

Table S2. Top SNP associations in the 9p21.3 region evaluated in Hispanic ALL patients from CCLS and validated in European-ancestry ALL patients from COG, with functional annotation.

rsID ¹	Position	MAF ²	P _{Meta} ³	OR _{meta} ³	P _{CCLS} ⁴	OR _{CCLS} ⁴	P _{COG} ⁵	OR _{COG} ⁵	Functional Annotation	RegulomeDB Score ⁶	Promoter ⁷	Enhancer ⁷	DNase Hypersensitivity ⁷	Motifs altered ⁸	eQTL ⁹
rs3731249	21970916	0.02	2.42E-12	2.92	3.78E-04	2.77	1.51E-09	2.99	missense	4		Yes			NA
rs113650570	21976402	0.02	2.66E-12	2.94	4.22E-04	2.82	1.51E-09	2.99	intronic	4	Yes	Yes			NA
rs36228834	21975319	0.02	2.68E-12	2.94	4.25E-04	2.82	1.51E-09	2.99	intronic	3a	Yes	Yes	Yes	Myb,MYBL	NA
rs56018935	21966527	0.10	4.29E-09	1.63	7.08E-04	1.92	1.00E-06	1.57	intergenic	4					CDKN2B
rs78545330	21995941	0.09	3.05E-08	1.59	5.27E-04	1.93	7.92E-06	1.52	intronic	2b	Yes	Yes	Yes	REST	NA
rs3218018	21998139	0.08	3.83E-08	1.61	3.25E-04	2.10	1.10E-05	1.52	intronic	5		Yes			CDKN2B
rs2069426	22006273	0.08	4.23E-08	1.60	3.23E-04	2.10	1.19E-05	1.51	intronic	2b		Yes	Yes	REST,Ascl2	CDKN2B
rs77920300	22012441	0.08	4.76E-08	1.60	3.15E-04	2.10	1.33E-05	1.51	intronic	NA					NA
rs3218007	21999800	0.09	5.28E-08	1.56	6.88E-04	1.88	1.12E-05	1.50	intronic	NA		Yes			CDKN2B
rs2069422	22008026	0.09	5.33E-08	0.63	4.58E-04	0.50	1.22E-05	0.67	intronic	5		Yes		CEBPA	CDKN2B
rs3218002	22000841	0.09	5.34E-08	1.56	6.86E-04	1.88	1.13E-05	1.50	intronic	5	Yes			Sox	CDKN2B
rs3218005	22000247	0.09	5.61E-08	1.56	7.53E-04	1.87	1.12E-05	1.50	intronic	5		Yes			CDKN2B
rs974336	22006348	0.09	6.48E-08	1.56	6.52E-04	1.89	1.38E-05	1.49	intronic	4	Yes	Yes	Yes		CDKN2B
rs2811712	21998035	0.10	6.59E-08	0.65	1.04E-03	0.55	1.06E-05	0.67	intronic	6		Yes		Dlx3	CDKN2B
rs77728904	22057530	0.07	6.76E-08	1.65	1.31E-03	2.01	8.52E-06	1.58	intronic	NA					NA
rs77284052	22025885	0.09	7.28E-08	1.58	2.99E-04	2.11	1.96E-05	1.49	intronic	NA					CDKN2B
rs116729641	22025432	0.09	7.39E-08	1.58	3.00E-04	2.11	1.99E-05	1.49	intronic	5					CDKN2B
rs79182326	22034267	0.09	7.92E-08	1.58	3.32E-04	2.10	1.98E-05	1.49	intronic	6		Yes		VAX,ELF,PA X4,ARID3A	CDKN2B
rs79985856	22033824	0.09	8.08E-08	1.58	3.43E-04	2.09	1.98E-05	1.49	intronic	5		Yes			CDKN2B
rs76213463	22040839	0.09	8.58E-08	1.58	3.67E-04	2.08	2.02E-05	1.49	intronic	4		Yes			CDKN2B
rs80166549	22041155	0.09	8.62E-08	1.58	3.69E-04	2.08	2.02E-05	1.49	intronic	4		Yes			CDKN2B
rs72652411	22085028	0.05	9.11E-08	1.87	2.00E-04	2.72	3.48E-05	1.71	intronic	4					
rs17694555	22051295	0.07	1.00E-07	1.65	1.45E-04	2.44	3.23E-05	1.53	intronic	4	Yes	Yes	Yes		CDKN2B
rs3731246	21971989	0.10	1.42E-07	1.55	2.13E-03	1.80	1.30E-05	1.50	intronic	5					CDKN2B
rs115939893	22054821	0.08	1.46E-07	1.58	1.42E-03	2.04	1.32E-05	1.51	intronic	6				ER	CDKN2B
rs74655961	22054164	0.08	1.75E-07	1.58	1.46E-03	2.04	1.53E-05	1.51	intronic	5					CDKN2B
rs17694572	22054356	0.08	1.80E-07	1.58	1.45E-03	2.04	1.56E-05	1.51	intronic	1f					CDKN2B
rs1333035	22044059	0.10	1.87E-07	0.66	3.60E-04	0.52	4.84E-05	0.70	intronic	4		Yes			CDKN2B
rs2427853	21950472	0.09	1.89E-07	0.62	3.25E-03	0.50	1.03E-05	0.65	intergenic	NA					CDKN2B
rs598664	22027551	0.09	2.08E-07	0.65	4.24E-04	0.49	4.11E-05	0.69	intronic	6		Yes		FOXP1	CDKN2B

rs2518716	21956470	0.09	2.29E-07	0.62	1.78E-03	0.48	1.70E-05	0.65	intergenic	NA						CDKN2B
rs2427852	21950432	0.09	2.80E-07	0.62	3.33E-03	0.50	1.45E-05	0.65	intergenic	6					Evi1,LEF1,T CF7,FOXP1, GATA4	CDKN2B
rs17756311	22053895	0.08	3.20E-07	1.56	1.48E-03	2.04	2.60E-05	1.49	intronic	6		Yes				NA
rs1333034	22044122	0.09	3.23E-07	0.66	5.09E-04	0.50	5.64E-05	0.69	intronic	5		Yes			TCF3	CDKN2B
rs72654280	22121357	0.06	3.25E-07	1.80	4.13E-04	2.85	4.98E-05	1.66	intergenic	NA						CDKN2B
rs72652409	22083994	0.05	3.57E-07	1.83	4.80E-04	2.63	7.04E-05	1.69	intronic	NA						CDKN2B
rs2263145	21950452	0.10	3.74E-07	0.65	1.74E-02	0.62	6.90E-06	0.66	intergenic	NA					AP1,PAX4, PAX5	CDKN2A
rs2811709	21980151	0.10	4.93E-07	0.66	2.03E-03	0.56	4.25E-05	0.69	intronic	6		Yes			MEF2	NA
rs662463	22030438	0.09	5.25E-07	0.66	4.93E-04	0.50	8.56E-05	0.70	intronic	1f	Yes	Yes	Yes	SkMC	CDKN2B	
rs2518721	21979204	0.11	5.29E-07	0.66	1.79E-03	0.56	5.00E-05	0.69	intronic	6		Yes			DMRT2,IRF 2,STAT1, ICSBP	CDKN2B
rs572738724	22095728	0.03	1.01E-06	2.08	2.07E-04	3.33	3.42E-04	1.83	intronic	NA	NA	NA	NA	NA	NA	NA
rs115574830	22059061	0.09	2.08E-06	1.51	1.23E-03	2.01	1.71E-04	1.43	intronic	NA						CDKN2B

¹42 SNPs having smaller p-values and larger effect sizes than tag SNP rs3731217 in CCLS discovery analyses, and also associated at P<1.1x10⁻³ in COG validation analyses, appear here. ²MAF calculated in CCLS and WTCCC controls combined; ³Totaling 1301 cases and 3078 controls; ⁴Totaling 321 Hispanic cases and 454 Hispanic controls; ⁵Totaling 980 European-ancestry cases and 2624 controls; ⁶RegulomeDB scores: 1f eQTL+TF binding/DNase peak; 2b, TF binding+any motif+DNase Footprint+DNase peak; 3a, TF binding+any motif+DNase peak; 4, TF binding+DNase peak; 5, TF binding or DNase peak; 6, other. ⁷Promoter Histone Marks, Enhancer Histone Marks and DNase hypersensitivity sites appearing in blood and extracted from ENCODE2 data using HaploRegV3; ⁸Altered motifs from footprinting and position-weight matrix analysis, extracted from RegulomeDB; ⁹Genotype-Tissue Expression (GTEx) project association p-values <0.05, in whole-blood, were taken to indicate a significant eQTL.