

Exploring SNP2GENE Genome-wide plots

1. What is the most significantly associated gene from the gene-based analysis?
 - a. AL138847.1
2. Based on MAGMA gene-set analysis, what processes are enriched in the GWAS risk regions?
 - a. Processes related to lipid homeostasis and metabolism
3. Based on MAGMA gene-set analysis, which tissue is significantly enriched amongst GWAS loci?
 - a. Liver – your liver makes cholesterol

Exploring SNP2GENE Results Summary

4. How many genomics risk loci are associated with LDL-C
 - a. 69
5. How many genes are mapped to these regions?
 - a. 569
6. Which genomic region (e.g. intronic, intergenic, 3'UTR etc) is significantly under-represented amongst candidate GWAS SNPs?
 - a. intergenic
7. What proportion of the candidate SNPs are intronic?
 - a. 0.532
8. Which genomic region is the most enriched amongst candidate SNPs?
 - a. UTR3 (2.23 fold enrichment)

Exploring SNP2GENE Results

9. What is the rs ID for the most significantly associated SNP?
 - a. rs646776
10. Which chromosome is this SNP located on?
 - a. Chr 1
11. What genomic region is the SNP located in (e.g. coding region for gene X, UTR3 for gene Y, downstream of gene X or intergenic between gene X and gene Y)?
 - a. downstream of CELSR2 or intergenic between CELSR2 and PSRC1
12. What is the CADD score for this SNP?
 - a. 2.441
13. What is the RegulomeDB score for this SNP and what does the score mean?
 - a. 2b
14. Which genes is the above SNP an eQTL for in GTEx Liver data?
 - a. CELSR2, SORT1, PSRC1, ATXN7L2 and SYPL2
15. Of the above identified genes, which have evidence of physical interaction between the GWAS associated region and the gene's promoter (use the circus plot to determine this)?
 - a. CELSR2, SORT1, ATXN7L2, SYPL2
16. Which Wiki pathways related to medication are enriched in LDL-C-associated loci?
 - a. Statin, evolocumab
17. Which non-cardiovascular GWAS data also show an enrichment of LDL-C-associated loci?
 - a. Bipolar disorder FASD1, FADS2, CIKLP2, GMIP