

TP 4

(from Thornton T.)

1. Read in the variants within "Gene1.txt" into R. Calculate the minor allele frequencies of the variants and plot a histogram. What do you notice?
2. Read in the trait value in the file "Trait1.txt". Then test for an association between each variant in Gene1 and the Trait1. Is anything significant after adjusting for multiple testing?
3. Test for an association between the rare variants in Gene1 and the quantitative trait. For now, let's define rare variants to be the variants with $MAF < 3\%$. Specifically, apply :
 - CAST (Binary collapsing approach)
 - MZ Test/GRANVIL (Count based collapsing)
 - Weighted Count Based Collapsing where weights can depend on MAF
 - SKAT (Sequence Kernel Association Test)
 - standard SKAT (default parameters)
 - omnibus SKAT
 - with different ρ values (0, 1, 0.5, ...)
 - with "optimal" ρ value
4. Redo the previous question using the variants inside of "Gene2.txt" and "Trait2.txt".
5. Let's consider alternative definitions for rare variant : $MAF < 5\%$, $< 3\%$, $< 1\%$, $< 0.5\%$.