

ESM Table 5. Most significantly associated SNPs for Type 2 diabetes ($P \leq 1 \times 10^{-5}$ in meta-analysis) in both individual and combined cohorts (including directly typed and imputed SNPs) in discovery stage.

SNP	Chr	Nearest gene(s)	RA/NRA	HK1 (99 T2D vs 99 controls)					HK2 (388 T2D vs 659 controls)					SHGWA (197 T2D vs 197 controls)					Combined			
				RA Freq	OR (95% CI)	$P_{adjusted}$	IMP	Rsq	RA Freq	OR (95% CI)	$P_{adjusted}$	IMP	Rsq	RA Freq	OR (95% CI)	P	IMP	Rsq	OR (95% CI)	P_{meta} (uncorrect)	P_{het}	I^2
rs10229583*	7	PAX4	G/A	0.849	1.48 (0.85, 2.59)	0.1645	0	1.00	0.835	1.56 (1.14, 2.13)	0.0055	0	1.00	0.825	1.92 (1.32, 2.79)	5.0×10^{-4}	0	1.00	1.66 (1.33, 2.07)	7.7×10^{-6}	0.6455	<0.001
rs2737230	8	TRPS1	A/G	0.671	1.50 (0.97, 2.32)	0.0629	1	0.97	0.672	1.54 (1.20, 1.97)	5.7×10^{-4}	1	0.97	0.639	1.60 (1.19, 2.16)	0.0018	1	0.98	1.55 (1.30, 1.85)	8.0×10^{-7}	0.9648	<0.001
rs2737231	8	TRPS1	A/G	0.671	1.50 (0.97, 2.32)	0.0629	1	0.97	0.672	1.54 (1.20, 1.96)	5.7×10^{-4}	1	0.97	0.639	1.60 (1.19, 2.16)	0.0017	1	0.98	1.55 (1.30, 1.85)	8.1×10^{-7}	0.9644	<0.001
rs727582	8	TRPS1	A/G	0.671	1.50 (0.97, 2.32)	0.0629	1	0.97	0.672	1.54 (1.20, 1.96)	5.8×10^{-4}	1	0.97	0.639	1.60 (1.19, 2.16)	0.0017	1	0.98	1.55 (1.30, 1.85)	8.1×10^{-7}	0.9644	<0.001
rs727581*	8	TRPS1	T/C	0.674	1.48 (0.95, 2.28)	0.0778	1	0.96	0.675	1.54 (1.21, 1.98)	5.2×10^{-4}	1	0.96	0.648	1.64 (1.21, 2.22)	0.0013	1	0.95	1.56 (1.31, 1.86)	6.3×10^{-7}	0.9203	<0.001
rs2737234	8	TRPS1	A/G	0.670	1.50 (0.97, 2.31)	0.0630	1	0.97	0.672	1.53 (1.20, 1.96)	6.0×10^{-4}	1	0.97	0.639	1.60 (1.19, 2.16)	0.0017	1	0.98	1.55 (1.30, 1.85)	8.4×10^{-7}	0.9622	<0.001
rs2178950	8	TRPS1	G/C	0.673	1.47 (0.95, 2.26)	0.0785	1	0.97	0.674	1.48 (1.16, 1.90)	0.0016	1	0.97	0.639	1.60 (1.19, 2.16)	0.0017	1	0.98	1.52 (1.28, 1.81)	2.6×10^{-6}	0.9094	<0.001
rs2721956	8	TRPS1	G/A	0.670	1.50 (0.97, 2.31)	0.0631	1	0.97	0.671	1.53 (1.20, 1.96)	6.1×10^{-4}	1	0.97	0.639	1.60 (1.19, 2.16)	0.0017	1	0.98	1.55 (1.30, 1.85)	8.4×10^{-7}	0.9609	<0.001
rs1983541	8	TRPS1	C/T	0.704	1.54 (0.95, 2.50)	0.0762	1	0.84	0.702	1.64 (1.25, 2.15)	3.0×10^{-4}	1	0.84	0.671	1.63 (1.18, 2.26)	0.0028	1	0.86	1.62 (1.34, 1.96)	7.3×10^{-7}	0.9749	<0.001
rs2721957	8	TRPS1	G/A	0.670	1.50 (0.97, 2.31)	0.0631	1	0.97	0.671	1.53 (1.20, 1.96)	6.3×10^{-4}	1	0.97	0.639	1.61 (1.19, 2.17)	0.0017	1	0.98	1.55 (1.30, 1.85)	8.6×10^{-7}	0.9590	<0.001
rs2721959	8	TRPS1	T/G	0.670	1.50 (0.97, 2.31)	0.0632	1	0.97	0.671	1.53 (1.20, 1.96)	6.4×10^{-4}	1	0.97	0.639	1.61 (1.19, 2.17)	0.0016	1	0.98	1.55 (1.30, 1.85)	8.6×10^{-7}	0.9577	<0.001
rs2721960*	8	TRPS1	G/A	0.704	1.54 (0.95, 2.50)	0.0763	1	0.84	0.702	1.64 (1.25, 2.15)	3.0×10^{-4}	1	0.84	0.671	1.63 (1.18, 2.26)	0.0028	1	0.86	1.62 (1.34, 1.96)	7.2×10^{-7}	0.9748	<0.001
rs2737239	8	TRPS1	A/G	0.670	1.50 (0.97, 2.31)	0.0632	1	0.97	0.671	1.53 (1.19, 1.96)	6.5×10^{-4}	1	0.97	0.639	1.61 (1.19, 2.17)	0.0016	1	0.98	1.55 (1.30, 1.85)	8.7×10^{-7}	0.9572	<0.001
rs2737240	8	TRPS1	A/G	0.724	1.35	0.1870	1	0.96	0.729	1.43	0.0066	1	0.95	0.698	1.77	4.2×10^{-4}	1	0.95	1.52	9.9×10^{-6}	0.5136	<0.001

					(0.86, 2.10)					(1.10, 1.86)									(1.28, 2.44)				(1.26, 1.82)				
rs2737241	8	<i>TRPS1</i>	G/A	0.670	1.50 (0.97, 2.31)	0.0634	1	0.97	0.670	1.53 (1.19, 1.95)	6.8×10 ⁻⁴	1	0.96	0.639	1.61 (1.19, 2.17)	0.0016	1	0.98	1.55 (1.30, 1.84)	9.0×10 ⁻⁷	0.9552	<0.001					
rs2737242	8	<i>TRPS1</i>	T/C	0.670	1.50 (0.97, 2.31)	0.0633	1	0.97	0.670	1.53 (1.19, 1.95)	6.8×10 ⁻⁴	1	0.96	0.639	1.61 (1.19, 2.17)	0.0016	1	0.98	1.55 (1.30, 1.84)	9.0×10 ⁻⁷	0.9552	<0.001					
rs2737243	8	<i>TRPS1</i>	G/C	0.670	1.50 (0.97, 2.31)	0.0633	1	0.97	0.670	1.53 (1.19, 1.95)	6.9×10 ⁻⁴	1	0.96	0.639	1.61 (1.19, 2.17)	0.0016	1	0.98	1.55 (1.30, 1.84)	9.1×10 ⁻⁷	0.9546	<0.001					
rs2737244	8	<i>TRPS1</i>	A/T	0.670	1.50 (0.97, 2.31)	0.0634	1	0.97	0.670	1.53 (1.19, 1.95)	6.9×10 ⁻⁴	1	0.96	0.639	1.61 (1.19, 2.17)	0.0016	1	0.98	1.55 (1.30, 1.84)	8.5×10 ⁻⁷	0.9533	<0.001					
rs2721962	8	<i>TRPS1</i>	T/A	0.652	1.46 (0.95, 2.24)	0.0838	1	0.98	0.644	1.44 (1.13, 1.83)	0.0031	1	0.97	0.618	1.63 (1.21, 2.19)	9.5×10 ⁻⁴	1	0.99	1.50 (1.27, 1.79)	3.2×10 ⁻⁶	0.8047	<0.001					
rs2178951	8	<i>TRPS1</i>	A/G	0.655	1.43 (0.93, 2.21)	0.1022	1	0.98	0.646	1.45 (1.14, 1.85)	0.0026	1	0.97	0.622	1.63 (1.21, 2.19)	0.0010	1	0.99	1.51 (1.27, 1.79)	3.4×10 ⁻⁶	0.8118	<0.001					
rs2205380	8	<i>TRPS1</i>	G/A	0.651	1.45 (0.95, 2.23)	0.0850	1	0.99	0.642	1.43 (1.12, 1.81)	0.0037	1	0.98	0.619	1.63 (1.22, 2.19)	9.1×10 ⁻⁴	1	0.99	1.50 (1.26, 1.78)	3.9×10 ⁻⁶	0.7780	<0.001					
rs2737247*	8	<i>TRPS1</i>	A/G	0.651	1.45 (0.95, 2.23)	0.0852	0	1.00	0.640	1.41 (1.11, 1.79)	0.0048	0	1.00	0.619	1.63 (1.22, 2.18)	8.9×10 ⁻⁴	0	1.00	1.49 (1.25, 1.76)	4.9×10 ⁻⁶	0.7400	<0.001					
rs2737248	8	<i>TRPS1</i>	A/G	0.651	1.45 (0.95, 2.23)	0.0848	1	0.99	0.640	1.41 (1.11, 1.79)	0.0048	1	0.99	0.619	1.63 (1.22, 2.18)	8.9×10 ⁻⁴	1	0.99	1.49 (1.25, 1.76)	4.9×10 ⁻⁶	0.7400	<0.001					
rs2737250*	8	<i>TRPS1</i>	A/G	0.654	1.46 (0.95, 2.23)	0.0812	0	1.00	0.641	1.40 (1.10, 1.77)	0.0058	0	1.00	0.617	1.64 (1.22, 2.19)	8.3×10 ⁻⁴	1	0.99	1.48 (1.25, 1.76)	5.6×10 ⁻⁶	0.7112	<0.001					
rs10965243	9	<i>CDKN2A/B</i>	A/G	0.596	2.13 (1.31, 3.45)	0.0017	1	0.82	0.623	1.44 (1.14, 1.82)	0.0019	0	1.00	0.621	1.42 (1.07, 1.89)	0.0135	0	1.00	1.51 (1.26, 1.81)	8.6×10 ⁻⁶	0.3268	0.106					
rs10965245	9	<i>CDKN2A/B</i>	G/A	0.596	2.12 (1.31, 3.44)	0.0017	1	0.82	0.623	1.44 (1.14, 1.82)	0.0019	1	0.98	0.621	1.42 (1.07, 1.89)	0.0138	1	0.99	1.51 (1.26, 1.81)	7.3×10 ⁻⁶	0.3310	0.096					
rs3858158*	10	<i>COL13A1</i>	T/C	0.586	1.55 (0.97, 2.46)	0.0628	1	0.82	0.569	1.71 (1.33, 2.20)	2.4×10 ⁻⁵	1	0.83	0.604	1.33 (0.98, 1.81)	0.0696	1	0.83	1.55 (1.29, 1.85)	2.1×10 ⁻⁶	0.4686	<0.001					
rs2395272*	10	<i>COL13A1</i>	G/A	0.583	1.50 (0.96, 2.33)	0.0728	1	0.89	0.566	1.67 (1.31, 2.13)	2.5×10 ⁻⁵	1	0.89	0.605	1.31 (0.97, 1.76)	0.0794	1	0.89	1.51 (1.27, 1.80)	3.1×10 ⁻⁶	0.4497	<0.001					
rs57703465*	10	<i>COL13A1</i>	G/A	0.717	1.57 (0.86, 2.86)	0.1363	1	0.58	0.704	1.95 (1.40, 2.70)	4.9×10 ⁻⁵	1	0.59	0.745	1.45 (0.94, 2.24)	0.0901	1	0.54	1.72 (1.35, 2.19)	9.1×10 ⁻⁶	0.5446	<0.001					
rs3861798	12	<i>P2RX7</i>	T/C	0.762	1.29 (0.78, 2.15)	0.3212	1	0.98	0.751	1.60 (1.22, 2.09)	4.9×10 ⁻⁴	1	0.97	0.712	1.67 (1.20, 2.32)	0.0022	1	0.98	1.57 (1.30, 1.91)	4.0×10 ⁻⁶	0.7063	<0.001					
chr12:12004	12	<i>P2RX7</i>	C/T	0.744	1.40	0.2345	1	0.78	0.762	1.63	9.8×10 ⁻⁴	1	0.83	0.732	1.73	0.0026	1	0.83	1.63	6.7×10 ⁻⁶	0.8136	<0.001					

9816					(0.80, 2.43)					(1.21, 2.20)										(1.20, 2.50)							(1.32, 2.01)
rs11065445	12	P2RX7	C/G	0.733	1.42 (0.84, 2.39)	0.1844	1	0.88	0.742	1.52 (1.17, 1.99)	0.0016	1	0.97	0.717	1.65 (1.18, 2.30)	0.0029	1	0.98	1.55 (1.28, 1.88)	8.4×10 ⁻⁶	0.8818	<0.001					
rs684201*	12	P2RX7	G/A	0.733	1.42 (0.84, 2.40)	0.1834	1	0.88	0.742	1.52 (1.17, 1.99)	0.0016	1	0.97	0.717	1.64 (1.18, 2.30)	0.0029	1	0.98	1.55 (1.28, 1.88)	8.6×10 ⁻⁶	0.8864	<0.001					
rs3900976	12	P2RX7	C/T	0.733	1.38 (0.82, 2.33)	0.2208	1	0.88	0.741	1.52 (1.16, 1.98)	0.0018	1	0.97	0.717	1.66 (1.19, 2.31)	0.0025	1	0.98	1.54 (1.27, 1.87)	9.8×10 ⁻⁶	0.8360	<0.001					
rs79362551	12	P2RX7	G/A	0.733	1.39 (0.83, 2.35)	0.2109	1	0.88	0.742	1.52 (1.17, 1.98)	0.0017	1	0.97	0.717	1.65 (1.18, 2.30)	0.0027	1	0.98	1.55 (1.27, 1.87)	9.7×10 ⁻⁶	0.8567	<0.001					
rs11065452	12	P2RX7	G/T	0.734	1.44 (0.85, 2.43)	0.1703	1	0.87	0.742	1.53 (1.17, 2.00)	0.0015	1	0.97	0.717	1.64 (1.17, 2.29)	0.0032	1	0.98	1.55 (1.28, 1.88)	8.5×10 ⁻⁶	0.9069	<0.001					
s11065453*	12	P2RX7	C/T	0.772	1.38 (0.81, 2.34)	0.2355	1	0.93	0.750	1.62 (1.23, 2.12)	4.2×10 ⁻⁴	1	0.96	0.721	1.66 (1.19, 2.32)	0.0025	1	0.97	1.60 (1.31, 1.94)	2.9×10 ⁻⁶	0.8362	<0.001					
rs10849849	12	P2RX7	A/G	0.772	1.52 (0.89, 2.62)	0.1237	1	0.91	0.755	1.63 (1.23, 2.15)	4.8×10 ⁻⁴	1	0.92	0.732	1.56 (1.11, 2.21)	0.0099	1	0.91	1.59 (1.30, 1.95)	5.7×10 ⁻⁶	0.9704	<0.001					
rs10849850	12	P2RX7	A/G	0.769	1.58 (0.94, 2.67)	0.0813	1	0.98	0.749	1.63 (1.25, 2.14)	2.8×10 ⁻⁴	1	0.98	0.725	1.50 (1.09, 2.08)	0.0134	1	0.99	1.58 (1.30, 1.92)	3.5×10 ⁻⁶	0.9286	<0.001					
rs208289	12	P2RX7	A/G	0.721	1.55 (0.91, 2.63)	0.1064	1	0.82	0.709	1.64 (1.24, 2.16)	3.8×10 ⁻⁴	1	0.85	0.692	1.51 (1.08, 2.11)	0.0154	1	0.86	1.58 (1.29, 1.92)	6.4×10 ⁻⁶	0.9312	<0.001					
rs208292	12	P2RX7	A/G	0.765	1.50 (0.90, 2.51)	0.1176	1	0.99	0.746	1.64 (1.26, 2.14)	2.2×10 ⁻⁴	1	0.99	0.724	1.47 (1.06, 2.04)	0.0184	1	0.99	1.56 (1.29, 1.89)	5.2×10 ⁻⁶	0.8717	<0.001					
rs10849851*	12	P2RX7	A/G	0.765	1.50 (0.90, 2.51)	0.1179	0	1.00	0.746	1.64 (1.26, 2.14)	2.2×10 ⁻⁴	0	1.00	0.724	1.47 (1.07, 2.04)	0.0185	0	1.00	1.56 (1.29, 1.89)	5.1×10 ⁻⁶	0.8704	<0.001					
rs6489794	12	P2RX7	G/A	0.765	1.50 (0.90, 2.51)	0.1174	1	0.99	0.746	1.64 (1.26, 2.14)	2.2×10 ⁻⁴	1	0.99	0.724	1.47 (1.07, 2.04)	0.0180	1	0.99	1.56 (1.29, 1.89)	4.9×10 ⁻⁶	0.8706	<0.001					
rs11065458	12	P2RX7	C/T	0.767	1.56 (0.93, 2.62)	0.0872	1	0.99	0.747	1.64 (1.25, 2.15)	2.6×10 ⁻⁴	1	0.98	0.724	1.48 (1.07, 2.05)	0.0170	1	0.99	1.57 (1.30, 1.91)	4.4×10 ⁻⁶	0.8894	<0.001					

“*” refers to the SNP selected for stage 2 replication. Nearest Entrez genes within 250 kb. $P_{adjusted}$, P , P_{meta} and P_{het} represent P values from logistic regression with/without adjustment for age and sex under the additive genetic model, meta-analysis under a random effects models (uncorrected for genomic control) and test of heterogeneity, respectively. Risk allele refers to the allele with a higher frequency in T2D patients than in controls in stage 1 meta-analysis. OR, odds ratio are reported with respect to the risk allele. RA, risk allele; NRA, non-risk allele; IMP, 1 = imputed, 2 = genotyped; Rsq, indicates the imputation quality provided from MACH.