

Final Project (Due May 6, midnight)
BMI 620 – Statistics in Human Genetics

1. We will grade your final projects based on the in-class presentation and the written report (< 1500 words).

2. Each team will be assigned a different GWAS. Significance of findings will not affect your grade. Instead, we value rigorous analyses and clear interpretations.

3. Prepare 4 sections in the report.

Introduction: Introduce the background information about the trait.

Methods: Introduce all the methods you applied in your analysis.

Results: Report the findings. You are encouraged to use figures and tables to present your results.

Conclusion: Summarize the implications. What do your results tell us about this trait?

In addition, include a paragraph briefly describing how the work is distributed between team members.

4. Submit your written report with annotated code/script for all analyses. Scripts will not be counted in the word limit.

5. Here is a list of required items in your report:

- Manhattan and QQ plots
- LocusZoom plot for the most significant locus (<http://locuszoom.sph.umich.edu>) in your GWAS. Showing the +/- 500kb window would suffice.
- LDSC results on intercept and heritability
- Partition trait heritability by tissue annotations you used in HW assignments and quantify h^2 enrichment
- Test pairwise genetic correlations with 5 other GWAS in the folder

6. Choose **two** methods from the list and apply to your data. If needed, you are allowed to use any GWAS in the folder.

- TWAS (PrediXcan or Fusion)
- Mendelian randomization with 5 other GWAS (IVW approach)
- Local genetic correlation with 5 other GWAS (SUPERGNOVA)
- Structural equation modeling (GSEM)
- Quantify PRS performance with sumstats-based cross-validation (PUMAS)