>LOC101927502</i>	C/T	0.369	0.458	0.405	0.411
9	rs11787792	<i>GPSM1</i>	G/A	0.049	0.034	0.029	0.032
10	rs10906115	<i>CDC123</i>	G/A	0.365	0.356	0.352	0.316
10	rs12779790	<i>CDC123</i>	G/A	0.190	0.174	0.183	0.174
10	rs1802295	<i>VPS26A</i>	T/C	0.110	0.111	0.099	0.078
10	rs12571751	<i>ZMIZ1</i>	G/A	0.410	0.463	0.463	0.436
10	rs1111875	<i>IDE-KIF11-HHEX</i>	C/T	0.314	0.290	0.301	0.289
10	rs7903146	<i>TCF7L2</i>	T/C	0.041	0.040	0.041	0.047
10	rs10886471	<i>GRK5</i>	T/C	0.217	0.198	0.226	0.185
11	rs231362	<i>KCNQ1</i>	T/C	0.070	0.101	0.100	0.093
11	rs2237892	<i>KCNQ1</i>	T/C	0.262	0.255	0.284	0.270
11	rs5219	<i>KCNJ11</i>	T/C	0.434	0.414	0.426	0.408
11	rs10751301	<i>TENM4</i>	C/G	0.230	0.209	0.204	0.209
11	rs1387153	<i>MTNR1B</i>	T/C	0.441	0.439	0.426	0.441
12	rs10842994	<i>LOC105369709</i>	T/C	0.181	0.189	0.188	0.161
12	rs1531343	<i>RPSAP52</i>	C/G	0.164	0.136	0.137	0.128
12	rs7961581	<i>LOC105369832</i>	C/T	0.175	0.245	0.245	0.203
13	rs9552911	<i>SGCG</i>	A/G	0.240	0.212	0.234	0.244
13	rs1359790	<i>LOC105370275</i>	T/C	0.252	0.241	0.273	0.270
15	rs7403531	<i>RASGRP1</i>	T/C	0.366	0.400	0.358	0.418
15	rs7172432	<i>NPM1P47</i>	G/A	0.341	0.363	0.361	0.394
15	rs1436955	<i>NPM1P47</i>	A/G	0.217	0.201	0.216	0.244
15	rs7178572	<i>HMG20A</i>	G/A	0.376	0.384	0.356	0.357
15	rs7177055	<i>LOC101929457</i>	A/G	0.351	0.352	0.349	0.339
15	rs11634397	<i>ZFAND6</i>	G/A	0.083	0.095	0.093	0.129
15	rs2028299	<i>AP3S2</i>	C/A	0.212	0.202	0.214	0.198

Supplementary Table 1 (Continued) Distributions of SNPs among stage 1 samples

Chr.	SNP	Gene	Minor/major allele	Minor allele frequency			
				Case-control study (1)		Case-control study (2)	
				Controls	DR only	DN only	DR&DN
15	rs8042680	<i>PRC1</i>	C/A	0.011	0.005	0.005	0.004
16	rs8050136	<i>FTO</i>	A/C	0.126	0.141	0.156	0.148
16	rs7202877	<i>CTRB1</i>	G/T	0.206	0.184	0.219	0.214
16	rs17797882	<i>MAF</i>	T/C	0.194	0.218	0.202	0.222
16	rs16955379	<i>CMIP</i>	T/C	0.267	0.257	0.239	0.235
17	rs391300	<i>SRR</i>	A/G	0.284	0.283	0.326	0.341
17	rs312457	<i>SLC16A13</i>	C/T	0.120	0.152	0.159	0.161
17	rs13342232	<i>SLC16A11</i>	G/A	0.117	0.147	0.140	0.158
17	rs4430796	<i>HNF1B</i>	G/A	0.393	0.349	0.296	0.300
18	rs12970134	<i>LOC342784</i>	A/G	0.218	0.220	0.212	0.251
19	rs10401969	<i>SUGP1</i>	C/T	0.080	0.089	0.093	0.090
19	rs3786897	<i>PEPD</i>	G/A	0.415	0.450	0.458	0.455
20	rs6017317	<i>FITM2</i>	G/T	0.494	0.477	0.416	0.476
20	rs4812829	<i>HNF4A</i>	A/G	0.487	0.458	0.468	0.450
X	rs5945326	<i>DUSP9</i>	G/A	0.308	0.350	0.355	0.398

Supplementary Table 2 Distribution of rs7756992 among patients with different severities of diabetic retinopathy

	n	Genotype count 11/12/22 <sup>a</sup>	Minor allele frequency	P value for trend analysis
Patients without DR	2,199	608/989/488	0.471	
Mild NPDR	709	242/323/129	0.419	
Moderate NPDR	396	126/174/79	0.438	
Severe NPDR	267	79/117/55	0.452	
PDR	116	38/53/20	0.419	0.96

<sup>a</sup> 11, major allele homozygotes; 12, heterozygotes; 22, minor allele homozygotes

DR = diabetic retinopathy; NPDR = non-proliferative diabetic retinopathy; PDR = proliferative diabetic retinopathy.