STAT/BIOST 550:online

Statistical Genetics – I
Mendelian traits in populations
and on pedigree members

1.1.1 GENETIC TERMINOLOGY

- Chromosome--- long string of double-strand DNA
- Cell nucleus --- has 46 chromosomes (22 pairs of autosomes, and 2 sex chromosomes, X,Y)
- Locus--- position on a chromosome, or DNA at that position,or the piece of DNA coding for a trait.
- Allele--- type of the DNA at a particular locus on particular chromosome.
- Genotype--- (unordered) pair of alleles at a particular locus in a particular individual.
- Homozygote-- a genotype with two like alleles.
- Heterozygote -- a genotype with two unlike alleles.
- Phenotype--- observable characteristics of an individual

# 1.1.2 EXAMPLE: ABO blood types

- The ABO locus is on chromosome 9
- The (main) alleles at the locus are A, B, and O
- The 6 genotypes are AA, AO, BB, BO, AB and OO.
-     Homozygotes are AA,BB,OO.
-     Heterozygotes are AO, BO and AB
- The 4 phenotypes are blood types A, B, AB and O.
-     The O allele is recessive to A and to B.
-     A and B are each dominant to O.
-     A and B are codominant
- What is a gene?? --- the chunk of DNA coding for a functional protein. Not a locus. Not an allele.

1.1.3 MENDEL's LAWS (1866)

- At any given locus, each individual has two genes, one maternal and the other paternal.
- 1. Each individual segregates a randomly chosen one of its two genes to each offspring, independently to each offspring, independently of gene segregated by the spouse, independently of gene segregated from parent.
- 2. Independently for different loci. (Not true; segregation of genes at loci on the same chromosome are dependent.)
- Mendel's first law says all transmissions of DNA from parent to offspring are independent.
- For every individual with both parents specified there are two transmissions, the one from his/her father (paternal) and the one from his/her mother (maternal).

1.2.1 REPRESENTATION OF PEDIGREES

- Graphical representations
  (i) parent-offspring links.
  (ii) sibship representation
  (iii) marriage-node graph.
- Founders (parents unspecified) and Non-founders (both parents specified).
- Gender: male, female, and unknown. (square, circle, diamond).
- Shading or labeling of individuals.
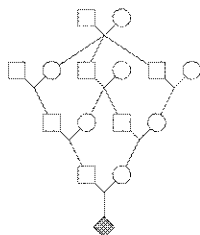
1.2.2 SPECIFICATION OF PEDIGREES

- Unique individual identifiers (``names'')
- Parent-offspring trios. (default: ind, dad, mom)
- Specification of founders. (parent ``names'' =0)
- Gender: male, female, and unknown. (1, 2, 0) or (M, F, U)
- Phenotypic, covariate, and marker data.
- ``Chronological'' (partial) ordering of pedigrees.

101	0	0	1	----
102	0	0	2	----
201	101	102	2	----
204	101	102	1	----
206	101	102	1	----
fred	0	0	1	----
203	0	0	2	
joe	fred	201	1	----
jane	204	203	2	
dave	204	203	1	
hugh	joe	jane	1	
etc.				

1.2.3 TYPES OF RELATIONSHIP

- Related: share common ancestor(s).
- Inbred: parents are related.
- Unilateral: related through one parent.
- Bilateral relatives: Both related through both parents. E.g. Double first cousins.
- Degrees of (regular, full) cousins:
- count down from the ancestors to first level to get degree: e.g. 2nd cousins share grt-grnd-pars.
- then count down from that level to the other
- individual to get count of generations "removed"