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Supplementary Material: *Reproduction, Fertility and Development*

Supplementary Material

Spermatogenic phenotype of testis-specific protein, Y-encoded, 1 (*TSPY1*) dosage deficiency is independent of variations in TSPY-like 1 (*TSPYL1*) and TSPY-like 5 (*TSPYL5*): a case-control study in a Han Chinese population

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Table S1. Testing for Hardy-Weinberg equilibrium of the *TSPYL1* and *TSPYL5* variations in the groups of patient and control

SNP	Genotype	Genotype frequency		Hardy-Weinberg equilibrium testing ($\alpha = 0.05$)	
		Patients, n (%)	Controls, n (%)	<i>P</i> -values*	<i>P</i> -values#
rs3828743	AA	98 (15.0)	97 (13.0)	0.079	0.6738
	AG	283 (43.2)	337 (45.1)		
	GG	274 (41.8)	313 (41.9)		
rs1128261	TC	74 (11.3)	75 (10.0)	0.1255	0.1486
	CC	581 (88.7)	672 (90.0)		
rs6468544	CG	33 (5.0)	26 (3.5)	0.5084	0.6283
	CC	622 (95.0)	721 (96.5)		
rs2583501	CC	7 (1.1)	2 (0.3)	0.2918	0.2226

	CT	99 (15.1)	110 (14.7)		
	TT	549 (83.8)	635 (85.0)		
rs2289496	GG	75 (11.5)	103 (13.8)	0.3843	0.3911
	GT	308 (47.0)	334 (44.7)		
	TT	272 (41.5)	310 (41.5)		
rs2583506	TT	9 (1.3)	3 (0.4)	0.4436	0.1251
	CT	119 (18.2)	134 (17.9)		
	CC	527 (80.5)	610 (81.7)		
rs1835740	GG	132 (20.2)	129 (17.3)	0.9389	0.6995
	AG	323 (49.3)	369 (49.4)		
	AA	200 (30.5)	249 (33.3)		

rs3763558	AA	8 (1.2)	2 (0.2)	0.0586	0.4301
	AC	91 (13.9)	96 (12.9)		
	CC	556 (84.9)	649 (86.9)		

* Hardy-Weinberg equilibrium test of patient group.

Hardy-Weinberg equilibrium test of control group.

Table S2. Comparison of the haplotype frequency and distribution of the *TSPYL5* between patients with spermatogenic failure and controls

Haplotype		Frequencies of the haplotypes			
rs2583501	rs2583506	Patients, <i>n</i> (%)	Controls, <i>n</i> (%)	OR (95 % CI)*	<i>P</i> -values ($\alpha = 0.05$)#
C	T	113 (8.6)	112 (7.5)	1.165 (0.887, 1.530)	0.296
T	C	1169 (89.2)	1355 (90.7)	0.850 (0.664, 1.089)	0.207
Others		28 (2.1)	27 (1.8)	1.187 (0.696, 2.024)	0.586
<i>P</i> -values ($\alpha = 0.05$)^			0.436		

Fisher exact test for the difference in the frequency of single haplotype between patients and controls.

^ Chi-squared test for the difference in haplotype distribution between patients and controls.

Table S3. Comparisons of sperm productions among different genotypes of the SNPs in the whole study population

SC, sperm concentration; TSC, total sperm count

SNP	Genotype	<i>n</i>	Median of SC (25th–75th) ($n \times 10^6/\text{ml}$)	Median of TSC (25th–75th) ($n \times 10^6/\text{ejaculate}$)	<i>P</i> -values ($\alpha = 0.05$)*	<i>P</i> -values ($\alpha = 0.05$)#
rs3828743	AA	195	19.1 (1.3-58.0)	52.9 (1.9-160.3)	0.475	0.708
	AG	620	28.5 (3.9-61.2)	77.0 (9.1-207.0)		
	GG	587	22.6 (2.4-61.5)	75.2 (5.9-188.8)		
rs1128261	TC	149	18.3 (2.2-59.2)	48.3 (3.1-162.1)	0.504	0.637
	CC	1253	27.9 (3.9-61.1)	77.3 (10.2-202.9)		
rs6468544	CG	59	12.0 (1.0-64.7)	44.0 (2.0-255.4)	0.643	0.878
	CC	1343	23.7 (3.0-60.5)	75.1 (6.5-189.7)		
rs2583501	CC	9	7.0 (0.7-19.3)	11.9 (2.0-85.7)	0.256	0.385

	CT	209	22.0 (3.9-59.3)	70.5 (6.7-197.3)		
	TT	1184	25.3 (3.2-61.5)	77.4 (8.3-193.4)		
rs2289496	GG	178	27.6 (7.5-60.9)	89.9 (23.0-195.2)	0.595	0.708
	GT	642	23.6 (2.1-59.1)	74.0 (4.0-189.1)		
	TT	582	23.0 (3.2-64.7)	77.0 (7.2-203.9)		
rs2583506	TT	12	2.3 (0.0-10.0)	5.0 (0.0-77.0)	0.157	0.275
	CT	253	22.0 (3.6-58.5)	70.8 (5.9-190.9)		
	CC	1137	25.4 (3.6-62.1)	74.7 (8.3-193.5)		
rs1835740	GG	261	15.5 (3.1-56.4)	67.1 (6.6-180.6)	0.512	0.471
	AG	692	25.5 (3.2-60.4)	83.9 (8.7-193.8)		
	AA	449	26.8 (3.0-65.6)	76.9 (6.9-205.9)		

rs3763558	AA	10	3.8 (0.0-18.9)	16.0 (0.0-37.6)	0.264	0.049
	AC	187	26.1 (6.5-61.8)	77.6 (15.7-196.9)		
	CC	1205	24.7 (2.9-60.6)	76.4 (6.0-193.0)		

* Mann-Whitney test for the difference in sperm concentration among different genotypes of the SNPs.

Mann-Whitney test for the difference in total sperm count among different genotypes of the SNPs.

Table S4. Testing for Hardy-Weinberg equilibrium of the *TSPYL1* and *TSPYL5* variations in the groups of patient and control with copy dosage insufficiency of the *TSPY1*

SNP	Genotype	Genotype frequency		Hardy-Weinberg equilibrium testing ($\alpha = 0.05$)	
		Patients, <i>n</i> (%)	Controls, <i>n</i> (%)	<i>P</i> -values*	<i>P</i> -values#
rs3828743	AA	33 (16.3)	15 (14.7)	0.4602	0.4990
	AG	91 (45.1)	44 (43.1)		
	GG	78 (38.6)	43 (42.2)		
rs1128261	TC	26 (12.9)	12 (11.8)	0.3283	0.5279
	CC	176 (87.1)	90 (88.2)		
rs6468544	CG	12 (5.9)	4 (3.9)	0.6635	0.8399
	CC	190 (94.1)	98 (96.1)		

rs2583501	CC	3 (1.5)		0.3673	0.4568
	CT	33 (16.3)	14 (13.7)		
	TT	166 (82.2)	88 (86.3)		
rs2289496	GG	24 (11.9)	13 (12.8)	0.8513	0.7785
	GT	93 (46.0)	45 (44.1)		
	TT	85 (42.1)	44 (43.1)		
rs2583506	TT	3 (1.5)		0.4780	0.3585
	CT	35 (17.3)	17 (16.7)		
	CC	164 (81.2)	85 (83.3)		
rs1835740	GG	43 (21.3)	18 (17.7)	0.9586	0.9054
	AG	100 (49.5)	49 (48.0)		

	AA	59 (29.2)	35 (34.3)		
rs3763558	AA	2 (1.0)		0.6033	0.4568
	AC	30 (14.9)	14 (13.7)		
	CC	170 (84.2)	88 (86.3)		

* Hardy-Weinberg equilibrium test of patient group.

Hardy-Weinberg equilibrium test of control group.

Table S5. Comparisons of sperm productions among different genotypes of the SNPs in the males carrying fewer than 21 *TSPY1* copies

SC, sperm concentration; TSC, total sperm count

SNP	Genotype [^]	<i>n</i>	Median of SC (25th–75th) (<i>n</i> × 10 ⁶ /ml)	Median of TSC (25th–75th) (<i>n</i> × 10 ⁶ /ejaculate)	<i>P</i> -values ($\alpha = 0.05$) [*]	<i>P</i> -values ($\alpha = 0.05$) [#]
rs3828743	AA	48	6.8 (0-25.2)	16.0 (0-61.4)	0.612	0.483
	AG	135	7.2 (0-28.9)	17.9 (0-76.5)		
	GG	121	7.7 (0-34.0)	20.4 (0-91.0)		
rs1128261	TC	38	6.7 (0-26.1)	16.2 (0-73.1)	0.731	0.769
	CC	266	7.4 (0-31.5)	18.7 (0-81.6)		
rs6468544	CG	16	5.0 (0-12.6)	11.9 (0-51.2)	0.206	0.311
	CC	288	7.6 (0-32.3)	19.6 (0-84.2)		
rs2583501	CC + CT	50	5.9 (0-23.0)	13.6 (0-52.7)	0.462	0.357

	TT	254	7.7 (0-33.2)	20.4 (0-86.7)		
rs2289496	GG	37	7.8 (0.1-34.0)	21.3 (0.3-89.3)	0.843	0.775
	GT	138	7.0 (0-28.9)	17.0 (0-76.5)		
	TT	129	7.5 (0-31.5)	18.7 (0-83.3)		
rs2583506	TT + CT	55	6.6 (0-24.4)	15.3 (0-65.9)	0.744	0.582
	CC	249	7.6 (0-32.8)	19.6 (0-87.5)		
rs1835740	GG	61	6.2 (0-25.5)	15.1 (0-62.9)	0.609	0.453
	AG	149	7.1 (0-29.8)	17.9 (0-77.4)		
	AA	94	8.2 (0.2-35.7)	21.0 (0.5-91.1)		
rs3763558	AA + AC	46	6.4 (0.0-23.9)	15.3 (0.0-66.2)	0.486	0.325
	CC	258	7.5 (0-34.6)	18.3 (0-85.1)		

[^] The homozygous genotype that was observed in less than five subjects was mixed with the heterozygous genotype.

^{*} Mann-Whitney test for the difference in sperm concentration among different genotypes of the SNPs.

[#] Mann-Whitney test for the difference in total sperm count among different genotypes of the SNPs.