Main Points: R05

Note that we will review and discuss the main points in class, so if anything here remains unclear after you read, definitely bring your questions to class!

Textbook: Ch 5

- **Linkage studies** leverage pedigrees/family history
  - looking for locations in the genome *inherited by* individuals with a condition and not by those without
  - identifies location in the genome whose presence is "linked" with a trait/condition
  - Linkage study success: HD and KE family and language
- **Association studies** leverage populations
  - Looking for locations in the genome *present* in individuals with a condition and not in those without
  - Requires much larger sample sizes than linkage studies
- **Candidate gene studies** are studies that are carried out b/c a scientist has a gene they're interested in studying b/c they *think* it plays a role in a condition/trait
- **Endophenotype** studies study a trait/condition associated with a condition of interest, rather than the trait directly

Textbook: Ch 6

- **GWAS (Genome-wide association study)** leverages **linkage disequilibrium** (the fact that parts of our genome are inherited in blocks) and genetic markers across the genome to find loci present more often in affected individuals than in controls
  - Manhattan plots summarize the results of GWAS
  - Individual loci effects on trait being studied tend to be small

Textbook: Ch 16-16.3

- **Reverse genetics**: phenotype -> genotype (see a phenotype and then work backward to figure out the genetic cause)
- **Forward Genetics**: genotype -> phenotype (genetically engineer a genotypic change and then determine if there's a phenotypic change)
  - Genetic engineering approaches: embryonic stem cells + homologous recombination, CRISPR