Genetic Analysis



Dr. Phillip Carpenter medpathwaymcat

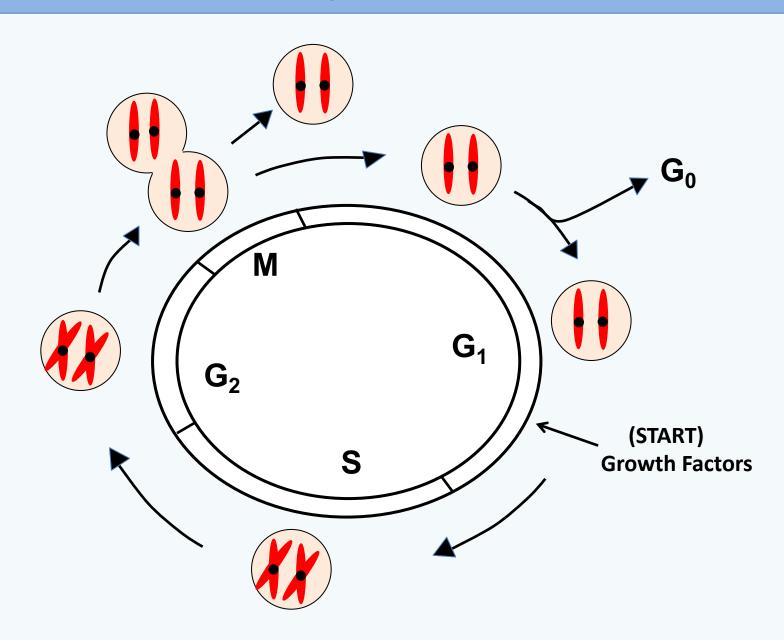


Med-pathway

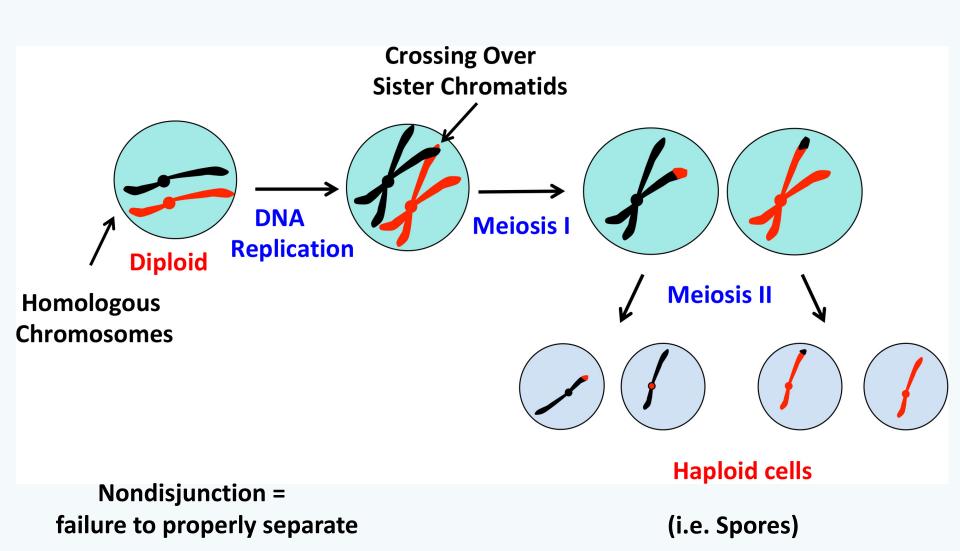


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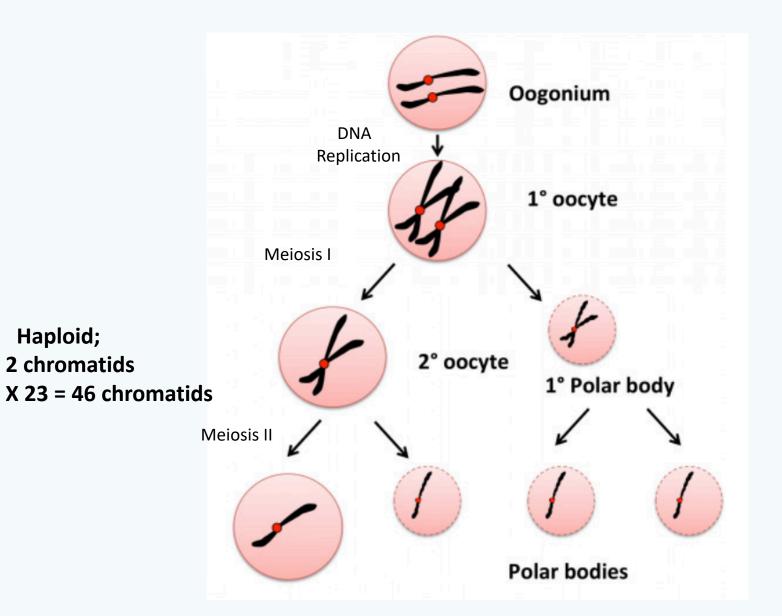
The Cell Cycle & Mitosis



Meiosis



Oogenesis



Haploid;

2 chromatids

Crossing Over

Occurs in Prophase I

Mendel

Examined inheritance of traits in peas

Discovered that traits were present in pairs of hereditary material called "alleles"

Law I: Segregation of alleles:

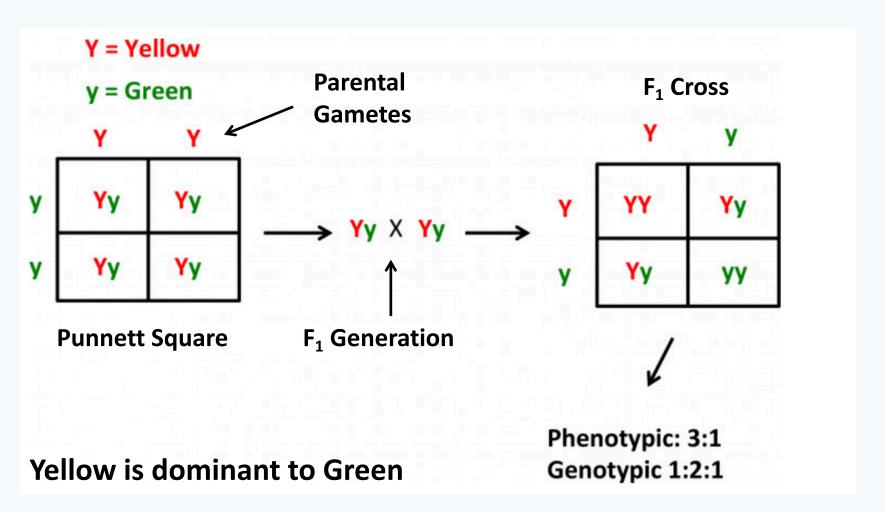
Alleles are individually transmitted from parent to offspring in a random manner

Law II: Independent assortment

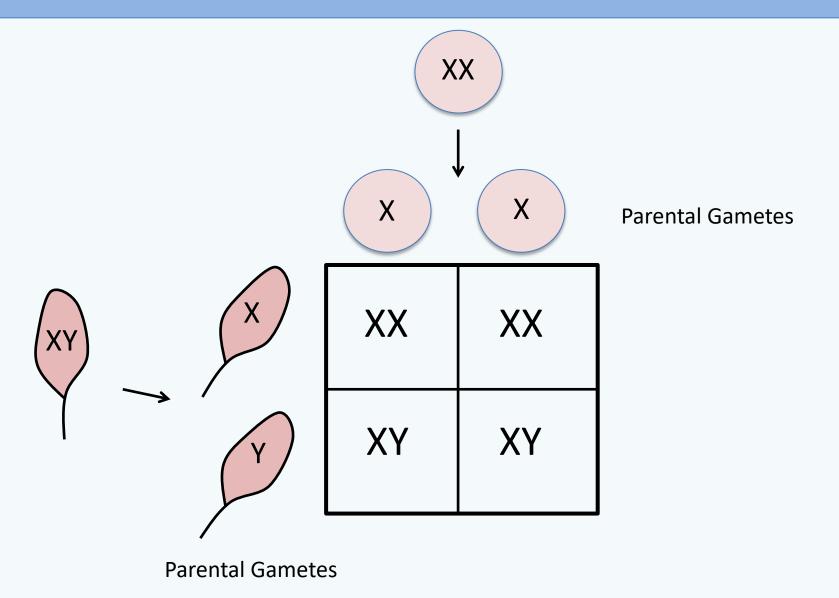
Alleles of one trait separate independently from alleles of a second trait. (Exception: Linkage of genes on same chromosome)

Predicting Crosses with Mendelian Laws

Pea Color: Yellow or Green



Sex Chromosomes

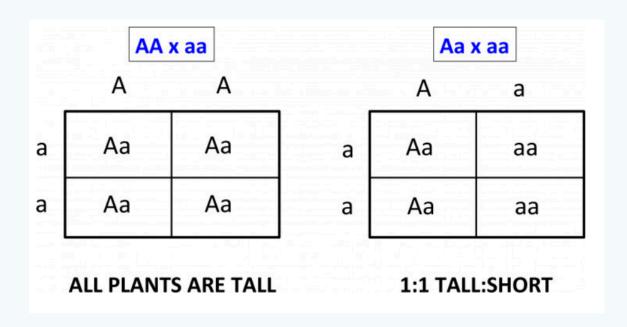


Test Cross

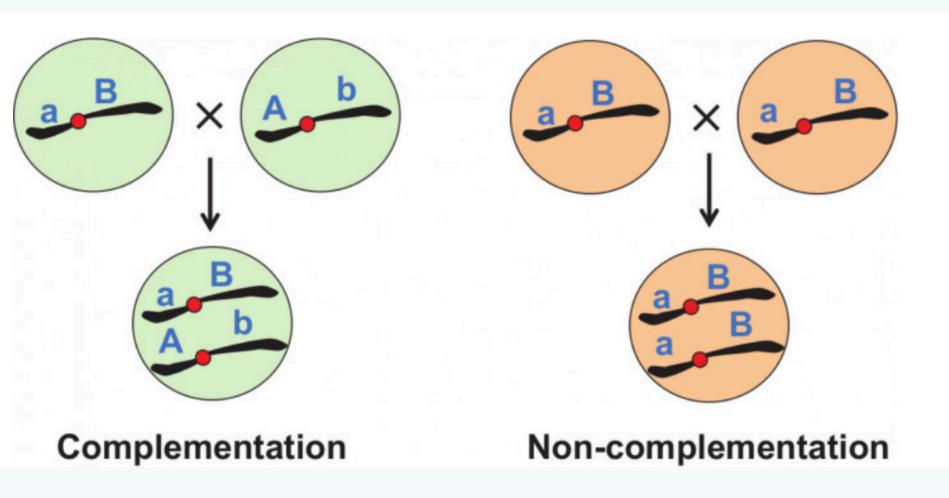
To determine the genotype of an unknown (i.e. Plant height: AA or Aa) Always includes a homozygous recessive.

Cross to a homozygous recessive (aa), Any recessive trait in the unknown will be revealed in the phenotype of the F_1 progeny.

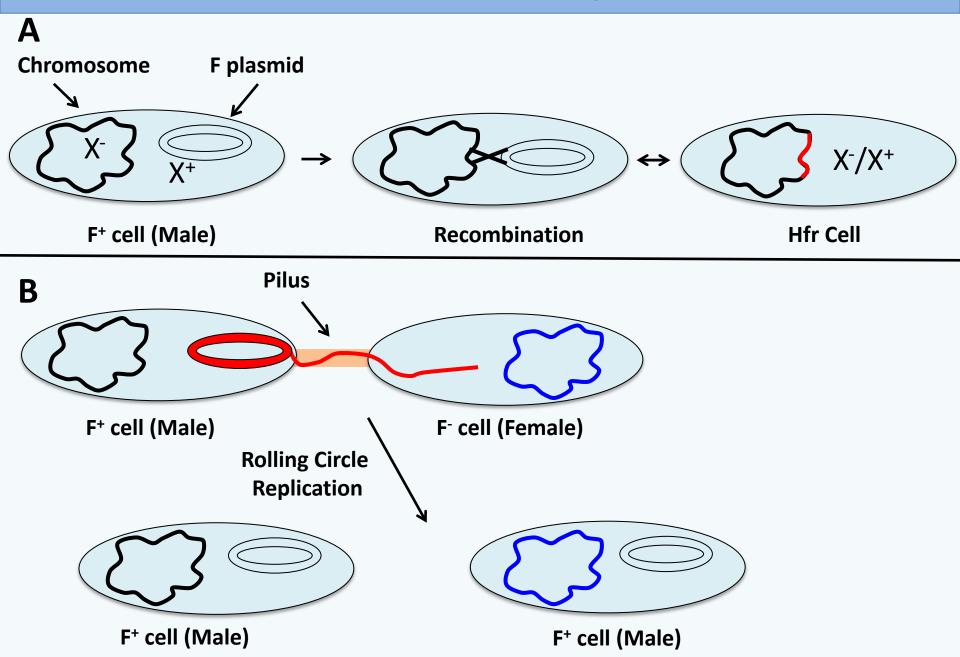
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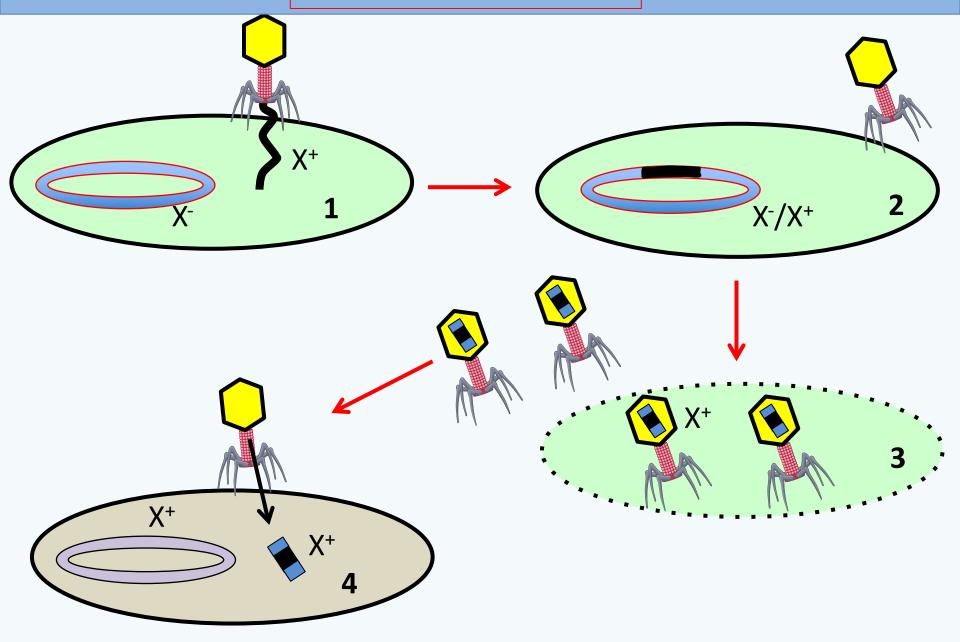
Complementation



Bacterial Partial Diploids



Specialized Transduction



Beyond Dominance & Recessiveness

Genotype Phenotype

LDL-R+/+ 120 mg/dL

LDL-R+/- 400 mg/dL

LDL-R-/- 700 mg/dL

Haploinsufficiency

RR = Red WW = White RW = Pink

Incomplete dominance (Blended)

Co-dominance (ABO Blood Type)

Each expressed regardless of status of other allele

More Complex Inheritance Patterns

Penetrance: Probability that a person with a genotype will express the corresponding phenotype

Pleiotropy: Gene alteration induces many aspects of phenotype

Polygenism: Traits influenced by numerous genes

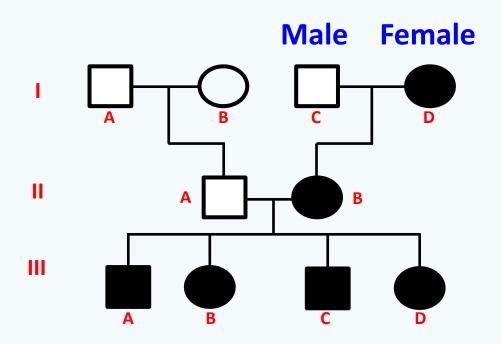
Epistasis: Interactions between genes where expression of one is dependent upon another

Baldness vs brown hair

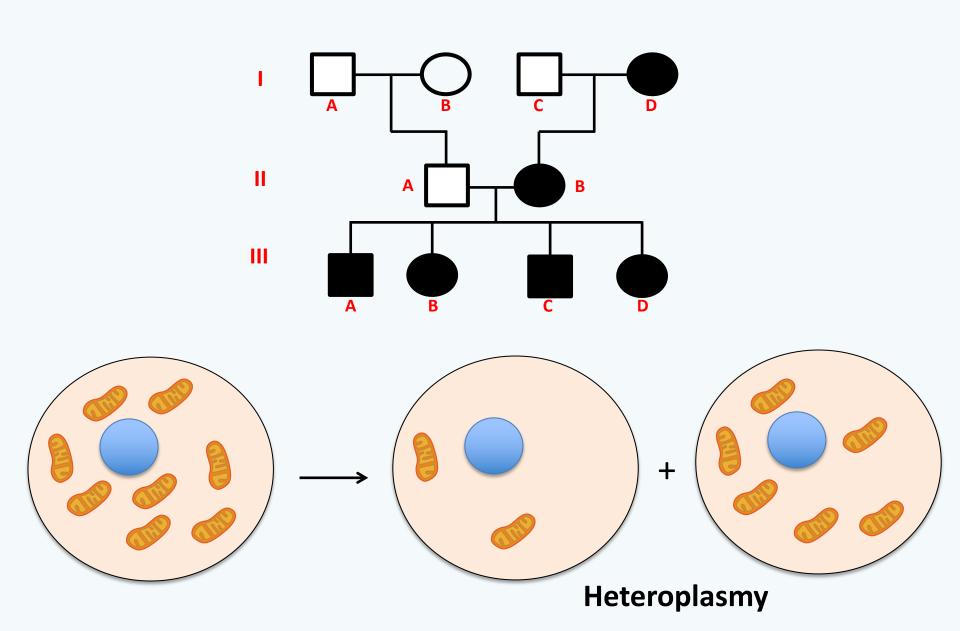
Pedigrees: Understanding Inheritance Patterns

- 1. Autosomal dominant: Sex Independent, AAD expresses phenotype (ADAD is lethal)
- 2. Autosomal recessive: Sex independent, skips generations
- Mitochondrial: Rare, Affected mothers pass on trait to males/females
- 4. X-linked Dominant: XX^D
- X-linked recessive: Skips generation, mostly affects males
- 6. Y-linked: Males only, inherited from affected father

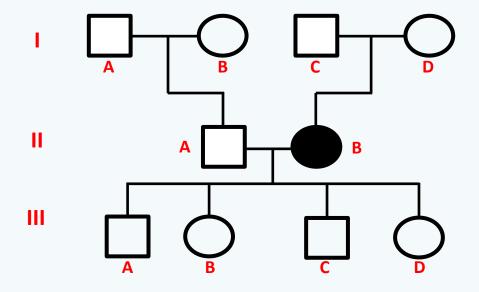
What type of inheritance?



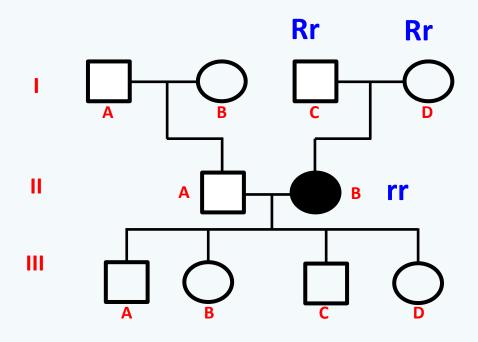
Mitochondrial Inheritance



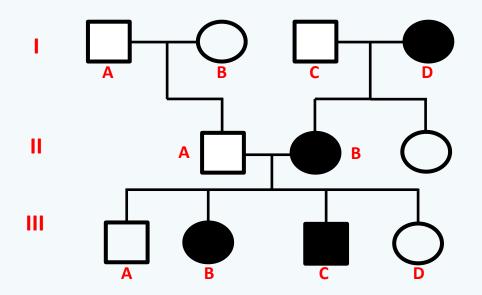
What type of Inheritance?



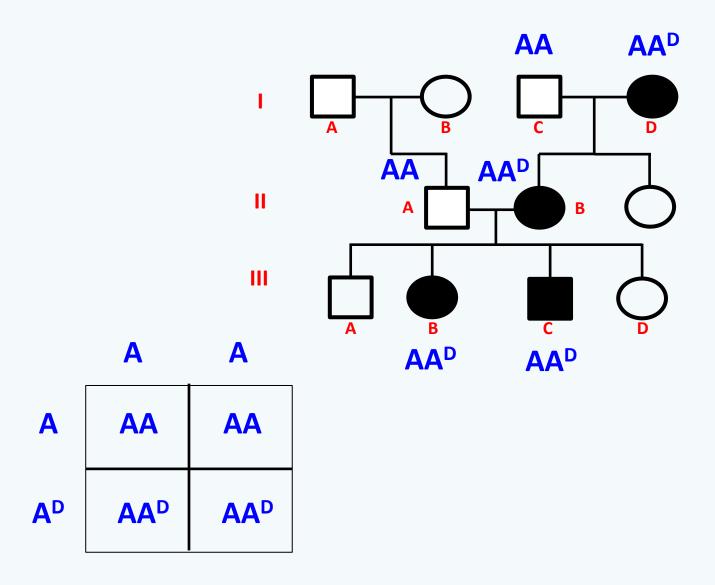
Autosomal Recessive



What type of Inheritance?



Autosomal Dominant



Rules of Probability

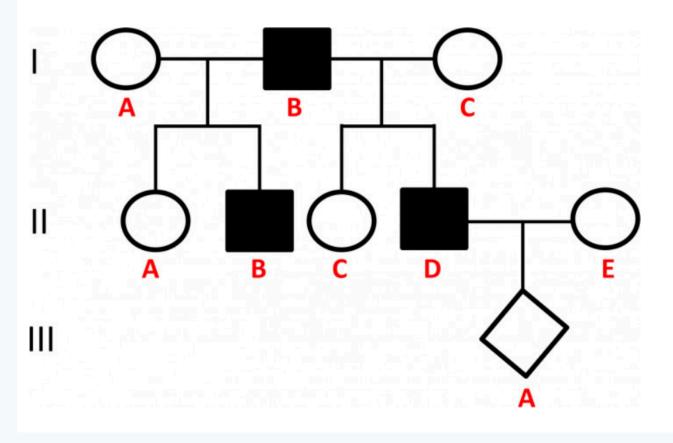
Rule of Multiplication: Probability of two events happening is the product of each event: $P = P(A) \times P(B)$

Rule of Addition: Probability of either of two events happening

$$P = P(A) + P(B) - P(A) \times P(B)$$

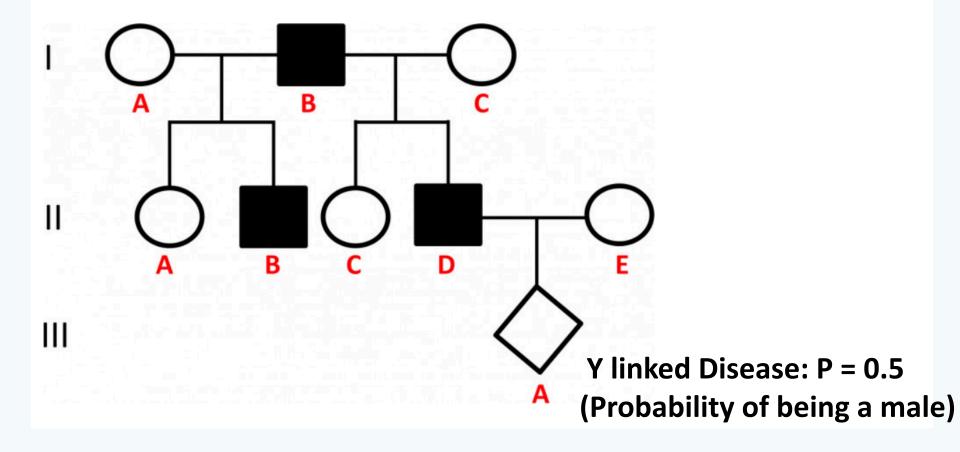
Pedigree Problem

A pedigree was constructed to understand the nature of transmission of a newly identified genetic disease. What is the probability that IIIA will be born with the disease?



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Population Genetics

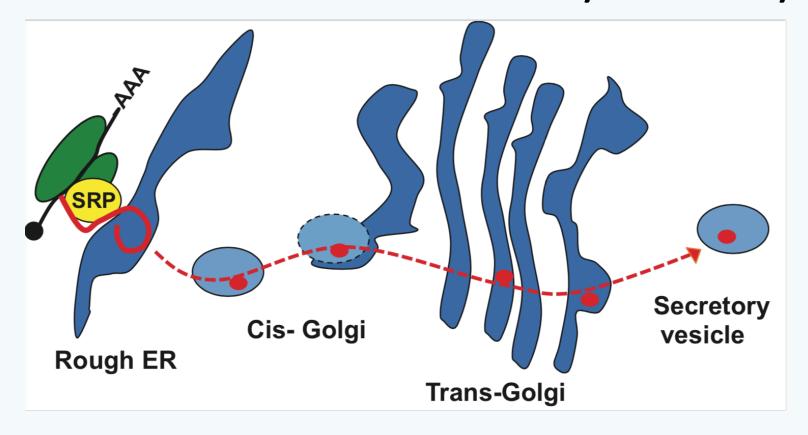
Gene Pool = Sum of genes in population

Hardy Weinberg: Frequency of alleles in population is constant provided that:

- 1. No net migration
- 2. No mutation
- 3. No natural selection
- 4. Random mating occurs
- 5. Population is large

Total # alleles: p + q = 1 or $p^2 + 2pq + q^2 = 1$ p and q = frequency of dominant, recessive alleles, respectively

Genetics of Yeast Secretory Pathway



Yeast: exist in haploid or diploid states

Temperature sensitive mutants: Permissive & Non-permissive states

