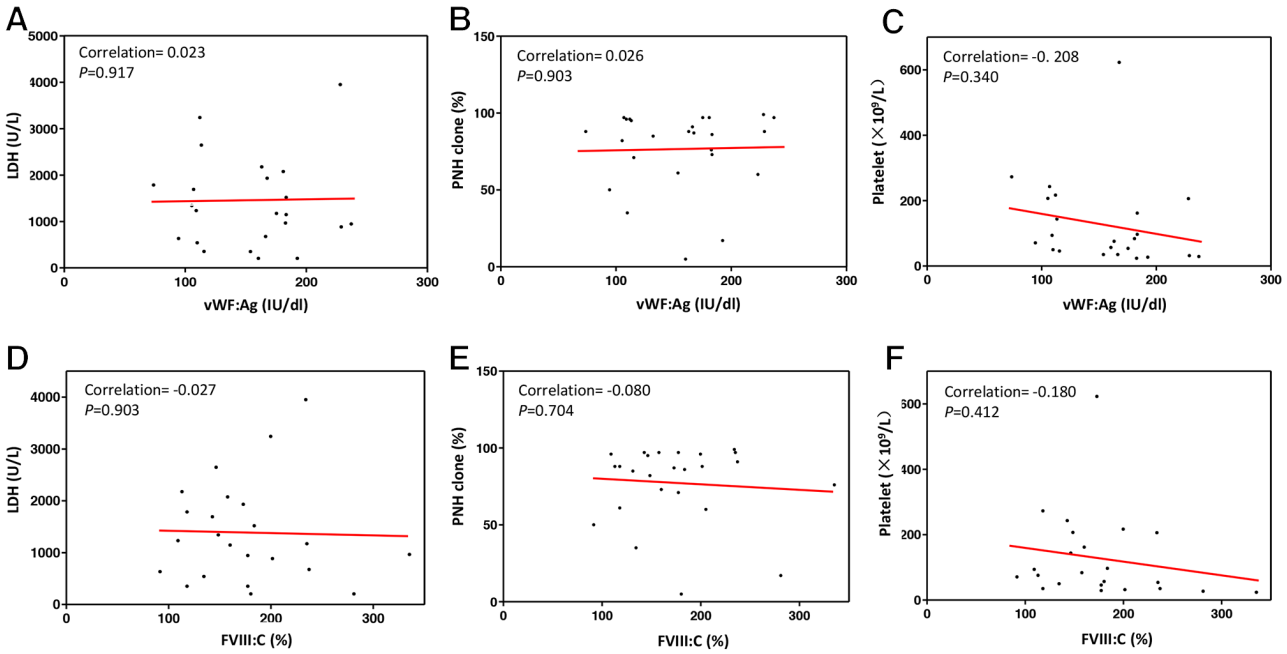
Polymorphism of the *ABO* gene associate with thrombosis risk in patients with paroxysmal nocturnal hemoglobinuria

SUPPLEMENTARY MATERIALS



Supplementary Figure 1: Correlation with the level of vWF:Ag and FVIII: C with possible factors among PNH patients: Correlation of the level of vWF:Ag with LDH **(A)**, PNH clone **(B)**, platelet **(C)**; correlation of the level of FVIII:C with LDH **(D)**, PNH clone **(E)**, platelet **(F)**.

Supplementary Table 1: Twenty-six alleles of 17 genes were detected in this study

Gene name	SNP	Associated phenotype
ABO	rs495828	vWF↑, VIII↑
	rs8176704	vWF↑, VIII↑
	rs8176719	vWF↑, VIII↑
	rs8176747	vWF↑, VIII↑
	rs8176750	vWF↑, VIII↑
	rs2519093	vWF↑, VIII↑
STXBP5	rs1039084	vWF↑
vWF	rs1063856	vWF↑
PROC	rs146922325	Protein C↓
GP6	rs1613662	platelet activation
THBD	rs16984852	thrombomudulin↓
F2	rs1799963	FII↑
MTHFR	rs1801133	methylenetetrahydrofolate reductase↓
	rs397507444	methylenetetrahydrofolate reductase↓
PROS1	rs199469491	Protein S↓
	rs199469495	Protein S↓
	rs121918474	Protein S↓
F11	rs2036914	FXI↑
	rs2289252	FXI↑
FGG	rs2066865	Fibrinogen YY'↓
SLC44A2	rs2288904	unkonwn
F5 leiden	rs6025	Resistance to activated pro C
KNG1	rs710446	aPTT↓
TSPAN15	rs78707713	unkonwn
PROCR	rs867186	sEPCR↑
VKORC1	rs9923231	vitamin K↑

Supplementary Table 2: Primers used for this study

SNP	1st-PCRP	2nd-PCRP	UEP_SEQ
rs495828	ACGTTGGATGCAGGTGTCATGGAGAAATGG	ACGTTGGATGACCTGTAACTGTTGCAAGGG	TTGCAAGGGAGGTAAA
rs8176704	ACGTTGGATGATTCACCAGACACGTCGAAG	ACGTTGGATGGGGCCTCATGGTTTGTAGAC	AAACTCCACTCTGCC
rs8176719	ACGTTGGATGTGTCGATGTTGAATGTGCCC	ACGTTGGATGCATGTGCAGTAGGAAGGATG	AGGATGTCCTCGTGGT
rs8176747	ACGTTGGATGACGAGGGCGATTTCTACTAC	ACGTTGGATGTGGTCGACCATCATGGCCTG	ACCGACCCCCCGAAGAAC
rs8176750	ACGTTGGATGTAAAACCAAGGGCGGGAGG	ACGTTGGATGGGTGCCCAAGAACCACCAG	AGGCGGTCCGGAACC
rs2519093	ACGTTGGATGTATACACCTACTATGAACCC	ACGTTGGATGAATGAATAAATAAGCCACC	ccccTAAGCCACCGACTGA
rs1039084	ACGTTGGATGGGAGGTATGTAGTATGCTTG	ACGTTGGATGTAACTTTGAGCACCCAAGCC	AAGCCCCAATTACCTCCG
rs1063856	ACGTTGGATGCATGGTCAAGCTGGTGTGTC	ACGTTGGATGTGCACTCCAGGTCATAGTTC	TCTGGCACGTTTTGG
rs146922325	ACGTTGGATGGGTCTTCTGTGTCTCGTTTC	ACGTTGGATGCTCAGTGAAGTTCCCTTGTG	ccgGGGAGGCCCTGGAAG
rs1613662	ACGTTGGATGATTTCCCAGGAACCTCTGTG	ACGTTGGATGATGGACCCTGCAGAACCTAC	tcACCTACCTGCTACCG
rs16984852	ACGTTGGATGGCACTTATAAACTCGAGCCC	ACGTTGGATGAGCCCAGACACTTCTTGCC	ctcaGCTGCGCGCAGCCCCTG
rs1799963	ACGTTGGATGTGAATAGCACTGGGAGCATT	ACGTTGGATGTGGAACCAATCCCGTGAAAG	gttcaCAATAAAAGTGACTCTCAGC
rs1801133	ACGTTGGATGCTTGAAGGAGAAGGTGTCTG	ACGTTGGATGCTTCACAAAGCGGAAGAATG	gaagGCTGCGTGATGATGAAATCG
rs397507444	ACGTTGGATGGCTGACCAGTGAAGAAAGTG	ACGTTGGATGCTCACTTTGTGACCATTCCG	tAGAGGTAAAGAACGAAGACT
rs199469491	ACGTTGGATGCACCTCCAGTTGTGATTTTG	ACGTTGGATGTTGCACTTCGTGGTGGAAAG	AAGATTGAAGTTCAGCTTAAGAA
rs199469495	ACGTTGGATGAGCTAAAGAAGCTGTGATGG	ACGTTGGATGACTTTGGTTTCCAGCAATCC	aGGCTTAAAAAGGGGTCCA
rs121918474	ACGTTGGATGTCTACCATCCTGCTCTTACC	ACGTTGGATGGGAAGTTACCACTGTTCCTG	aaagGGTTTTGTTATGCTTTCAAAT
rs2036914	ACGTTGGATGTGTCTTGGAGACAAGGAGTG	ACGTTGGATGAAAGCAGCAGCTTTTGCCAG	AGCAGCTTTTGCCAGTAAAGA
rs2289252	ACGTTGGATGTTTTCCTCCCTCAGGTTCAG	ACGTTGGATGAGTTGGATGAGGAGTTAGCG	GTGAGGCTTGTCTCTCTCT
rs2066865	ACGTTGGATGATTACTACTGATGGTTGCCC	ACGTTGGATGGTTTCCTAAGACTAGATAC	TTCCTAAGACTAGATACATGGTA
rs2288904	ACGTTGGATGATCTCTCTTGGCAGGGAGTG	ACGTTGGATGTGACTCACAGGGTTTGCTGG	GGGCAGTCACCATCT
rs6025	ACGTTGGATGCTCTGGGCTAATAGGACTAC	ACGTTGGATGCTTCAAGGACAAAATACCTG	AAGGACAAAATACCTGTATTCCT
rs710446	ACGTTGGATGTCAGGGATCCAATCGTCATC	ACGTTGGATGCAGGACTCTGATCTCATTGC	ATATCACCAGCTCCCA
rs78707713	ACGTTGGATGTTCGACTTCACCTTCCACCC	ACGTTGGATGGGAAGCTGAGCAGAAATGAG	cTGTGAGCAAAACAGCG
rs867186	ACGTTGGATGACAAGCCGCTCCTACACTTC	ACGTTGGATGAAGATGCCTACAGCCACACC	ACCAGCAATGATGAAAC
rs9923231	ACGTTGGATGTCTGGGAAGTCAAGCAAGAG	ACGTTGGATGGCTAGGATTATAGGCGTGAG	aggaGTGAGCCACCGCACC

Supplementary Table 3: Sites of thrombosis in 17 patients

Thrombus site	Events, n	% of total
Venous		
Portal vein	6	28.6
Mesenteric vein	3	14.3
Cerebral vein	3	14.3
Splenic vein	3	14.3
Deep vein	3	14.3
Pulmonary embolus	1	4.8
Arterial		
Cornary	2	9.5
Total	21	100

Supplementary Table 4: Hardy-Weinberg equilibrium and minor allele frequency of 17 SNPs

Gene name	SNP	x^2	*P* value	MAF	MAF in database
ABO	rs495828	1.136	0.286	0.1875	0.1603
	rs8176704	104	<0.001	0.0577	0.0463
	rs8176719	3.119	0.077	0.4952	0.3438
	rs8176747	0.479	0.489	0.3173	0.1528
	rs2519093	2.36	0.125	0.1779	0.1414
STXBP5	rs1039084	0.68	0.41	0.2981	0.4571
vWF	rs1063856	2.649	0.104	0.0481	0.3363
GP6	rs1613662	0.063	0.802	0.024	0.1573
THBD	rs16984852	0.022	0.881	0.0144	0.0014
F11	rs2036914	0.283	0.595	0.2356	0.3936
	rs2289252	6.481	0.011	0.2596	0.3195
FGG	rs2066865	0.337	0.562	0.4856	0.3027
SLC44A2	rs2288904	0.238	0.626	0.3413	0.1827
KNG1	rs710446	0.415	0.519	0.2548	0.4153
TSPAN15	rs78707713	0.002	0.96	0.0048	0.0479
PROCR	rs867186	0.265	0.607	0.0481	0.096
VKORC1	rs9923231	1.177	0.278	0.0962	0.3556