##### GWAS Results

##### Less Stringent QC

##### # This command generates the association P-values for each individual SNP in relation to the phenotype.

##### plink --bfile smokescreen\_s6\_p2 –assoc --out assoc\_smokescreen\_s6\_p2

##### # This command generates the association P-values for each individual SNP in relation to the phenotype and the adjusted values with different correction methods. Results for the significant SNP seen below.

##### plink --bfile smokescreen\_s6\_p2 --assoc --adjust --out adj\_assoc\_smokescreen\_s6\_p2

##### Genomic inflation est. lambda (based on median chisq) = 1 (adjust)

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **CHR** | SNP | UNADJ | GC | BONF | HOLM | SIDAK\_SS | SIDAK\_SD | FDR\_BH | FDR\_BY |
| 9 | AX-42555763 | 1.88E-08 | 1.88E-08 | 0.006782 | 0.006782 | 0.006759 | 0.006759 | 0.006782 | 0.09071 |

AX-42555763 = rs4573325

The AX to rs conversion was done with an excel provided by the smokescreen project staff. This excel contains all the AX SNPs codes and their rs counterparts.

##### More Stringent QC

##### # This command generates the association P-values for each individual SNP in relation to the phenotype.

##### plink --bfile smokescreen\_stringent\_p2\_s6 –assoc --out assoc\_smokescreen\_stringent\_p2\_s6

##### # This command generates the association P-values for each individual SNP in relation to the phenotype and the adjusted values with different correction methods. Results for the significant SNP seen below.

##### plink --bfile smokescreen\_stringent\_p2\_s6 --assoc --adjust --out adj\_assoc\_smokescreen\_stringent\_p2\_s6

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| CHR | SNP | UNADJ | GC | BONF | HOLM | SIDAK\_SS | SIDAK\_SD | FDR\_BH | FDR\_BY |
| 9 | AX-42555763 | 1.44E-08 | 1.51E-08 | 0.004713 | 0.004713 | 0.004702 | 0.004702 | 0.004713 | 0.06256 |

Genomic inflation est. lambda (based on median chisq) = 1.00282 (--adjust).

The significantly different SNP rs4573325 is part of a non-coding (intron) region of the HMCN2 gene. Below you can find the list of the 25 proteins that are more closely associated with the HMCN2 gene.

![A picture containing map

Description automatically generated]()

Regulome DB report can be found on the following link:

<http://www.regulomedb.org/snp/chr9/133274264>

An analysis on the PolyPhen2 website demonstrated that the rs4573325 SNP has no variant that directly affects the structure of any protein (as is expected for an intron region SNP).

<http://genetics.bwh.harvard.edu/ggi/pph2/198e031152c86ee8e1050d4beb5cc2f60fc919b7/1/pph2-full.txt>

PS: The stringent QC data set was submitted to further analyses to control for population stratification. Principal component analyses was utilized to control for stratification, SNP rs4573325 remained significant even after PCA, More detailed information on the population stratification process and results can be found in “Population Stratification.docx”