

Supplemental Data

Association of common *TMPRSS6* and *TF* gene variants with hepcidin and iron status in healthy rural Gambians.

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Supplemental information

Supplemental Table S1. Allele configuration of the allele risk score from the 6 *TMPRSS6* SNPs

SNP	rs2235321	rs855791	rs4820268	rs2235324	rs2413450	rs5756506	Total Risk score
major/minor	G/A	G/A	A/G	A/G	G/A	G/C	
Risk allele*	A	A	A	G	G	C	
normal allele	G	G	G	A	A	G	
Risk genotype (ARS)	A/A (2)	A/A (2)	A/A (2)	G/G (2)	G/G (2)	C/C (2)	12
normal group	G/G (2)	G/G (0)	G/G (0)	A/A (0)	A/A (0)	G/G (0)	0

ARS, allele risk score

* Allele linked to low iron status from the previously-published studies
Bolded alleles are those associated with the risk of low iron status.

Supplemental Table S2. Details of the allele configuration for the allele risk score from the 2 *TF* SNPs

SNP	rs1799852	rs3811647	Genotype combination	ARS	N
major/minor	G/A	G/A			
Risk allele*	G	A			
WT/WT	GG	GG	GG /GG	2	825
Het/Het	AG	AG	AG /AG	2	10
Homo/Homo	AA	AA	AA/ AA	2	0
WT/Homo	GG	AA	GG/ AA	4	27
Homo/WT	AA	GG	AA/GG	0	9
Het/WT	AG	GG	AG/GG	1	154
WT/Het	GG	AG	GG/ AG	3	290
Het/Homo	AG	AA	AG/ AA	3	1
Homo/Het	AA	AG	AA/AG	1	0

* Allele linked to low iron status from previously-published studies; bolded letters indicates risk alleles

Supplemental Table S3. A list of all the 94 genotype combinations generated from the 6 *TMPRSS6* SNPs

Genotype group	N	<i>TMPRSS6</i> SNP ARS
GG/GG/GG/AA/AA/GG	18	0
GG/GG/GG/AA/AG/GG	16	1
GG/GG/GG/GA/AA/GG	10	1
GG/GG/GA/AA/AG/GG	26	2
GG/GG/GG/GA/AG/GG	20	2
AG/GG/GG/AA/AG/GG	4	2
AG/GG/GG/GA/AA/GG	4	2
GG/AG/GG/AA/AA/CG	3	2
GG/GG/GG/GG/AA/GG	3	2
GG/GG/GG/GG/AA/GG	2	2
GG/GG/GG/AA/GG/GG	1	2
GG/GG/GG/GA/AA/CG	1	2
AG/GG/GA/AA/AG/GG	72	3
GG/GG/GA/AA/AG/CG	16	3
GG/GG/GA/AA/GG/GG	16	3
GG/GG/GA/GA/AG/GG	12	3
AA/GG/GG/AA/AG/GG	1	3
AG/GG/GA/NA/AG/NA	1	3
AG/GG/GG/GA/AG/GG	1	3
GG/AG/GA/AA/AG/GG	1	3
GG/AG/GG/GA/AA/CG	1	3
AG/GG/GA/GA/AG/GG	81	4
GG/GG/GA/GA/AG/CG	52	4
AG/GG/GA/AA/GG/GG	29	4
GG/AG/GA/GA/AG/GG	20	4
GG/GG/AA/AA/GG/GG	14	4
GG/GG/GA/GA/GG/GG	9	4
GG/GG/GA/AA/GG/CG	6	4
AG/AG/GG/AA/AG/CG	2	4
AG/GG/GG/GA/GG/GG	2	4
GG/GG/GA/GG/AG/GG	2	4
GG/GG/GG/GG/GG/GG	2	4
AA/GG/GA/AA/AG/GG	1	4
GG/GG/GG/GG/AG/CG	1	4

Supplemental Table S3 cont.

Genotype group	N	<i>TMPRSS6</i> SNP ARS
AG/GG/AA/AA/GG/GG	42	5
AG/GG/GA/GA/GG/GG	42	5
GG/GG/GA/GA/GG/CG	25	5
AG/GG/GA/GG/AG/GG	16	5
GG/GG/GA/GG/AG/CG	8	5
GG/GG/AA/GA/GG/GG	7	5
AA/GG/GA/AA/GG/GG	5	5
AG/AG/GA/AA/AG/CG	5	5
GG/AG/GA/GA/AG/CG	5	5
GG/GG/AA/AA/GG/CG	5	5
GG/AG/GA/GA/GG/GG	4	5
AA/GG/GA/GA/AG/GG	3	5
AG/GG/GA/AA/GG/CG	3	5
GG/GG/GA/GG/GG/GG	2	5
AG/AG/GA/GA/AG/GG	1	5
AG/GG/GA/GA/AG/CG	1	5
GG/AG/AA/AA/GG/GG	1	5
GG/AG/GA/AA/AG/CC	1	5
NA/GG/AA/GA/GG/GG	1	5
AA/GG/AA/AA/GG/GG	75	6
AG/GG/AA/GA/GG/GG	56	6
AG/GG/AA/AA/GG/CG	24	6
GG/GG/AA/GA/GG/CG	20	6
GG/AG/AA/GA/GG/GG	19	6
AG/GG/GA/GG/GG/GG	16	6
AA/GG/GA/GA/GG/GG	12	6
GG/GG/GA/GG/GG/CG	7	6
GG/AG/GA/GG/GG/GG	5	6
AA/GG/GA/GG/AG/GG	4	6
AG/AG/GA/GA/AG/CG	3	6
AG/GG/GA/GA/GG/CG	2	6
GG/AG/GA/GA/GG/CG	2	6
GG/GG/AA/AA/GG/CC	2	6
AG/AG/GA/GA/GG/GG	1	6
AG/GG/GA/GG/AG/CG	1	6

Supplemental Table S3 cont.

Genotype group	N	<i>TMPRSS6</i> SNP ARS
GG/AG/GA/GA/AG/CC	1	6
GG/GG/AA/GG/GG/GG	1	6
NA/GG/AA/GA/GG/CG	1	6
AA/GG/AA/GA/GG/GG	123	7
AG/GG/AA/GA/GG/CG	62	7
AG/AG/AA/GA/GG/GG	32	7
AG/GG/AA/GG/GG/GG	19	7
GG/GG/AA/GA/GG/CC	8	7
GG/GG/AA/GG/GG/CG	6	7
GG/AG/AA/GA/GG/CG	4	7
GG/AG/AA/GG/GG/GG	2	7
AG/GG/AA/GG/GG/CG	50	8
AA/GG/AA/GG/GG/GG	46	8
AG/AG/AA/GG/GG/GG	30	8
GG/GG/AA/GG/GG/CC	14	8
GG/AG/AA/GG/GG/CG	13	8
AG/AG/AA/GA/GG/CG	6	8
GG/AG/GA/GG/AG/GG	6	8
GG/AA/AA/GG/GG/GG	1	8
GG/AG/AA/GG/GG/CC	6	9
AG/AG/AA/GG/GG/CG	2	9
AG/GG/AA/GG/GG/CC	2	9
AA/GG/AA/GG/GG/CG	1	9
GG/AA/AA/GG/GG/CG	1	9
GG/AA/AA/GG/GG/CC	3	10

Abbreviations: ARS, allele risk score

Legend: NA, genotypes that were not available for that particular SNP in the combination.

TMPRSS6 SNP ARS, number of iron-lowering alleles within a genotype combination

Supplemental Table S4. The effects of *TMPRSS6* allele risk score on hepcidin, controlling for age, sex and CRP

TMPRSS6 ARS*	N	Mean Hepcidin (ng/ml)	Std. error	Beta	P-value
0	18	13.4	3.66	Reference group	
1	26	8.6	4.48	-4.80	0.285
2	61	10.8	3.89	-2.64	0.498
3	123	11.7	3.66	-1.68	0.645
4	221	10.9	3.55	-2.49	0.483
5	185	9.0	3.58	-4.39	0.220
6	253	9.2	3.54	-4.26	0.229
7	254	10.4	3.54	-3.04	0.390
8	160	9.8	3.61	-3.61	0.317
9	12	13.6	5.41	0.16	0.977
10	3	3.5	9.04	-9.96	0.270

* Number of risk alleles based on **Table S1**