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Supplemental Data

**A Genome-wide Association Study Reveals that Variants
within the HLA Region Are Associated with Risk
for Nonobstructive Azoospermia**

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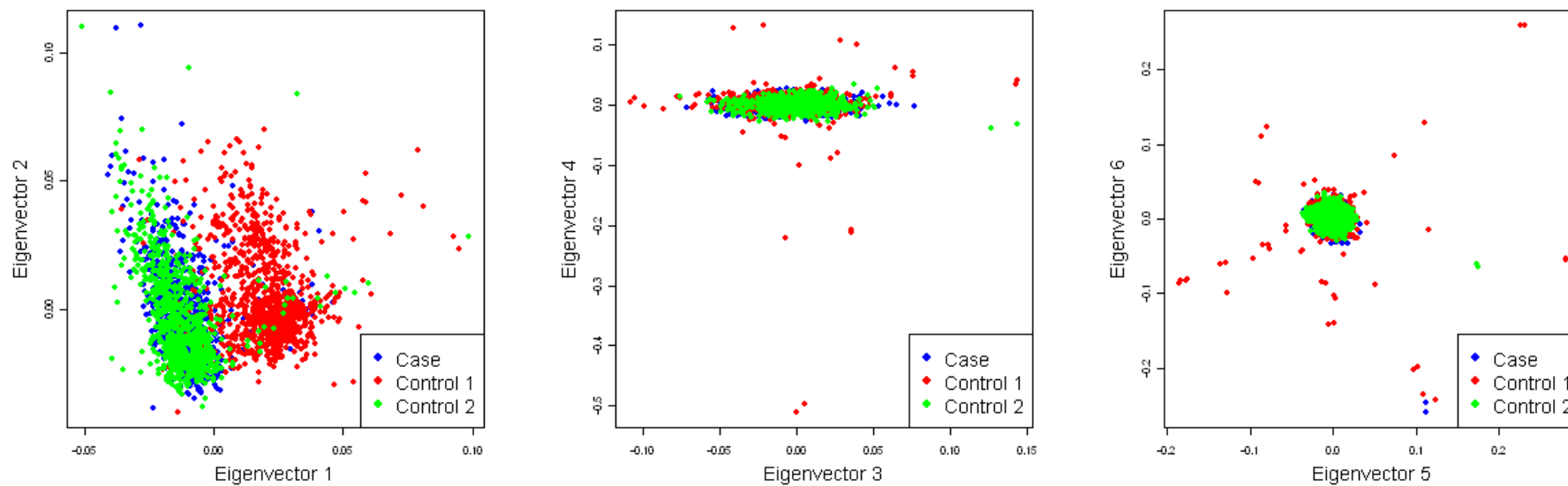


Figure S1. Principal Component Analysis of GWAS Samples of Nonobstrusive Assospermia (NOA) for the First Six Dimensions

From left to right, X-axis and Y-axis present the first vs the second, the third vs the fourth, and the fifth vs the sixth eigen vector identified by principal component analysis using Eigenstrat software, respectively. Control 1 refers to the 1,000 male controls and Control 2 refers to the 863 female controls.

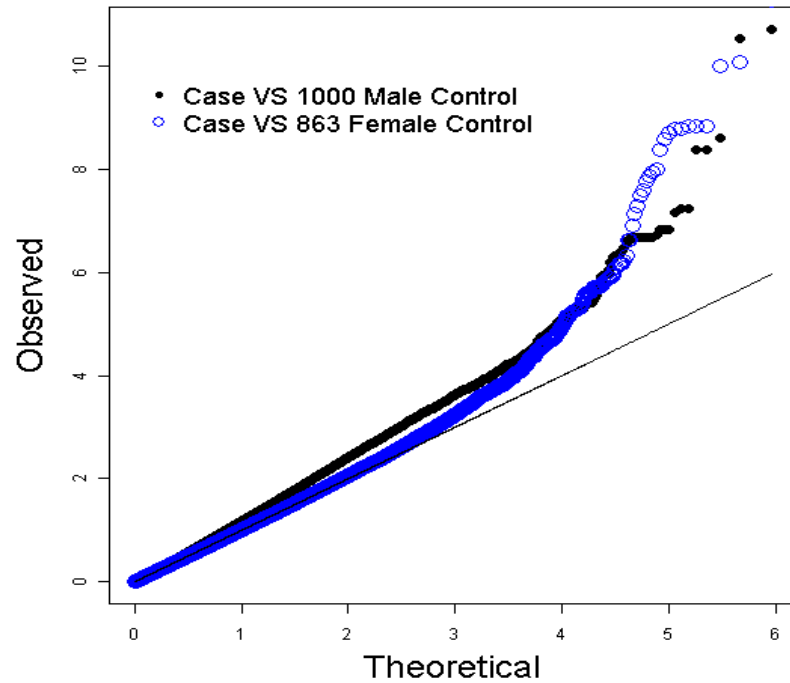


Figure S2. Quantile-Quantile Plots

The quantile-quantile (Q-Q) plot of observed versus expected test statistics was adjusted for age and first eigen (χ^2 test). The line in black indicated the association results for NOA cases vs 1,000 male controls. The line in blue indicated the association results for NOA cases vs 863 female controls.

Table S1. SNPs That Are Associated with NOA Risk in the Chinese Population

CHR	SNP	BP	Alleles		GWAS					Confirmation (Northern China)				
					Frequency of A1		OR	P	HWE	Status	Frequency of A1		OR	P
			A1	A2	Cases (n=802)	Controls (n=1858)					Cases (n=818)	Controls (n=1755)		
1	rs4845911	12,615,712	A	G	0.28	0.22	1.39	1.33E-05	0.39	G/G	0.23	0.24	0.95	0.55
1	rs4653551	226,694,536	G	A	0.26	0.33	0.71	3.01E-06	0.89	I/G	0.31	0.32	0.96	0.49
2	rs17033369	67,360,228	C	T	0.09	0.06	2.05	1.60E-08	0.35	G/I	0.08	0.08	0.95	0.7
2	rs3731611	119,459,081	G	T	0.43	0.5	0.72	9.70E-07	0.41	I/I	0.44	0.47	0.88	0.07
2	rs950233	201,302,430	A	G	0.14	0.19	0.66	5.75E-06	0.95	I/G	0.16	0.17	0.94	0.54
3	rs9876109	1,525,301	G	A	0.29	0.36	0.72	6.27E-06	0.96	I/G	0.34	0.34	1.01	0.89
4	rs10032586	25,009,566	G	A	0.1	0.07	1.75	1.47E-06	0.70	G/I	0.07	0.08	0.92	0.47
6	rs3130139	30,465,155	T	C	0.39	0.31	1.34	2.28E-05	0.11	G/I	0.35	0.33	1.11	0.1
6	rs975195	30,585,926	G	A	0.39	0.35	1.30	1.82E-04	0.03	G/I	0.35	0.35	0.99	0.88
6	rs2261033	31,711,570	A	G	0.45	0.5	0.80	1.09E-03	0.67	G/G	0.45	0.49	0.87	0.04
6	rs12614	32,022,158	T	C	0.12	0.06	2.37	8.99E-13	0.59	G/I	N/A	N/A	N/A	N/A
6	rs415929	32,297,010	C	T	0.26	0.19	1.43	3.18E-06	0.18	G/G	0.23	0.21	1.08	0.33
6	rs498422	32,394,739	G	T	0.2	0.13	1.81	1.53E-11	0.55	G/I	0.17	0.13	1.37	1.47E-04
6	rs2076531	32,471,690	G	A	0.11	0.06	2.03	1.00E-09	0.11	I/G	0.1	0.07	1.42	9.75E-04
6	rs3129878	32,516,713	C	A	0.4	0.29	1.63	1.03E-12	0.03	G/G	0.36	0.3	1.32	1.48E-05
6	rs7192	32,519,624	T	G	0.32	0.26	1.42	1.02E-06	0.88	G/I	0.32	0.27	1.26	3.71E-04
6	rs7747521	32,539,083	G	A	0.28	0.21	1.59	2.00E-09	0.35	I/G	0.29	0.21	1.55	2.02E-10
6	rs35120848	32,778,473	T	C	0.14	0.09	1.62	1.92E-06	0.14	G/G	N/A	N/A	N/A	N/A

CHR	SNP	BP	GWAS						Confirmation (Northern China)					
			Alleles		Frequency of A1		OR	P	HWE	Status	Frequency of A1		OR	P
			A1	A2	Cases (n=802)	Controls (n=1858)					Cases (n=818)	Controls (n=1755)		
6	rs6912492	32,977,687	A	G	0.22	0.16	1.42	2.23E-05	0.69	I/I	0.19	0.18	1.09	0.25
7	rs2385466	104,298,151	C	T	0.07	0.11	0.59	1.42E-05	0.25	I/G	0.1	0.11	0.93	0.55
7	rs7802723	137,579,847	T	C	0.1	0.06	1.89	2.07E-07	0.29	G/I	N/A	N/A	N/A	N/A
8	rs4976955	143,348,657	C	T	0.08	0.05	1.74	1.94E-05	0.32	G/I	0.06	0.07	0.85	0.27
9	rs1326807	118,625,673	C	T	0.1	0.06	1.79	1.59E-06	1.00	G/I	0.1	0.08	1.17	0.19
11	rs11040248	5,980,157	G	A	0.1	0.06	1.72	7.72E-06	0.67	G/I	0.1	0.1	0.99	0.93
11	rs10834232	23,883,206	C	T	0.09	0.06	1.91	1.02E-06	0.02	G/I	0.08	0.08	1.04	0.78
11	rs13377211	80,197,054	G	A	0.09	0.05	1.87	1.62E-06	0.62	I/I	0.09	0.09	0.95	0.72
11	rs12419806	92,640,128	C	A	0.19	0.26	0.69	5.93E-06	0.55	I/G	0.24	0.24	1	0.96
12	rs11057353	122,831,640	T	C	0.09	0.06	1.76	6.14E-06	0.21	G/I	0.07	0.07	1.02	0.88
13	rs4770720	18,537,078	T	G	0.11	0.08	1.66	8.74E-06	0.80	G/I	N/A	N/A	N/A	N/A
13	rs278040	29,357,712	C	T	0.1	0.06	1.75	5.06E-06	0.88	G/I	0.07	0.09	0.8	0.05
13	rs1320526	112,608,380	T	C	0.12	0.07	1.68	2.77E-06	0.71	G/I	0.09	0.1	0.85	0.11
14	rs7149094	50,948,557	A	G	0.11	0.08	1.68	4.51E-06	0.39	I/I	N/A	N/A	N/A	N/A
14	rs8006145	63,769,203	A	C	0.18	0.12	1.56	1.06E-06	0.94	G/I	0.16	0.15	1.1	0.29
15	rs12592037	35,623,271	T	C	0.13	0.18	0.67	1.40E-05	0.35	I/G	0.2	0.17	1.16	0.1
15	rs16964657	50,061,705	T	C	0.18	0.13	1.62	1.41E-07	0.52	G/I	0.14	0.16	0.85	0.1
17	rs2665969	71,633,804	C	T	0.13	0.08	1.79	3.50E-08	0.08	G/I	0.12	0.12	0.97	0.74
18	rs11152112	54,813,028	A	G	0.09	0.06	1.78	2.20E-06	0.55	G/I	0.1	0.09	1.16	0.2

N/A: SNPs wherein genotyping failed.