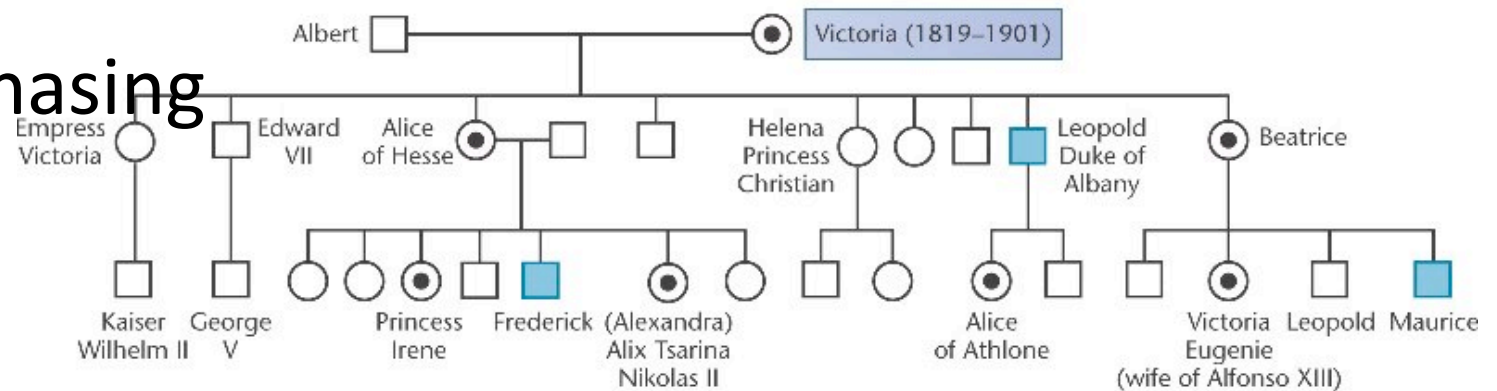
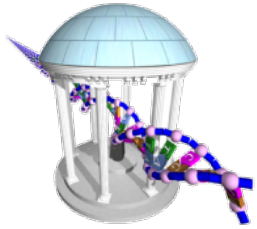


Pedigree and Genotype



- Genotype
- Allele States
- Inherited Allele States
- Trio Phasing

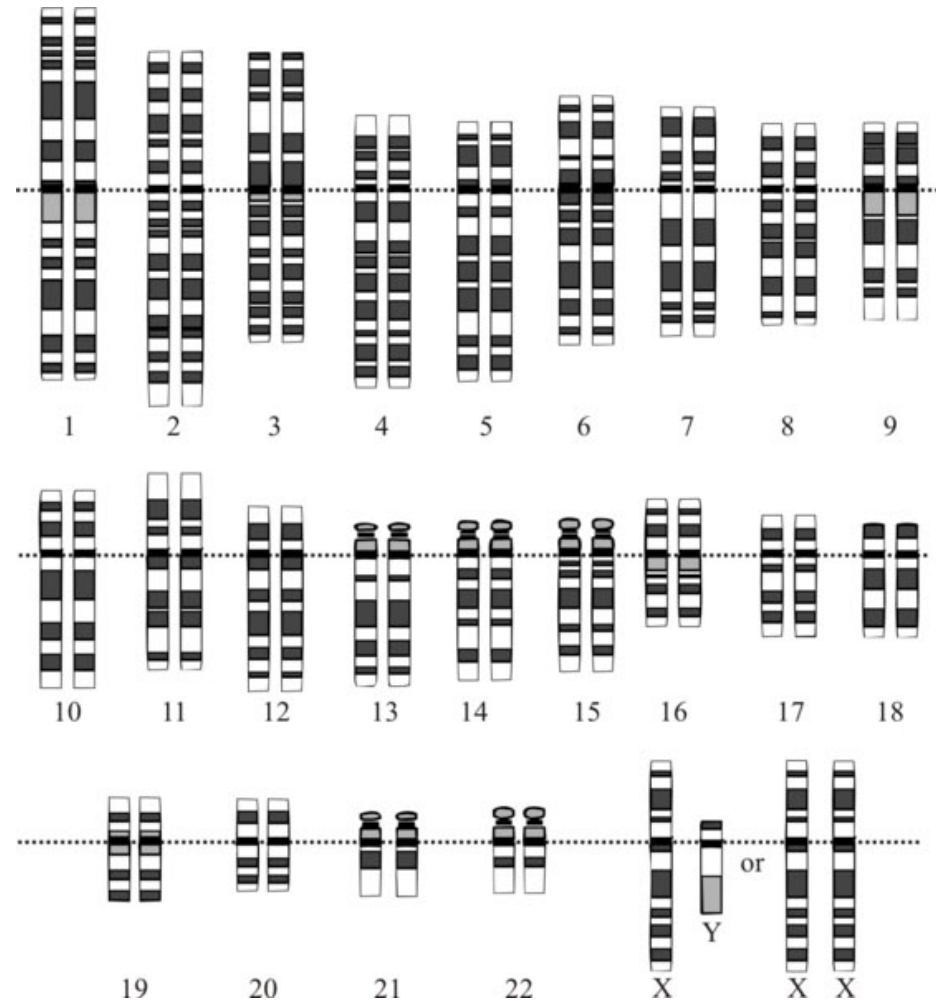


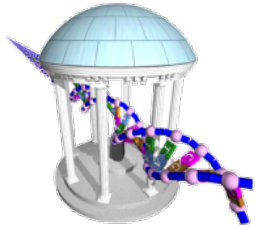


Diploidy



- Ploidy is the number of chromosome sets in a cell's nucleus.
- Cells with a ploidy of two are called diploid.
- Generally, one chromosome set is inherited from mom and the other from pop.
- Nearly all mammals are diploid

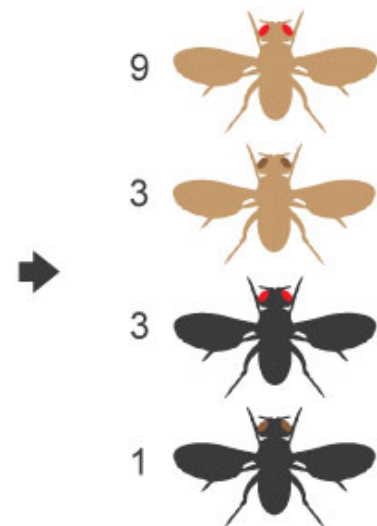
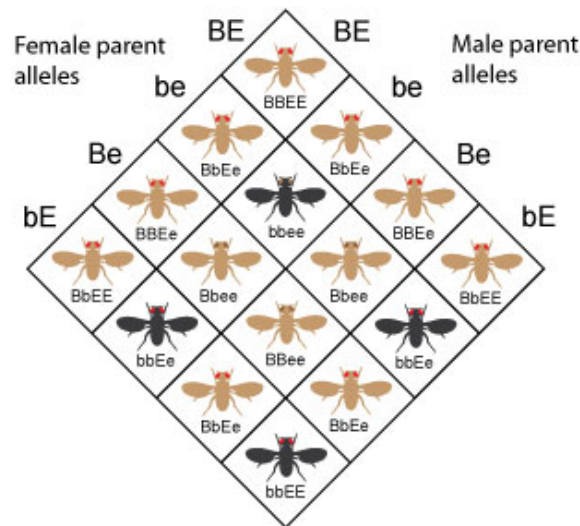


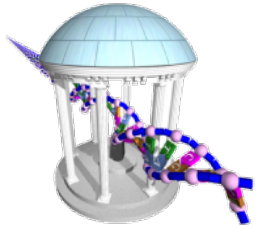


Genotype



- Genotype is the genetic state of an organism
- Genotyping decomposes genotype into one or more component alleles (genetic state variables)
- Since diploid organisms have two homologous copies of their state, genotyping provides *pairs* of alleles
- Genotyping can be in terms of any genetic feature including DNA fragment size, Microsatellite, Single Nucleotide Polymorphisms, repeat copy number, insertions, deletions, ...



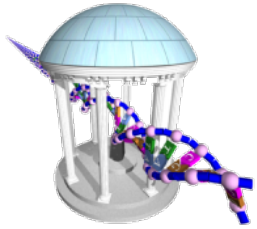


Binary Alleles



- Most Alleles come in two variants (e.g. wild, mutant)
- We will consider binary alleles as having one of two binary states 0 or 1
- And a diploid organism has two alleles which can be in one of 3 states: $[(0,0), (0,1), (1,1)]$
- Sometimes denoted:
 $[0,1,2]$

Note: For now we assume the chromosome origin of each order is unknown, thus $(0,1) \equiv (1,0)$



Allele Types



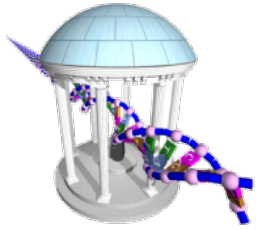
- An organism is *homozygous* when the same allele is present on all homologous chromosomes.
- A diploid organism is *heterozygous* when it has different alleles on its homologous chromosomes.
- A chromosome in a diploid organism is *hemizygous* when only one copy is present (e.g. X in males)

Allele-type notation:

Homozygous: AA or aa

Heterozygous: Aa

Hemizygous: A or a



Inherited Allele States

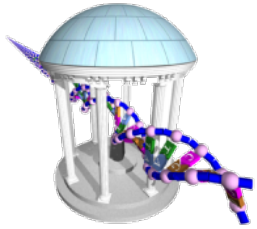


- Punnett squares: A diagram representing all possible outcomes of a cross.

	A	a
A	AA	Aa
a	Aa	aa

	A	a
A	AA	Aa
A	AA	Aa

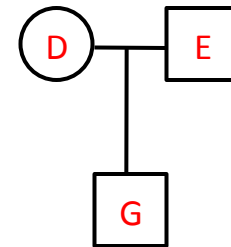
	a	a
A	Aa	Aa
A	Aa	Aa
A	A	A
A	AA	AA
A	AA	AA

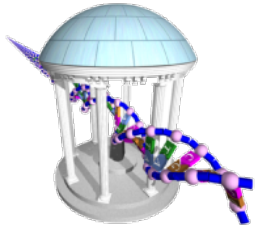


Genotype Phasing



- Phasing addresses questions of allele origin
- In other words, which allele was inherited from mom and which was inherited from pop
- This is a difficult problem given only a single genotype, but it becomes almost trivial when given a Trio pedigree.

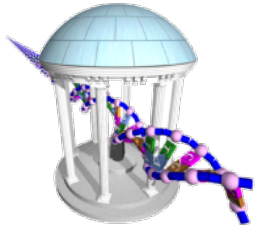




Trio Cases



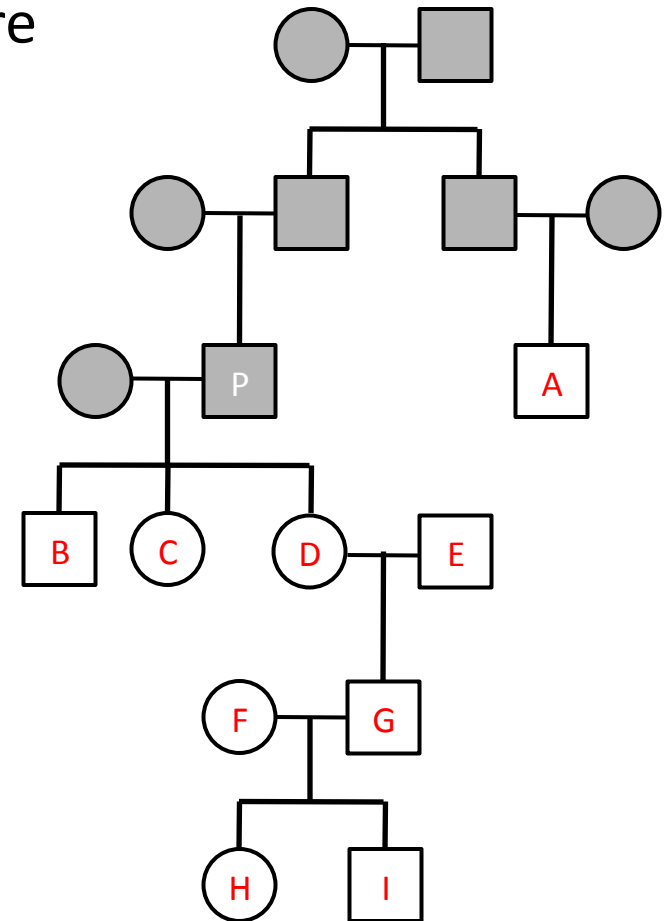
Maternal Allele	Paternal Allele	Child's Allele	Maternal Contribution	Paternal Contribution
AA	AA	AA	A	A
AA	aa	Aa	A	a
Aa	AA	Aa	a	A
Aa	AA	AA	A	A
AA	Aa	Aa	A	a
AA	Aa	AA	A	A
Aa	Aa	AA	A	A
Aa	Aa	aa	a	a
Aa	Aa	Aa	?	?

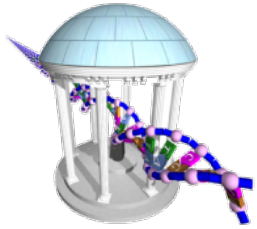


Concrete Example



- Consider the pedigree given last lecture
- Genotypes are available on-line
- Goto lecture web page for support
- Before the next class meeting write a phasing algorithm
 - Rewrite Ordered Alleles
 - Use lower case to indicate unphased alleles
 - How to treat No-Calls (impute or ignore)





Next Time



- Using allele sequence to infer missing phase state
- Recombination
- Mapping haplotype origin
- Inference in non-Trio relationships