**Population genetics, Quantitative genetics, and QTLs: Study guide and readings**

***Required readings:***

Cistue et al. 2011. Theor. Appl. Genet. 122: 1399-1410

1. Full paper

Munoz-Amatriain et al. 2014. PLOS One. <https://journals.plos.org/plosone/article?id=10.1371/journal.pone.0094688>

1. Full paper

**Population genetics**

1. In the context of population genetics distinguish between population, gene pool, and gene frequency.
2. Distinguish between the terms “genotype frequency” and “allele frequency”.
3. In your own terms, explain four key elements of the Hardy Weinberg theorem
4. Given p and q values of 0.8 and 0.2, respectively, what are the frequencies of AA, Aa, and aa genotypes in a population that is Hardy-Weinberg equilibrium?
5. What are the four factors that can alter Hardy-Weinberg equilibrium in a population?
6. Why is inbreeding depression in angiosperms a key consideration given that the
ancestral and basal condition is hermaphroditic?

**Quantitative genetics**

1. Given a phenotypic frequency distribution, be able to describe it as qualitative or quantitative and be able to defend your choice of terms.
2. In the progeny of a cross, what are transgressive segregants and what is a genetic explanation for their occurrence?
3. Why are quantitatively inherited traits often described as “multigenic”?
4. What are the components of phenotypic variance?
5. Why does Vp = Ve in the F1 generation of the cross between two inbred parents?
6. Why might you expect Vd in the F2 offspring of two inbred parents but not in the doubled haploid progeny of the same cross.
7. What is the difference between broad sense and narrow sense heritability?
8. If you read that in an experiment the narrow sense heritability of bread flavor is 7%, what expectation would you have for the heritability of the same trait in different wheat germplasm?
9. How does heritability relate to predicted response to selection?
10. If heritability estimates are, to some extent, under your control when you design an experiment, of what use are they?

**Biparental QTL mapping**

1. What are the principal steps in a biparental QTL analysis?
2. What is the importance of a linkage map for biparental QTL analysis?
3. Explain, in your own words, the meaning and implications of typical QTL metrics: e.g. location, LOD, additive effect, and R2.
4. Explain how, in the case of a species with complete linkage maps, the number of linkage groups will equal the n number of chromosomes.
5. How can a QTL analysis assist in understanding the genetics basis of transgressive segregation in a population?
6. What is the importance of population size in determining, via biparental QTL analysis, if an association of two phenotypic characters is due to linkage or pleiotropy?

**GWAS**

1. Why might you choose GWAS as a strategy for QTL mapping, rather than using a biparental population?
2. What is intragenic recombination and what are the implications if the cross-over event is reciprocal vs. if the cross-over event is non-reciprocal?
3. What is a Manhattan plot and how does linkage disequilibrium relate to the number of significant markers and their positions relative to the underlying map?
4. Why is MAF a consideration in GWAS but not in the analysis of a biparental mapping population, comprised of homozygous siblings, that is derived from the cross of two inbred parents?
5. What factors determine the rate of LD decay?

**Integration**

1. If you identify a candidate gene using QTL analysis, is that the end of the story? If not, what other steps are necessary to conclusively prove that the gene is indeed responsible for the phenotype under study?
2. What is a perfect marker, and is it possible to have more than one perfect marker in a gene?
3. Give three defensible reasons for mapping QTLs.

***Cistue et al.***

1. What is segregation distortion and what effect did it have on mapping QTLs in the Oregon Wolfe Barley?
2. Do these authors conclude that coincident QTL peaks for two different traits are definitive evidence for pleiotropy?

***Munoz-Amatriain et al.***

1. In what way can GWAS expedite the incorporation of genetic diversity into breeding programs?
2. Why are there different rates of LD decay in modern varieties vs. land races?
3. Of what value were a consensus linkage map and a barley genome sequence for GWAS?
4. What is genetic redundancy in the context of germplasm collections and why did it go undetected prior to advent of cost-effective high density marker data sets?

**Supplementary resources: Not required.**

Asins et al. 2009. QTL analysis in Plant Breeding. [https://link.springer.com/content/pdf/10.1007%2F978-90-481-2967-6\_1.pdf](https://link.springer.com/content/pdf/10.1007/978-90-481-2967-6_1.pdf)

Gibson, G. 2018. Population genetics and GWAS. [https://link.springer.com/content/pdf/10.1007%2F978-90-481-2967-6\_1.pdf](https://link.springer.com/content/pdf/10.1007/978-90-481-2967-6_1.pdf)

Hernandez et al. 2019. Stem rust resistance GWAS. <https://apsjournals.apsnet.org/doi/pdf/10.1094/PHYTO-09-18-0350-R>