GENETICS-Chapter 14

Observable characteristic = **TRAIT**

Alternative choices for a gene = ALLELES

DOMINANT allele hides the recessive allele

RECESSIVE allele is hidden by dominant allele if it is present

HOMOZYGOUS organisms have two of the same alleles for a trait (EX: TT or tt)

HETEROZYGOUS organisms have two different alleles for a trait (EX: Tt)

PHENOTYPE= character that is expressed; "way it looks"

GENOTYPE = genetic makeup; "what genes it has"

P_1 (Parental)

↓

F_1 (Filial=offspring)

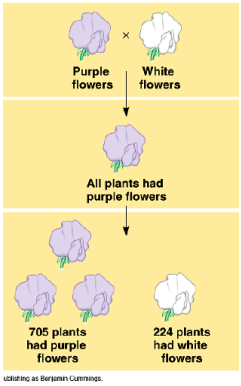
↓

F_2

TRUE BREEDING PARENTS in genetic cross = PARENTAL (P_1) generation

Their offspring = first filial generation (F_1)

Off spring of the F_1 generation = SECOND FILIAL generation (F_2)



MENDELIAN INHERITANCE

MONOHYBRID CROSSES – cross to study only ONE character

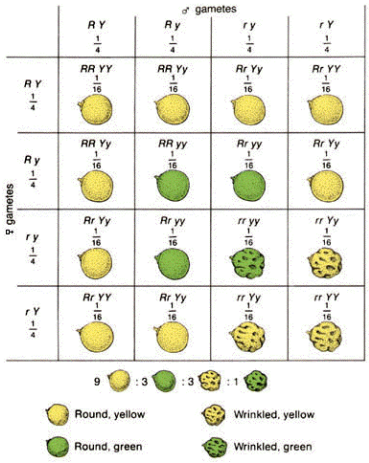
Aa X Aa = 3:1 ratio

LAW of SEGREGATION = two alleles separate during gamete formation (separation of homologous partners during ANAPHASE I)

DIHYBRID CROSS- cross to study TWO characters

LAW of INDEPENDENT ASSORTMENT= Each pair of alleles segregates (separate) independently in meiosis

Maternal and paternal chromosomes mix up in different combinations during gamete formation (ANAPHASE I)



AaBb X AaBb = 9:3:3:1 ratio

9 = dominant trait 1/dominant trait 2

3 = dominant trait 1/recessive trait 2

3 = recessive trait 1/dominant trait 2

1 = recessive trait 1/recessive trait 2

For TRIHYBRID crosses or if parents NOT HETEROZYGOUS:

Use MULTIPLICATION and ADDITION RULES to determine probability of phenotypes and ratios

Possible different gamete combinations = 2^n where n= number of chromosome pairs

3 pairs of chromosomes (AaBbCc) = 2^3 = 8 possible combinations

TESTCROSS-

Used to determine if individual with dominant phenotype is heterozygous or homozygous

Unknown genotype is CROSSED WITH HOMOZYGOUS RECESSIVE (A_ X aa)

NON-MENDELIAN INHERITANCE

1) **NATURE vs NURTURE** – environment influences phenotypic expression
   Siamese cats/Himalayan rabbits- darker fur on areas that are cooler
   Hydrangea flower color depends on soil pH
2) **SPECTRUM OF DOMINANCE**
   INCOMPLETE DOMINANCE- heterozygote = blended intermediate phenotype
      (EX: red X white four o'clocks; heterozygotes = pink
   CO-DOMINANCE- Both alleles are expressed at same time
      (EX: Roan horse has BOTH white and red hair; AB blood type)
3) **LINKED GENES**
   X-LINKED- carried on X chromosome EX: Color blindness; hemophilia; Duchenne Muscular Dystrophy
   Y-LINKED- carried on Y chromosome EX: Hairy pinnae; SRY=gene for "maleness"
4) **MULTIPLE ALLELE TRAIT-** More than 2 choices
   EX: A, B, O blood alleles produce A, B, O, or AB blood types
5) **POLYGENIC TRAIT**- trait determined by more than one gene
   EX: skin color, intelligence, eye color
   "bell curve"
6) **EPISTASIS-** Gene at one locus alters the phenotypic expression of a gene at another locus
   EX: Coat color pigment not deposited in hair without color gene;
7) **PLEIOTROPY-** one gene has multiple phenotypic effects
   EX: dwarfism; cystic fibrosis
8) **MULTIFACTORIAL**- Genetic component + environmental factors influence disease
   (EX: heart disease, diabetes, cancer, manic depression, schizophrenia)
   Not well understood; educate people about risk factors/promote healthy lifestyle

CHI-SQUARE (X^2) - Used to determine if observed results are significantly different from expected results
KNOW HOW TO USE FORMULA and HOW TO INTERPRET RESULTS

NULL HYPOTHESIS: "There is NO DIFFERENCE between observed and expected"

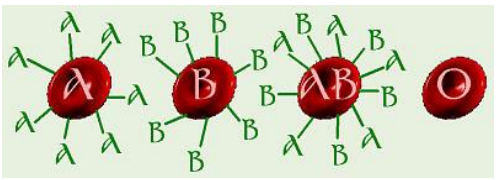
DEGREES OF FREEDOM = # of classes - 1

If X^2 < 0.05 p value; then difference can be due to random chance alone: "Accept null hypothesis"

$$X^2 = \sum \frac{(\text{observed} - \text{expected})^2}{\text{expected}}$$

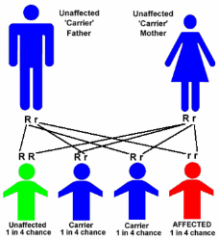
$\sum$ = the **sum of the values**
(the differences, squared, divided by the number of expected)

BLOOD TYPES:



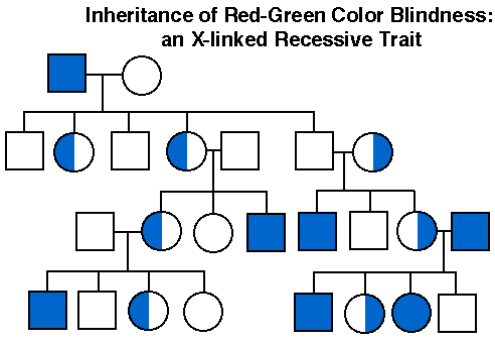
GLYCOPROTEINS on surface determine blood type
Important in transfusions/transplants
I^A and I^B are **CODOMINANT**; ii (type O) is recessive to A or B
Type O = **UNIVERSAL DONOR;** Type AB= **UNIVERSAL RECIPIENT**
Differences in Rh factor (Mom Rh^- and baby Rh^+) can result in
ERYTHROBLASTOSIS FETALIS

GENETIC SCREENING & COUNSELING



Basedon Mendialian genetics and probability rules
Tests for identifying carriers
Fetal testing
Newborn screening

CARRIER = Heterozyous individual that doesn't show trait, but can pass it on to offspring

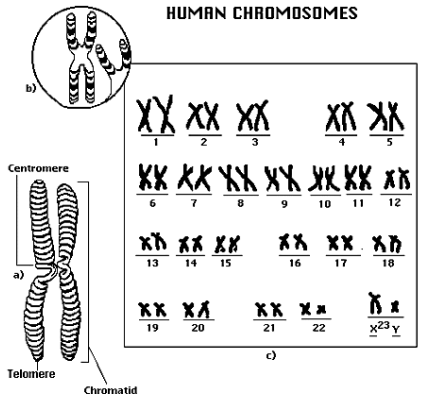


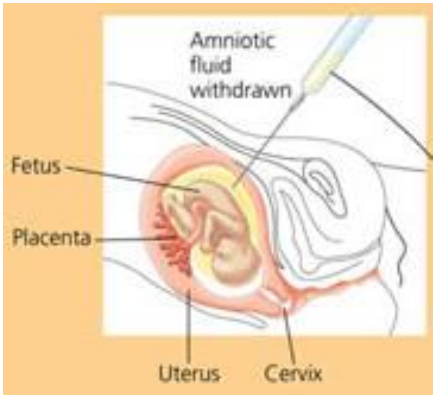
PEDIGREE-

Diagram that shows how traits are passed over generations;
Circles = females;
Squares = males;
Individual with trait= Filled in
CARRIER = half/half filled in

KARYOTYPE-

organizes picture of an individual's chromosomes
Can show sex: XX = female / XY = male
Can show some genetic disorders:
missing/extra chromosomes (Down, Turner, Klinefelter)
large translocations/deletions (Fragile X)
Can't show gene mutations (EX: PKU, sickle cell)





AMNIOCENTESIS

Can't be done until 14-16 weeks
Needle inserted through abdomen
Sample of amniotic fluid removed
Biochemical tests done immediately on fluid
OR later on cultured cells (EX: karyotype)
Takes weeks
Risk of miscarriage

CHORIONIC VILLUS SAMPLING (CVS)

Can be done sooner (8-10 weeks)
Suction tube inserted through cervix
Biochemical tests/karyotyping can be done on fetal cells immediately
Faster/ results within days
but greater chance of miscarriage than amniocentesis