

## ПРИЛОЖЕНИЕ

к статье В.В. Климонтова, К.С. Шишина, Р.А. Иванова, М.П. Пономаренко, К.А. Золотаревой, С.А. Лашина  
 «База данных о генах и белках, ассоциированных с нарушениями метаболизма глюкозы (GlucoGenes®):  
 описание и возможности применения в биоинформационических исследованиях»

Клинические SNP-маркеры заболеваний, которые аннотированы в базе данных ClinVar и статистически значимо изменяют средство TATA-связывающего белка (ТВР) к промоторам 52 исследованных генов человека, ассоциированных с нарушениями метаболизма глюкозы (НМГ)

Интернет-портал GlucoGenes® (эта работа)		База данных ClinVar (Landrum et al., 2014)				База данных Human_SNP_TATAbd (Филонов и др., 2023)								
№	Символ гена	Статус гликемии	Ассоциации с SNP		База данных dbSNP (Day, 2010)				$K_{D_r}$ нМ/л, <i>in silico</i>		Достоверность			
			Заболевание	dbSNP ID:min	5'-flank, 10 bp	WT → min	3'-flank, 10 bp	WT $M_0$	min $M_0$	$\pm$ SEM	$z$	$p$	$p$	$\Delta$
1	ABCA1	[High and Low Glucose] & [Low Glucose] & [High Glucose];	Hyperinsulinemic hypoglycemia; Hereditary hyperinsulinism; Maturity onset diabetes mellitus in young; Transitory neonatal diabetes mellitus; Transient Neonatal Diabetes; Dominant/Permanent neonatal diabetes mellitus; Hyperinsulinism; Dominant/Recessive Maturity onset diabetes mellitus in young; ABCC8-related disorder; Monogenic diabetes	rs886063317:C	agccgaatct	A→C	taaaaaggaac	2.23	0.22	9.87	0.9	21.9	$10^{-6}$	A
				rs886063317:G	agccgaatct	A→G	taaaaaggaac	2.23	0.22	7.05	0.66	16.69	$10^{-6}$	A
2	ABCC8	[High and Low Glucose] & [Low Glucose] & [High Glucose]	ABCC8-related disorder, hypoglycemia, hyperinsulinism, diabetes mellitus, monogenic diabetes	rs1409900082:A	agcactttcc	G→A	catcttggcc	74.04	5.71	30.75	2.85	14.58	$10^{-6}$	A
				rs1409900082:T	agcactttcc	G→T	catcttggcc	74.04	5.71	52.37	3.26	6.99	$10^{-6}$	A
				rs1955706366:A	cctgtccagt	G→A	cagagatccg	53.2	4.54	12.08	1.4	20.58	$10^{-6}$	A
				rs1957150758:A	cttggccgac	C→A	cttcgggagg	74.04	5.71	47.43	3.75	8.06	$10^{-6}$	A
				rs541244107:A	agctgcaagg	G→A	acagaggcgc	59.19	4.91	50.3	3.4	3.04	$10^{-2}$	C
				rs541244107:C	agctgcaagg	G→C	acagaggcgc	59.19	4.91	47.44	3.82	3.83	$10^{-3}$	B
				rs565662182:A	tgcgccccc	G→A	agccccacacc	53.2	4.54	31.21	2.78	8.65	$10^{-6}$	A
				rs58241708:T	tgcagagatc	C→T	gtgaggagca	53.2	4.54	45.33	3.8	2.68	$10^{-2}$	C
3	ADAMTS13	[High and Low Glucose]	Abnormal bleeding; Thrombocytopenia Upshaw-Schulman syndrome	rs782563004:G	tccagttatg	T→G	cctgtcctcc	7.54	0.75	10.29	0.98	4.5	$10^{-3}$	B
4	ADGRV1	[High Glucose]	Usher syndrome type 2C	rs587712950:C	acccttagcca	T→C	ggcccccgtcc	103.92	6.75	57.35	5.28	10.55	$10^{-6}$	A
5	AGTR1	[High Glucose]	Renotubular dysgenesis	rs529360494:G	cgcgcctcac	T→G	ataaaattcgg	1.87	0.19	6.42	0.58	18.28	$10^{-6}$	A
6	APOA1	[High Glucose]	Adenylosuccinate lyase deficiency	rs909669:T	agattctccg	C→T	agccggcagc	69.13	5.55	55.06	6.65	3.14	$10^{-2}$	C
7	APOB	[High and Low Glucose]	Familial hypobetalipoproteinemia 1; Hypercholesterolemia, autosomal dominant, type B	rs1266180253:A	ctgagtgccc	T→A	tctcggttgc	71.83	4.85	63.65	4.82	2.38	0.05	D
8	APP	[High Glucose]	Early-onset autosomal dominant Alzheimer disease	rs200621906:A	gggggtggcc	G→A	gatcagctga	89.97	7.19	55.96	3.68	9.17	$10^{-6}$	A
				rs459543:T	gcacgcggag	G→T	agcgtgcgcg	120.77	10.01	52.72	4.74	13.55	$10^{-6}$	A
				rs459543:A	gcacgcggag	G→A	agcgtgcgcg	120.77	10.01	93.07	6.72	4.74	$10^{-3}$	B
9	CASP8	[High Glucose]	Autoimmune lymphoproliferative syndrome type 2B	rs886056998:T	cctcggcagc	G→T	gtaggcgaga	120.77	10.01	101.55	8.7	2.91	$10^{-2}$	C

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№	Символ гена	Статус гликемии	Ассоциации с SNP			База данных dbSNP (Day, 2010)			$K_D$ , нМ/л, <i>in silico</i>			Достоверность			
			Заболевание	dbSNP ID:min	5'-flank, 10 bp	WT→min	3'-flank, 10 bp	WT $M_0$	min $M_0$	$\pm$ SEM	$z$	$p$	$\rho$	$\Delta$	
10	<i>CDH3</i>	[High Glucose]	EEM syndrome, i.e., Ectodermal dysplasia, Ectrodactyly and Macular dystrophy syndrome	rs537339409:T	GACCTCGTGG	C→T	GCTGGACCAA	69.56	3.98	63.44	4.54	2.01	0.05	D	
				rs74674351:T	GTCGACTTCA	G→T	ATGTGTCTCC	9.19	0.83	8.05	0.67	2.15	0.05	D ↑	
				rs886052227:T	ACCTCGTGGC	G→T	CTGGACCAAT	69.56	3.98	63.79	4.00	2.04	0.05	D	
11	<i>CDKN1B</i>	[High and Low Glucose] & [High Glucose]	Multiple endocrine neoplasia type 4; Hereditary cancer-predisposing syndrome	rs1555085477:A	tgtcaaacgt	G→A	cgagtgtcta	22.24	1.44	16.02	1.56	5.62	$10^{-6}$	A ↑	
				rs756190836:A	ggagaaaagat	G→A	tcaaaacgtgc	22.24	1.44	11.01	0.91	13.36	$10^{-6}$	A ↑	
12	<i>COL3A1</i>	[High Glucose]	Ehlers-Danlos syndrome type 4; Familial thoracic aortic aneurysm and aortic dissection	rs763316592:G	ccatatttca	A→G	ttttactctg	8.28	0.76	6.88	0.72	2.68	$10^{-2}$	C ↑	
				rs766564882:T	ttcaatttta	C→T	tctgtagggt	8.28	0.76	5.32	0.47	6.98	$10^{-6}$	A ↑	
13	<i>DCN</i>	[High Glucose]	Congenital stromal corneal dystrophy	rs189853358:G	ttaccatatg	A→G	tctaatgtgg	5.11	0.52	6.64	0.53	4.02	$10^{-3}$	B ↓	
14	<i>DNMT1</i>	[Low Glucose] & [High Glucose]	Hereditary sensory neuropathy-deafness-dementia syndrome; Inborn genetic diseases	rs149362098:A	agtgaagccc	G→A	tagagtggga	30.01	2.3	18.9	1.94	7.23	$10^{-6}$	A ↑	
				rs554894511:A	aaccaagcaa	G→A	aagtgaagcc	30.01	2.3	22.51	1.47	5.72	$10^{-6}$	A ↑	
15	<i>EIF2AK3</i>	[Low Glucose]	Wolcott-Rallison dysplasia	rs200991366:C	tgaatcttct	T→C	ataaaatttgg	2.62	0.32	6.49	0.64	11.69	$10^{-6}$	A ↓	
				rs766524866:A	gccagcttat	G→A	ccagacaca	20.18	1.85	6.2	0.71	16.15	$10^{-6}$	A ↑	
16	<i>FBN1</i>	[High Glucose]	Stiff skin syndrome; Familial thoracic aortic aneurysm and aortic dissection; Weill-Marchesani syndrome; Ectopia lentis 1, isolated, autosomal dominant; Marfan syndrome; Acromicric dysplasia; Geleophysic dysplasia	rs1890299170	tcttttttta	A→T	aaaaagtatt	5.43	0.48	1.97	0.19	15.52	$10^{-6}$	A	
				rs3825792	aaaagtattt	C→G	tctcgcgaga	9.94	0.9	7.41	0.7	4.52	$10^{-3}$	B ↑	
17	<i>FBP1</i>	[Low Glucose]	Fructose-biphosphatase deficiency	rs921192380:T	gccgaccgc	G→T	tctaaagggt	19.48	1.62	12.54	0.99	7.68	$10^{-6}$	A ↑	
18	<i>FTO</i>	[High and Low Glucose]	Lethal polymalformative syndrome, Boissel type; Inborn genetic diseases	rs150450891:A	agcataacaac	G→A	taaccttgct	7.17	0.73	8.48	0.8	2.42	0.05	D	
				rs375930208:A	atcattttctt	G→A	tagacaaatg	6.38	0.45	3.57	0.35	9.6	$10^{-6}$	A	
				rs527438785:A	attttagcaca	G→A	gagaatttcc	20.2	1.63	15.89	1.58	3.74	$10^{-3}$	B ↑	
				rs886052100:C	tgcattcctgg	G→C	agttgttagtt	8.7	0.86	6.79	0.67	3.56	$10^{-3}$	B	
19	<i>G6PC3</i>	[High and Low Glucose]	Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency	rs28370440:T	caggaggaaa	C→T	agtaccggct	33.75	2.38	14.85	1.15	15.68	$10^{-6}$	A ↑	
				rs746461072:A	cctggactct	G→A	gtttccgccc	50.87	3.79	26.58	2.12	11.88	$10^{-6}$	A ↑	
20	<i>GFAP</i>	[High and Low Glucose]	Scoliosis; Alexander disease	rs1057518828:C	ttgcaggagt	A→C	ccagggacctg	25.7	2.7	39.32	3.12	6.46	$10^{-6}$	A ↓	
				rs797044574:C	ctggaacagc	A→C	aaacaaggcg	29.97	2.11	40.47	2.77	6.11	$10^{-6}$	A ↓	
21	<i>GLUD1</i>	[High Glucose]	Hyperinsulinism, dominant	rs886047380:G	ctgcggctta	A→G	aaggcaacc	9.87	0.82	23.31	2.17	13.79	$10^{-6}$	A ↓	

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			Заболевание	dbSNP ID:min	5'-flank, 10 bp	WT→min	3'-flank, 10 bp	WT $M_0$	min $M_0$	$\pm$ SEM	$\pm$ SEM	z	p	$\rho$
22	<i>HSPG2</i> [High Glucose]	Schwartz-Jampel syndrome; Lethal Kniest-like syndrome		rs140197256:T	ccaccatata	G→T	caccacggtc	12.63	1.17	9.14	0.81	5.05	$10^{-6}$	A
				rs17459139:A	tagtgcattt	G→A	cagctggatg	23.53	1.97	7.36	0.7	18.29	$10^{-6}$	A ↑
				rs761025182:T	gccctcagt	G→T	tagtgcattt	23.53	1.97	11.83	1.29	10	$10^{-6}$	A
				rs761025182:G	gccctcagt	C→G	tagtgcattt	23.53	1.97	20.53	1.99	2.13	0.05	D
23	<i>IL1B</i>	[High and Low Glucose] & [Low Glucose] & [High Glucose]	Gastric cancer susceptibility after H, pylori infection	rs1143627:T	ttttgaaagc	C→T	ataaaaacag	4.5	0.39	1.76	0.17	14.56	$10^{-6}$	A ↑
24	<i>INS</i>	[High and Low Glucose] & [Low Glucose] & [High Glucose]	Neonatal diabetes mellitus; Autosomal recessive DOPA responsive dystonia; Maturity onset diabetes mellitus in young; Transient Neonatal Diabetes, dominant/recessive; Maturity-onset diabetes of the young type 10	rs5505:T	agatca	T→C	tttctgccat	53.29	3.56	43.36	3.16	4.17	$10^{-3}$	B ↑
25	<i>INSR</i>	[High and Low Glucose] & [High Glucose]	Insulin-resistant diabetes mellitus & acanthosis nigricans; Rabson-Mendenhall syndrome, leprechaunism syndrome	rs532052290:T	cgcctcgagg	C→T	atgaccccccg	62.14	4.83	17.87	1.92	18.81	$10^{-6}$	A ↑
26	<i>KRIT1</i> [High and Low Glucose]	Angiokeratoma corporis diffusum with arteriovenous fistulas; Cerebral cavernous malformation		rs370781912:C	ggaagctctt	T→C	caccgccaac	54.24	4.15	65.33	5.02	3.43	$10^{-3}$	B ↓
				rs1437344467:A	tgcagg	T→A	tgaaaaggag	32.96	2.33	19.6	1.33	10.63	$10^{-6}$	A
				rs553392669:T	gactgcagg	C→T	cagtggaaag	32.96	2.33	29.32	1.84	2.48	0.05	D
				rs58075588:T	ccacccttagt	C→T	tccttagtcgc	28.89	2.57	19.7	1.77	6.06	$10^{-6}$	A ↑
				rs886062497:A	ttcatagcca	C→A	ggtaacggcg	11.23	1.02	9.44	0.87	2.67	$10^{-2}$	C
				rs932427766:A	ccagtgaaaa	G→A	gagcaggaac	32.96	2.33	26.37	1.92	4.4	$10^{-3}$	B
27	<i>MAPT</i> [High Glucose]	MAPT-related spectrum disorders		rs886053025:C	cctcaggaa	G→C	cgcctcttc	77.48	5.36	92.87	7.45	3.42	$10^{-3}$	B ↓
				rs550530601:A	cgcctcttc	C→A	ctggggaggc	210.23	14.64	136	11.37	8.01	$10^{-6}$	A ↑
				rs886053025:T	cctcaggaa	G→T	cgcctcttc	77.48	5.36	66.22	5.19	3.01	$10^{-2}$	C
28	<i>MEF2C</i>	[High Glucose]	Intellectual disability, autosomal dominant; Inborn genetic diseases	rs1554102556:C	ttttgttaac	A→C	ggaaacgggt	10.89	1.09	20.48	1.31	10.64	$10^{-6}$	A ↓

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№	Символ гена	Статус гликемии	Ассоциации с SNP			База данных dbSNP (Day, 2010)			$K_D$ , нМ, <i>in silico</i>			Достоверность		
			Заболевание	dbSNP ID:min	5'-flank, 10 bp	WT→min	3'-flank, 10 bp	WT $M_0$	min $M_0$	$\pm$ SEM	$z$	$p$	$\rho$	$\Delta$
29	MEN1	[High Glucose]	Multiple endocrine neoplasia, type 1; Hereditary cancer-predisposing syndrome	rs1035192873:G	gcccgccact	A→G	tttccaggct	17.12	1.38	50.35	3.72	19.73	$10^{-6}$	A
				rs1060499980:G	ccagacagtgc	A→G	atgccggtgt	31.88	2.66	61.32	4.47	11.81	$10^{-6}$	A
				rs1438685841:G	cagacagtca	A→G	tgcgggtgtgg	31.88	2.66	45.52	3.43	6.34	$10^{-6}$	A
				rs1565647698:G	ggccagacag	T→G	caatccgggt	31.88	2.66	61.32	4.47	11.81	$10^{-6}$	A
				rs1592650685:C	ggggccagac	A→C	gtcaatgccg	31.88	2.66	39.7	3.15	3.81	$10^{-3}$	B
				rs756287855:G	aggggccaga	C→G	agtcaatgcc	31.88	2.66	38.21	3.2	3.06	$10^{-2}$	C
				rs1114167479:T	cctggcacgg	C→T	aaggcAACG	31.88	2.66	25.34	2.28	3.75	$10^{-3}$	B
				rs1565647751:C	gcaggggcca	G→C	acagtcaatg	31.88	2.66	23.95	1.84	5.04	$10^{-6}$	A
				rs1565647751:T	gcaggggcca	G→T	acagtcaatg	31.88	2.66	13.85	1.68	11.34	$10^{-6}$	A
				rs1565647767:T	ccgcaggggc	C→T	agacagtcaa	31.88	2.66	25.43	1.98	3.97	$10^{-3}$	B
30	NLRP3	[High and Low Glucose] & [High Glucose]	Chronic infantile neurological, cutaneous and articular syndrome; Familial amyloid nephropathy with urticaria AND deafness; Familial cold autoinflammatory syndrome 1	rs138900557:A	tcaaagtgtc	G→A	ggattacagg	16.3	1.37	13.02	1.08	3.81	$10^{-3}$	B
				rs140404523:T	gagaaagcca	G→T	acagacgcag	31.27	2.49	13.02	1.31	13.67	$10^{-6}$	A
31	NPHS1	[High Glucose]	Congenital nephrotic syndrome; Finnish congenital nephrotic syndrome	rs73928331:T	cagagacaca	C→T	agagagggac	31.27	2.49	12.97	1.36	13.34	$10^{-6}$	A
32	PARK7	[Low Glucose]	Parkinson Disease, recessive	rs17523802:T	ggggctgagg	G→T	aggccggacg	61.68	4.4	47.85	5.56	3.72	$10^{-3}$	B
33	PAX3	[High Glucose]	Waardenburg syndrome; Craniofacial-deafness-hand syndrome	rs45501095:T	cacaggagga	G→T	actcaggcag	42.17	2.9	28.27	2.81	6.62	$10^{-6}$	A
				rs867534042:A	gacttaggaac	C→A	gacagcccccc	36.02	3.21	28.35	2.4	3.89	$10^{-3}$	B
34	PAX4	[High Glucose]	Maturity onset diabetes mellitus in young	rs867534042:T	gacttaggaac	C→T	gacagcccccc	36.02	3.21	31.47	2.61	2.22	0.05	D
35	PAX6	[High Glucose]	carboxymethyl-dextran-A2-gadolinium-DOTA; Aniridia; Cerebellar Ataxia And Intellectual Disability; Autosomal dominant keratitis; Anophthalmia-microphthalmia syndrome; Foveal hypoplasia 111p partial monosomy syndrome; Congenital aniridia	rs886048209:C	gcggagtgtat		agtgggttg	16.89	1.13	19.65	1.3	3.22	$10^{-2}$	C
				rs199928376:T	tgccgccaac	G→T	gggttaaggga	46.52	3.87	36.68	3.28	3.89	$10^{-3}$	B
36	PGM1	[Low Glucose]	PGM1-congenital disorder of glycosylation, Inborn genetic diseases											

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			Заболевание	dbSNP ID:min	5'-flank, 10 bp	WT→min	3'-flank, 10 bp	WT	min	$M_0$	$\pm$ SEM	$M_0$	$\pm$ SEM	z	p	$\rho$	$\Delta$
37	PTEN	[High Glucose]	PTEN hamartoma tumor syndrome; Hereditary breast ovarian cancer syndrome; Hereditary cancer-predisposing syndrome; Cowden syndrome 1	rs369849061:G	agcagccatt	A→G	cccggtcgcg	38.64	3.63	73.43	6.44	9.99	10 <sup>-6</sup>	A			
				rs535471450:G	ccgcacgacg	C→G	attaccggc	38.64	3.63	73.43	6.44	9.99	10 <sup>-6</sup>	A	↓		
				rs587782793:C	ctttatgcgc	T→C	gcggcaggat	7.97	0.7	9.01	0.8	1.96	0.05	D			
				rs144620057:A	cgcctgtccc	C→A	cttcattaccgc	45.97	4.29	27.75	2.77	7.39	10 <sup>-6</sup>	A			
				rs1589593824:T	cccccttcata	C→T	cgcgcctgtc	45.97	4.29	35.03	3.27	4.12	10 <sup>-3</sup>	B			
				rs564216039:T	cccagagacta	C→T	cgcctgtccc	45.97	4.29	34.92	3.54	3.99	10 <sup>-3</sup>	B			
				rs587779989:A	cagcgcctgt	G→A	agcagccgcg	83.24	6.54	23.25	2.81	17.71	10 <sup>-6</sup>	A			
				rs587781501:T	tctgccccct	C→T	ctaccgcggc	45.97	4.29	21.51	2.08	11.31	10 <sup>-6</sup>	A			
				rs587781871:T	tgcgcgtcgca	C→T	ccagagctac	45.97	4.29	37.02	3	3.5	10 <sup>-3</sup>	B	↑		
				rs587781981:T	tggcgggact	C→T	tttatgcgc	7.97	0.7	5.26	0.47	6.65	10 <sup>-6</sup>	A			
				rs587782079:C	ctcgcaccca	G→C	agctaccgc	45.97	4.29	31.27	2.88	5.88	10 <sup>-6</sup>	A			
				rs587782230:T	gcgctcgcac	C→T	cagagctacc	45.97	4.29	24.09	1.86	10.66	10 <sup>-6</sup>	A			
				rs587782230:A	gcgctcgcac	C→A	cagagctacc	45.97	4.29	25.24	2.01	9.77	10 <sup>-6</sup>	A			
				rs786204920:T	gcggcttggg	G→T	actctgcgc	45.97	4.29	30.29	3.14	5.98	10 <sup>-6</sup>	A			
38	RUNX2	[High and Low Glucose]	Cleidocranial dysostosis	rs886061492:T	gtatttacaa	C→T	agagggtaca	4.65	0.48	3.84	0.41	2.58	10 <sup>-2</sup>	C	↑		
39	RYR2	[High Glucose]	Catecholaminergic polymorphic ventricular tachycardia 1	rs373127797:C	gaaaccactt	T→C	atttattatag	2.99	0.26	3.47	0.26	2.56	0.05	D	↓		
40	SERPINF1	[High Glucose]	Osteogenesis Imperfecta, recessive	rs541151948:T	gagtgcaggt	C→T	gctttaagaa	10.24	0.88	8.51	0.77	2.97	10 <sup>-2</sup>	C	↑		
41	SLC16A1	[Low Glucose]	Exercise-induced hyperinsulinism	rs886045087:T	cctgcgccac	C→T	acgtcacgca	61.19	4.18	35.72	2.89	10.18	10 <sup>-6</sup>	A	↑		
42	SLC2A1	[High and Low Glucose] & [Low Glucose] & [High Glucose]	Dystonic disorder; GLUT1 deficiency syndrome	rs561646847:T	gtcctataaa	C→T	gctacggtcc	4.31	0.56	2.51	0.25	6.57	10 <sup>-6</sup>	A	↑		
					atctctgttag	G→T	aagaggcggt	6.31	0.86	4.45	0.47	4.07	10 <sup>-3</sup>	B	↑		
43	SMAD3		Familial thoracic aortic aneurysm and aortic dissection	rs1555413991:T	tctactacat	C→T	ggagggggagg	6.31	0.86	4.85	0.85	2.38	0.05	D	↑		
					rs750779804:C	A→C	gcaagtgtga	20.15	1.69	34.31	3.25	8.41	10 <sup>-6</sup>	A	↓		
44	SMPD1	[High Glucose]	Niemann-Pick disease, types A and B; Sphingomyelin/cholesterol lipidosis	rs1423504237:T	gcgaatacatag	C→T	aagtgtgacc	20.15	1.69	7.52	0.64	16.56	10 <sup>-6</sup>	A			
					gcaatacag	C→A	aagtgtgacc	20.15	1.69	13.83	1.18	6.29	10 <sup>-6</sup>	A			
					atactggggc	G→T	aatacagcaa	20.15	1.69	6.5	0.9	13.96	10 <sup>-6</sup>	A	↑		
					atactggggc	G→A	aatacagcaa	20.15	1.69	11.98	1.54	6.77	10 <sup>-6</sup>	A			
					atactggggc	G→C	aatacagcaa	20.15	1.69	16.43	2.06	2.71	10 <sup>-2</sup>	C			
					atactggggc	G→C	aatacagcaa	20.15	1.69								

**Продолжение приложения**

Интернет-портал GlucoGenes® (эта работа)			База данных ClinVar (Landrum et al., 2014)						База данных Human_SNP_TATAbd (Филонов и др., 2023)						
№	Символ гена	Статус гликемии	Ассоциации с SNP			База данных dbSNP (Day, 2010)			$K_D$ , нМ/л, <i>in silico</i>			Достоверность			
			Заболевание	dbSNP ID:min	5'-flank, 10 bp	WT→min	3'-flank, 10 bp	WT $M_0$	min $M_0$	$\pm$ SEM	$\pm$ SEM	z	p	$\rho$	
45	SNCA	[High Glucose]	Parkinson Disease, dominant	rs757451373:A	ggacggcgac	G→A	accagaaggg	43.79	3.04	36.74	2.28	3.77	$10^{-3}$	B	↑
46	SOD1	[High and Low Glucose] & [High Glucose]	Amyotrophic lateral sclerosis type 1	rs7277748:G	ggtctggcct	A→G	taaagttagtc	2.44	0.23	6.97	0.59	16.67	$10^{-6}$	A	↓
47	STAT3	[High Glucose]	Hyper-IgE recurrent infection syndrome 1, autosomal dominant	rs780393027:A rs902564848:T	ggagccgggg gcgcagactg	G→A G→T	ttccgacgtc gagggggagc	105.37 105.37	7.43 7.43	72.66 37.34	4.84 2.77	7.66 20.27	$10^{-6}$	A	↑
48	STK11	[High Glucose]	Peutz-Jeghers syndrome; Hereditary cancer-predisposing syndrome	rs2080796950:C	tgtggtgccg	T→C	acttggagga	17.1	1.96	26.24	2.08	6.14	$10^{-6}$	A	
				rs773049570:C	ctgtggtgcc	G→C	tacttggagg	17.1	1.96	25.1	2.82	4.78	$10^{-3}$	B	↓
				rs876658160:C	gccgtacttg	G→C	aggacctgca	17.1	1.96	26.24	2.08	6.14	$10^{-6}$	A	
				rs1057521267:T	ggtggcgcag	C→T	atgactgtgg	17.1	1.96	8.58	0.86	9.04	$10^{-6}$	A	
				rs1323638467:T	ttttttctt	G→T	taaaaattttg	8.04	0.82	5.9	0.47	4.8	$10^{-3}$	B	
				rs1407068597:A	tttctttttt	C→A	tttgtaaaaat	8.04	0.82	4.39	0.29	9.94	$10^{-6}$	A	
				rs1407068597:T	tttctttttt	C→T	tttgtaaaaat	8.04	0.82	6.29	0.52	3.73	$10^{-3}$	B	
				rs1555739243:T	ggcgcagcat	G→T	actgtggtgc	17.1	1.96	10.28	0.77	7.43	$10^{-6}$	A	↑
				rs1555739243:A	ggcgcagcat	G→A	actgtggtgc	17.1	1.96	6.8	0.61	12.65	$10^{-6}$	A	
				rs187744790:A	tcttcgacat	C→A	gaggatgaca	17.1	1.96	12.01	1.1	4.82	$10^{-3}$	B	
49	TP53	[High and Low Glucose] & [High Glucose]	Li-Fraumeni syndrome; Hereditary cancer-predisposing syndrome	rs2080662705:A	acgtttgcgg	G→A	gaggggggcg	59.52	5.44	51.84	4.76	2.13	0.05	D	
				rs769644352:T	gactgtggt	C→T	cgtacttgg	17.1	1.96	13.96	1.45	2.62	$10^{-2}$	C	
				rs786202471:T	tggtgcccgt	C→T	ttggaggacc	17.1	1.96	12.34	1.27	4.23	$10^{-3}$	B	
				rs1457582183:T	TCTTTCCCTAG	C→T	actgccccaa	7.46	0.63	6.04	0.61	3.22	$10^{-2}$	C	
				rs34361146:A	TTGCCTCTTT	C→A	ctagcactgc	7.46	0.63	4.96	0.51	6.17	$10^{-6}$	A	↑
50	VHL	[High and Low Glucose] & [Low Glucose]	Squamous cell carcinoma of the head and neck	rs1597400604:A	CAGTCAGGAG	C→A	TTACCCAATC	47.8	4.43	34.32	2.52	5.61	$10^{-6}$	A	
				rs1034934219:T	cgggagcgcg	C→T	acgcagctcc	85.18	7.05	41.72	4.85	10	$10^{-6}$	A	
				rs1192379474:A	tggtctggat	C→A	gcggagggaa	85.18	7.05	40.3	4.07	11.47	$10^{-6}$	A	
				rs1192379474:G	tggtctggat	C→G	gcggagggaa	85.18	7.05	66.48	5.34	4.3	$10^{-3}$	B	↑
				rs1696106670:T	gttccatcct	C→T	taccgagcgc	31.82	2.87	24.85	2.55	3.61	$10^{-3}$	B	
				rs886057700:A	cgagcgcgtt	C→A	catcctctac	31.82	2.87	9.42	0.88	18.71	$10^{-6}$	A	
				rs886057701:A	cccgccggcgt	C→A	cggccccgggt	85.18	7.05	67.81	7.47	3.31	$10^{-3}$	B	

## Продолжение приложения

Интернет-портал GlucoGenes® (эта работа)		База данных ClinVar (Landrum et al., 2014)					База данных Human_SNP_TATAdb (Филонов и др., 2023)										
№	Символ гена	Статус гликемии	Ассоциации с SNP			База данных dbSNP (Day, 2010)			$K_D$ , нМ/л, <i>in silico</i>			Достоверность					
			Заболевание	dbSNP ID:min	5'-flank, 10 bp	WT→min	3'-flank, 10 bp	WT	min	$M_0$	$\pm SEM$	$M_0$	$\pm SEM$	z	p	$p$	Δ
51	TSC2	[High Glucose]	Tuberous sclerosis 2; Hereditary cancer-predisposing syndrome	rs1449727037:C	ctccctccctg	T→C	accagtcacag	21.96	2.54	39.18	2.93	8.41	10 <sup>-6</sup>	A			
				rs1555496967:C	cggtcgcgga	T→C	ctgttgcagc	48.4	3.9	59.07	4.25	3.69	10 <sup>-3</sup>	B			
				rs1555496967:A	cggtcgcgga	T→A	ctgttgcagc	48.4	3.9	56.45	3.96	2.88	10 <sup>-2</sup>	C			
				rs1555512861:G	cctccctgtta	C→G	cagtccagot	21.96	2.54	27.41	2.86	2.85	10 <sup>-2</sup>	C			
				rs1596404506:C	tcccgttacc	A→C	gtcccgactgc	21.96	2.54	28.17	2.92	3.21	10 <sup>-2</sup>	C			
				rs2090259818:G	ctctttctcc	T→G	ccctgttacca	21.96	2.54	34.58	5.51	4.61	10 <sup>-3</sup>	B			
				rs45493394:G	tgatotcccta	T→G	agagcgcag	8.19	1.04	36.94	3.54	18.98	10 <sup>-6</sup>	A	↓		
				rs45493394:C	tgatotcccta	T→C	agagcgcag	8.19	1.04	22.44	2.44	12.09	10 <sup>-6</sup>	A			
				rs45493394:A	tgatotcccta	T→A	agagcgcag	8.19	1.04	25.37	1.97	15.23	10 <sup>-6</sup>	A			
				rs761713908:T	tggcccttctt	C→T	tcctccctgt	21.96	2.54	27.26	4.05	2.3	0.05	D			
				rs766029525:C	tctccctccct	G→C	taccagtccaa	21.96	2.54	39.18	2.93	8.41	10 <sup>-6</sup>	A			
				rs774727624:C	ctgatctccct	A→C	tagagcgcag	8.19	1.04	25.53	2.88	13.41	10 <sup>-6</sup>	A			
				rs1190158836:A	gaaggcggtc	G→A	cggatctgtt	48.4	3.9	41.44	3.53	2.65	10 <sup>-2</sup>	C			
				rs1325874616:T	acctgatctc	C→T	tatagagcgc	8.19	1.04	4.47	0.48	7.3	10 <sup>-6</sup>	A			
				rs1325874616:G	acctgatctc	C→G	tatagagcgc	8.19	1.04	5.87	0.79	3.62	10 <sup>-3</sup>	B			
				rs1330915229:T	tctttctctt	C→T	cctgttaccag	21.96	2.54	15.45	1.5	4.65	10 <sup>-3</sup>	B			
				rs1369594860:A	aaggcggtcg	C→A	gatctgttg	48.4	3.9	41.36	3.57	2.66	10 <sup>-2</sup>	C			
				rs1410163756:A	gccttccctt	G→A	cagtggcctc	21.96	2.54	12.06	1.23	7.78	10 <sup>-6</sup>	A			
				rs142580483:T	gcagtgaaag	C→T	actctggaaag	48.4	3.9	22.17	1.95	13.09	10 <sup>-6</sup>	A			
				rs1466279830:A	tgcgcgatct	G→A	ttgcagccgg	48.4	3.9	18.06	1.76	15.59	10 <sup>-6</sup>	A			
				rs1567394860:A	aagcaactctg	G→A	aaggcggtcg	48.4	3.9	25.59	1.86	11.73	10 <sup>-6</sup>	A			
				rs202187148:T	tcctataagag	C→T	gcagtcacatc	8.19	1.04	5.92	0.69	3.78	10 <sup>-3</sup>	B			
				rs2085359439:T	cacgcagttgg	A→T	agcaactctgg	48.4	3.9	37.1	2.75	4.86	10 <sup>-3</sup>	B			
				rs2085359793:A	cgcagttggaa	G→A	cactctggaa	48.4	3.9	36.7	3.48	4.44	10 <sup>-3</sup>	B	↑		
				rs2086770532:C	atctccata	G→C	agcgcagtc	8.19	1.04	6.1	0.93	2.98	10 <sup>-2</sup>	C			
				rs374146919:T	tttctcttcc	C→T	tgttaccatc	21.96	2.54	9.79	1	10.49	10 <sup>-6</sup>	A			
				rs397514912:A	aggcggtcgc	G→A	gatctgttgc	48.4	3.9	35.31	3.07	5.32	10 <sup>-6</sup>	A			
				rs397514933:A	gaagcaactct	G→A	gaaggcggtc	48.4	3.9	16.83	1.61	16.87	10 <sup>-6</sup>	A			
				rs45485999:T	ggtcgcggat	C→T	tgttgcagcc	48.4	3.9	20.94	1.54	15.37	10 <sup>-6</sup>	A			
				rs45485999:G	ggtcgcggat	C→G	tgttgcagcc	48.4	3.9	28.17	2.53	8.97	10 <sup>-6</sup>	A			
				rs45517099:T	ggatctgttg	C→T	agccggagcg	48.4	3.9	12.83	1.17	21.86	10 <sup>-6</sup>	A			
				rs543752709:A	ggtgcgcctt	T→A	ctccgcgtcg	47.51	3.28	29.99	2.39	8.73	10 <sup>-6</sup>	A			
				rs761713908:A	tggcccttctt	C→A	tcctccctgt	21.96	2.54	8.51	0.74	13.08	10 <sup>-6</sup>	A			
				rs766029525:A	tctccctccct	G→A	taccagtccaa	21.96	2.54	9.39	1.61	8.23	10 <sup>-6</sup>	A			
				rs770054036:T	cttcccttgc	C→T	agtggcctct	21.96	2.54	10.04	1.02	10.17	10 <sup>-6</sup>	A			

Примечание. WT и min – анцестральный (норма) и минорный (патология) аллели SNP;  $K_D$  – равновесная константа диссоциации комплекса ТВР-промотор, выраженная в наномолях на литр, (нМ/л);  $M_0$  и SEM – контекстно-зависимая оценка *in silico* и ее стандартизированная ошибка; z, p и ρ – значение z-статистики Фишера и уровень ее значимости, а также эвристическая приоритизация оценок *in silico* от наилучшей (A) до наихудшей (D) в алфавитном порядке; Δ – экспрессия выше (↑) или ниже (↓).

## Окончание приложения

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