

# GENOME-WIDE ASSOCIATION STUDY IDENTIFIES TWO LOCI STRONGLY AFFECTING GLYCAN VARIATION IN CARBOHYDRATE-DEFICIENT TRANSFERRIN

## SUPPLEMENTARY DATA

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**Supplementary Table 1.** SNPs with significant ( $p < 5 \times 10^{-8}$ ) association with CDT percent in the initial meta-analysis, using data obtained by all three methods (capillary electrophoresis, CDText and N-Latex CDT). SNPs are ordered by their chromosomal location (HapMap release 27, February 2010, on NCBI B36 assembly). P values are also shown for heterogeneity of the associations across methods and for the allelic associations from meta-analysis of the capillary electrophoresis and N-Latex CDT methods only.

SNP	Chromosome	Location, bp (PhaseIII/Rel#2, Feb09)	Rank	Type	Closest gene	P (meta-analysis, all 3 methods)	P (hetero- geneity)	P (meta-analysis, CE and Latex methods)
rs2301054	1	63,879,616	74	INTRONIC	<i>PGM1</i>	6.81E-08	0.112	1.04E-06
rs2269247	1	63,879,872	86	INTRONIC	<i>PGM1</i>	2.98E-07	0.135	7.05E-06
rs2269246	1	63,880,079	77	INTRONIC	<i>PGM1</i>	7.45E-08	0.118	1.13E-06
rs2269245	1	63,880,481	76	INTRONIC	<i>PGM1</i>	7.43E-08	0.118	1.13E-06
rs2269241	1	63,881,359	69	INTRONIC	<i>PGM1</i>	3.57E-08	0.126	5.49E-07
rs2269240	1	63,881,852	78	INTRONIC	<i>PGM1</i>	8.21E-08	0.116	1.24E-06
rs3790857	1	63,884,454	83	INTRONIC	<i>PGM1</i>	1.37E-07	0.105	2.02E-06
rs11576729	1	63,887,017	79	INTRONIC	<i>PGM1</i>	9.15E-08	0.104	1.50E-06
rs2269238	1	63,890,125	75	INTRONIC	<i>PGM1</i>	7.19E-08	0.104	1.18E-06
rs2269235	1	63,891,503	68	INTRONIC	<i>PGM1</i>	3.45E-08	0.094	7.59E-07
rs2269234	1	63,891,564	70	INTRONIC	<i>PGM1</i>	4.47E-08	0.091	9.73E-07

SNP	Chromosome	Location, bp (PhasIII/Rel#2, Feb09)	Rank	Type	Closest gene	P (meta-analysis, all 3 methods)	P (hetero- geneity)	P (meta-analysis, CE and Latex methods)
rs2269233	1	63,891,934	67	INTRONIC	<i>PGM1</i>	3.15E-08	0.096	6.99E-07
rs12027168	1	63,893,201	72	INTRONIC	<i>PGM1</i>	6.25E-08	0.094	1.46E-06
rs2749100	1	63,897,567	91	INTRONIC	<i>PGM1</i>	7.63E-07	0.379	1.03E-05
rs11208264	1	63,897,660	95	INTRONIC	<i>PGM1</i>	9.37E-07	0.371	1.25E-05
rs8294	1	63,897,956	55	3PRIME_UTR	<i>PGM1</i>	2.32E-09	0.218	9.10E-08
rs4643	1	63,898,027	54	3PRIME_UTR	<i>PGM1</i>	2.31E-09	0.218	9.06E-08
rs855350	1	63,899,592	96	DOWNSTREAM	<i>PGM1</i>	9.58E-07	0.278	1.56E-05
rs2749097	1	63,900,056	52	DOWNSTREAM	<i>PGM1</i>	1.89E-09	0.224	7.52E-08
rs1506397	1	63,917,871	71	INTERGENIC	<i>PGM1</i>	6.01E-08	0.429	9.21E-07
rs7548548	1	63,923,957	80	INTERGENIC	<i>PGM1</i>	1.04E-07	0.596	9.21E-07
rs4854723	3	134,736,948	66	INTERGENIC		2.78E-08	0.021	1.25E-09
rs4854727	3	134,740,575	93	INTERGENIC		8.08E-07	0.075	1.14E-07
rs4017074	3	134,743,117	92	DOWNSTREAM		7.76E-07	0.075	1.10E-07
rs3860498	3	134,743,206	94	DOWNSTREAM		9.11E-07	0.075	1.44E-07
rs6785365	3	134,745,135	88	DOWNSTREAM		4.64E-07	0.094	7.96E-08
rs6763627	3	134,745,257	89	DOWNSTREAM		4.71E-07	0.094	8.12E-08
rs10935068	3	134,745,497	87	DOWNSTREAM		4.61E-07	0.094	7.90E-08
rs7614812	3	134,746,941	61	DOWNSTREAM		1.26E-08	0.031	7.20E-10
rs872134	3	134,748,222	62	UPSTREAM		1.27E-08	0.031	7.25E-10
rs6778062	3	134,749,843	63	UPSTREAM		1.28E-08	0.024	7.84E-10
rs10433415	3	134,768,220	65	INTERGENIC	<i>CDV3</i>	2.13E-08	0.022	1.22E-09
rs4854733	3	134,769,755	64	INTERGENIC	<i>CDV3</i>	2.11E-08	0.022	1.22E-09
rs1444600	3	134,771,196	73	UPSTREAM	<i>CDV3</i>	6.62E-08	0.023	4.42E-09
rs10935070	3	134,824,678	59	NON_SYNONYMOUS_CODING	<i>TOPBP1</i>	1.10E-08	0.312	3.42E-09
rs9830001	3	134,916,160	51	INTRONIC	<i>TF</i>	6.31E-11	0.0026	8.41E-13
rs4525863	3	134,918,826	46	INTRONIC	<i>TF</i>	3.99E-12	5.80E-05	6.53E-15

SNP	Chromosome	Location, bp (Phasell/Rel#2, Feb09)	Rank	Type	Closest gene	P (meta-analysis, all 3 methods)	P (hetero- geneity)	P (meta-analysis, CE and Latex methods)
rs6782523	3	134,921,607	38	INTRONIC	TF	2.74E-12	4.94E-05	4.10E-15
rs10935073	3	134,921,874	37	INTRONIC	TF	2.72E-12	4.92E-05	4.06E-15
rs11921527	3	134,923,857	45	INTRONIC	TF	3.89E-12	2.32E-05	6.08E-15
rs12639304	3	134,924,284	42	INTRONIC	TF	3.81E-12	2.31E-05	5.95E-15
rs6774822	3	134,924,702	43	INTRONIC	TF	3.84E-12	1.62E-05	4.60E-15
rs6775042	3	134,924,935	44	INTRONIC	TF	3.85E-12	1.63E-05	4.62E-15
rs6439431	3	134,930,799	40	INTRONIC	TF	3.78E-12	1.61E-05	4.51E-15
rs7646118	3	134,932,729	41	INTRONIC	TF	3.78E-12	1.61E-05	4.51E-15
rs6787177	3	134,933,410	32	INTRONIC	TF	1.29E-12	4.88E-06	8.02E-16
rs8177178	3	134,945,962	47	INTRONIC	TF	4.02E-12	5.32E-05	5.86E-15
rs8177313	3	134,946,792	20	INTRONIC	TF	1.65E-13	5.80E-06	1.06E-16
rs4459901	3	134,948,389	39	INTRONIC	TF	3.18E-12	4.39E-05	4.76E-15
rs1800277	3	134,948,845	17	INTRONIC	TF	1.60E-14	1.08E-06	7.03E-18
rs6796795	3	134,948,912	16	INTRONIC	TF	1.69E-15	4.73E-07	4.30E-19
rs6785596	3	134,949,147	15	INTRONIC	TF	1.53E-15	4.50E-07	3.77E-19
rs8177191	3	134,950,829	11	INTRONIC	TF	1.76E-18	2.20E-04	1.32E-21
rs8177197	3	134,952,220	48	INTRONIC	TF	5.24E-12	1.47E-05	3.78E-15
rs8177213	3	134,954,917	85	INTRONIC	TF	2.92E-07	0.035	2.08E-08
rs8177220	3	134,956,315	14	INTRONIC	TF	1.33E-15	4.65E-07	4.00E-19
rs1799852	3	134,958,412	18	SYNONYMOUS_CODING	TF	6.28E-14	2.16E-06	5.30E-17
rs1799899	3	134,958,502	6	NON_SYNONYMOUS_CODING	TF	2.05E-28	1.77E-07	2.04E-33
rs8177240	3	134,960,391	25	INTRONIC	TF	3.12E-13	6.90E-07	1.39E-16
rs8177252	3	134,962,864	24	INTRONIC	TF	2.56E-13	6.63E-07	1.12E-16
rs8177272	3	134,965,560	23	INTRONIC	TF	2.13E-13	6.44E-07	9.15E-17
rs3811647	3	134,966,719	22	INTRONIC	TF	2.07E-13	6.39E-07	2.20E-06
rs1358024	3	134,966,878	81	INTRONIC	TF	1.09E-07	1.10E-04	8.85E-17
rs1525892	3	134,967,402	21	INTRONIC	TF	2.05E-13	6.39E-07	8.77E-17

SNP	Chromosome	Location, bp (Phasell/Rel#2, Feb09)	Rank	Type	Closest gene	P (meta-analysis, all 3 methods)	P (hetero- geneity)	P (meta-analysis, CE and Latex methods)
rs2692695	3	134,968,144	3	INTRONIC	<i>TF</i>	4.70E-29	6.12E-07	1.20E-33
rs1525889	3	134,972,723	28	INTRONIC	<i>TF</i>	8.21E-13	1.41E-06	5.83E-16
rs1049296	3	134,977,044	2	NON_SYNONYMOUS_CODING	<i>TF</i>	1.47E-31	1.96E-08	2.90E-37
rs7638018	3	134,978,151	31	INTRONIC	<i>TF</i>	1.16E-12	1.75E-06	9.09E-16
rs4854760	3	134,981,431	30	UPSTREAM	<i>TF</i>	1.14E-12	1.08E-06	7.19E-16
rs4854761	3	134,981,479	35	UPSTREAM	<i>TF</i>	2.00E-12	1.30E-06	1.39E-15
rs4854762	3	134,981,753	19	UPSTREAM	<i>TF</i>	1.07E-13	5.40E-06	9.41E-17
rs9843728	3	134,984,097	36	UPSTREAM	<i>SRPRB</i>	2.09E-12	4.81E-06	1.65E-15
rs1830084	3	134,991,154	33	INTRONIC	<i>SRPRB</i>	1.93E-12	3.73E-05	5.96E-15
rs9881405	3	135,000,888	26	INTRONIC	<i>SRPRB</i>	3.71E-13	0.0012	1.97E-15
rs6794945	3	135,001,153	27	INTRONIC	<i>SRPRB</i>	6.06E-13	6.39E-06	9.67E-16
rs9853615	3	135,002,671	4	INTRONIC	<i>SRPRB</i>	6.28E-29	1.17E-07	7.11E-34
rs12490148	3	135,006,663	34	INTRONIC	<i>SRPRB</i>	1.98E-12	1.95E-05	4.33E-15
rs1107413	3	135,007,407	29	NON_SYNONYMOUS_CODING	<i>SRPRB</i>	1.13E-12	7.50E-04	4.53E-15
rs3817524	3	135,009,093	5	INTRONIC	<i>SRPRB</i>	1.18E-28	7.54E-08	9.93E-34
rs7650925	3	135,009,859	49	INTRONIC	<i>SRPRB</i>	2.74E-11	0.0056	4.71E-13
rs6439441	3	135,012,284	50	INTRONIC	<i>SRPRB</i>	4.69E-11	0.0062	8.21E-13
rs1464937	3	135,019,345	7	INTRONIC	<i>SRPRB</i>	6.15E-26	1.10E-07	9.88E-31
rs1534165	3	135,019,607	82	INTRONIC	<i>SRPRB</i>	1.15E-07	0.0021	2.62E-09
rs1534166	3	135,019,757	1	INTRONIC	<i>SRPRB</i>	3.39E-39	2.64E-09	4.27E-46
rs6439442	3	135,023,049	84	NMD_TRANSCRIPT	<i>SRPRB</i>	2.62E-07	0.569	2.61E-07
rs17376019	3	135,023,573	8	NMD_TRANSCRIPT	<i>SRPRB</i>	8.05E-24	4.50E-04	8.41E-27
rs6439443	3	135,023,605	90	NMD_TRANSCRIPT	<i>SRPRB</i>	6.52E-07	0.433	3.70E-07
rs6773354	3	135,024,464	53	NMD_TRANSCRIPT	<i>SRPRB</i>	2.29E-09	0.030	1.14E-10
rs17310413	3	135,024,651	10	NMD_TRANSCRIPT	<i>SRPRB</i>	3.25E-20	0.0064	2.26E-22
rs12637730	3	135,028,987	57	3PRIME_UTR	<i>RAB6B</i>	7.32E-09	0.035	4.02E-10
rs2293374	3	135,029,935	58	3PRIME_UTR	<i>RAB6B</i>	1.03E-08	0.037	5.88E-10

SNP	Chromosome	Location, bp (PhaseIII/Rel#2, Feb09)	Rank	Type	Closest gene	P (meta-analysis, all 3 methods)	P (hetero- geneity)	P (meta-analysis, CE and Latex methods)
rs11921187	3	135,034,332	12	INTRONIC	<i>RAB6B</i>	5.22E-18	0.015	8.25E-20
rs6793673	3	135,035,962	9	INTRONIC	<i>RAB6B</i>	8.61E-21	0.0010	2.08E-23
rs2692672	3	135,038,190	60	INTRONIC	<i>RAB6B</i>	1.22E-08	0.504	6.47E-09
rs17310798	3	135,045,629	13	INTRONIC	<i>RAB6B</i>	1.98E-17	0.149	3.65E-18
rs1880663	3	135,064,149	56	INTRONIC	<i>RAB6B</i>	3.79E-09	0.659	5.83E-09

**Supplementary Figure 1.** Manhattan plots for associations with CDT percentage, total CDT concentration and total transferrin. For CDT% and total CDT the TF region showed heterogeneity between the CE, N-Latex vs CDText methods. For this region only CE and N-Latex cohorts were meta-analysed.

