GENETICS

History of genetics

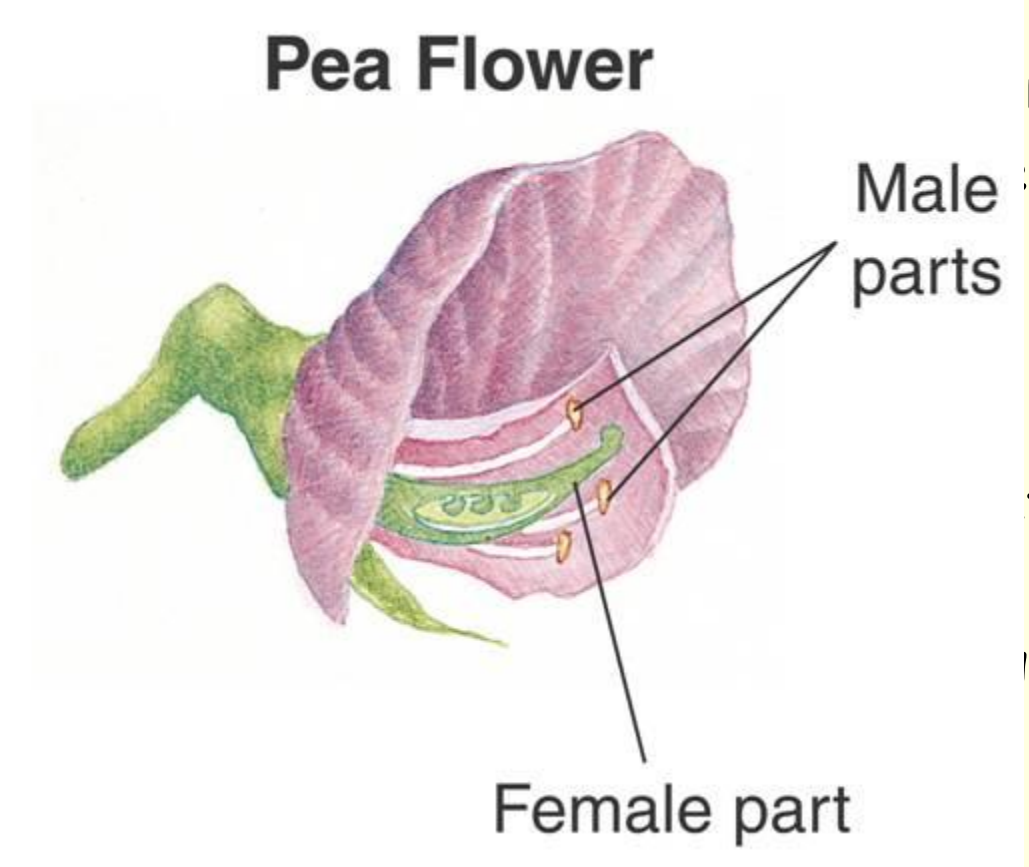
- Genetics- study of inheritance
- Trait- characteristic that can be passed from parent to child
  - Ex. Hair color, eye color, etc.
  - Ex. Pea plants- seed shape, height, flower color
- In the past, people explained heredity by saying that traits from parents were "blended" (blending hypothesis)
- We now know that during cell division, chromosomes are distributed through gametes

Mendel's experiments

- Gregor Mendel
  - Father of genetics
  - Austrian monk
  - Began work in 1860's
- Used 20,000 pea plants to study how traits are passed from one generation to the next
- Found that his results did not support the "blending hypothesis" (Why was this? How did he know it wasn't right?)

Mendel's experiments (cont.)

- Purebred
  - Org rents
  - Pea eding (spe
  - Also
- Hybrid
  - Org tic trait fro
  - Pea from diff
  - Also



Mendel's observations

- Started by crossing 2 different purebred plants (called parent or **P generation**) by cross pollination
- 1st generation of offspring (what he got from crossing two parent plants) called **F_1 generation**
  - All hybrid offspring only had the character of ONE of the parents (NO BLENDING!)
- Allowed F_1 plants to self-fertilize to produce **F_2 generation**

Mendel's observations (cont.)

Generation	Cross (pea color)	Results
P	*Yellow x Green*	*All yellow*
F_1	*Yellow x Yellow*	*3 Yellow* *1 Green*
F_2	*Differs*	*Differs*

Mendel's observations (cont.)

- Repeated experiment with 6 other traits
  - Flower color (purple/white)
  - Flower position (axial/terminal)
  - Pea shape (round/wrinkled)
  - Pod color (green/yellow)
  - Pod shape (inflated/constricted)
  - Height (tall/short)
- Defined each form as dominant (75% of F_2) or recessive (25% of F_2)

Gene

- Section of a chromosome that codes for a trait
- Allele
  - Different form of a gene (usually two)
  - Represented by a letter
  - Ex. B- brown eyed allele, b- blue eyed allele
- Two alleles make up a gene
  - Dominant
  - Recessive

Gene (cont)

- Dominant allele
  - Form of allele that is expressed (shown) when different alleles are present
  - Always represented by a CAPITAL letter
  - Ex. T- tall plant, P- purple flowers
- Recessive allele
  - Not expressed when the dominant allele is present (only when both are recessive)
  - Always represented by lower case letter that is the SAME as the letter that represents dominant allele
  - Ex. t-short plant, p- white flowers

Genotype

- Genetic make-up of an organism
- Included both genes in a homologous pair of chromosomes
- In the form of **LETTERS that represent genes**
- Ex. Bb (brown eyes), PP (purple flowers)

Phenotype

- Outward expression of a trait
- In the form of **WORDS that describe the trait**
- Ex. Green eyes, button nose, green pea color

	Genotype	Phenotype
Homozygous	TT	Tall (dominant)
Homozygous	tt	Short (recessive)
Heterozygous	Tt	Tall (hybrid) *Sometimes this could be a mix of two traits*

Example problem

- Ex. Flower color
  - Purple is dominant, white is recessive
  - Use P for dominant, p for recessive
  - Genotype for homozygous?
    - PP OR pp
  - Genotype for heterozygous?
    - Pp
  - Phenotype for homozygous?
    - Purple or white
  - Phenotype for heterozygous?
    - Only purple

Mendel's Laws

- **Law of Segregation**
  - When individual produces gametes, the copies of a gene separate so that each gamete receives only one copy (p. 265-266)
- **Law of Independent Assortment**
  - Alleles of different genes assort independently of one another during gamete formation (p. 270-271)
- **Law of Dominance**
  - Some alleles are dominant and some are recessive (p.264-265)

Mendel's Laws

- **Law of Segregation**
  - Only one characteristic can be found in a gamete
  - Ex. Blue eyes vs. brown eyes (can only have one of them)
- **Law of Independent Assortment**
  - For two characteristics, the genes are inherited independently of one another
  - Also called the "Inheritance Law"
  - Ex. If you had the genotype **AaBb** you would make four kinds of gametes: they would contain the combinations of either **AB**, **Ab**, **aB** or **ab**.

Mendel's Laws (cont.)

- **Law of Dominance-** When an organism has two different alleles for a trait (is a hybrid or heterozygous), the allele that is expressed is the dominant one
  - Ex. Blue eyes vs. Brown eyes (Bb- brown is expressed therefore it is dominant)

One trait (monohybrid)

- Punnett Square
  - Grid used for organizing genetic info and making predictions
  - When studying the inheritance of only one trait, called a **monohybrid cross**
  - For the first examples, we will only be testing the **complete dominance** condition (where one allele completely dominates over the other)

Rules for monohybrid crosses

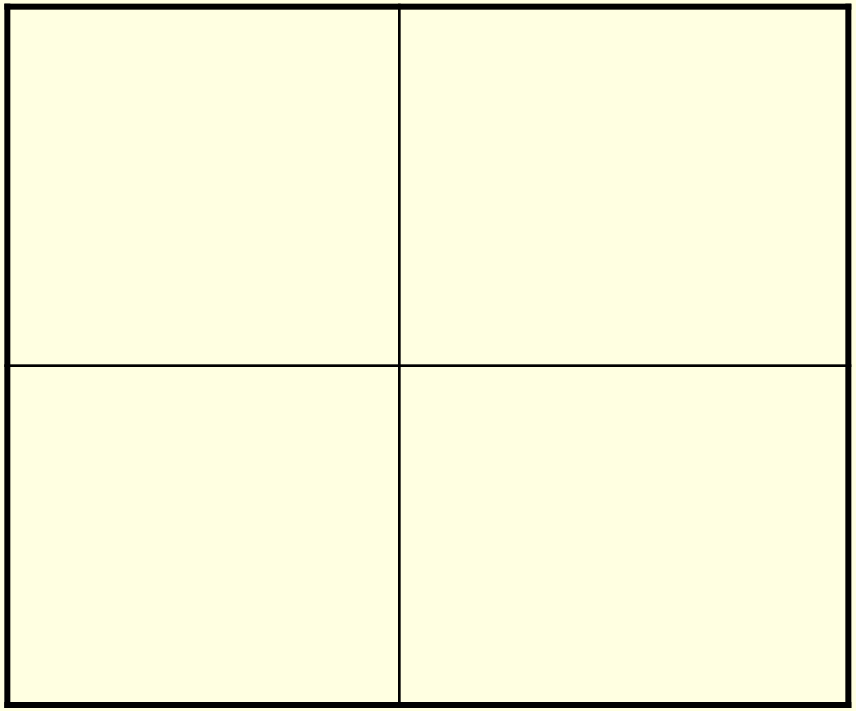
1. Determine parent genotypes (big letter for dominant, little letter for recessive)
2. Segregate alleles and place alleles of each parent on top and side of four-squared grid (mom's on one side and dad's on the other)
3. Combine parent alleles inside boxes (letters inside boxes show POSSIBLE genotypes of offspring- not ACTUAL)
4. Determine phenotypic ratio and possible genotypes in the following format:
   - Fraction, probability statement, phenotype (genotype)
   - Ex. $\frac{3}{4}$ cbetb (Could Be Expected To Be) purple (PP, Pp)

     $\frac{1}{4}$ cbetb white (pp)
- MUST BE LIKE THIS EVERY TIME!!!!

Example monohybrid cross

Yy x Yy

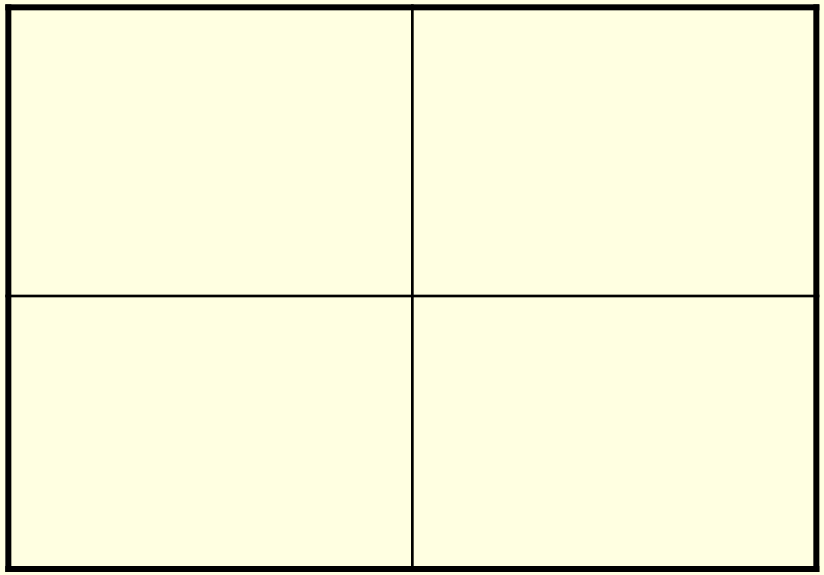
Y = yellow, y = green



Example monohybrid cross

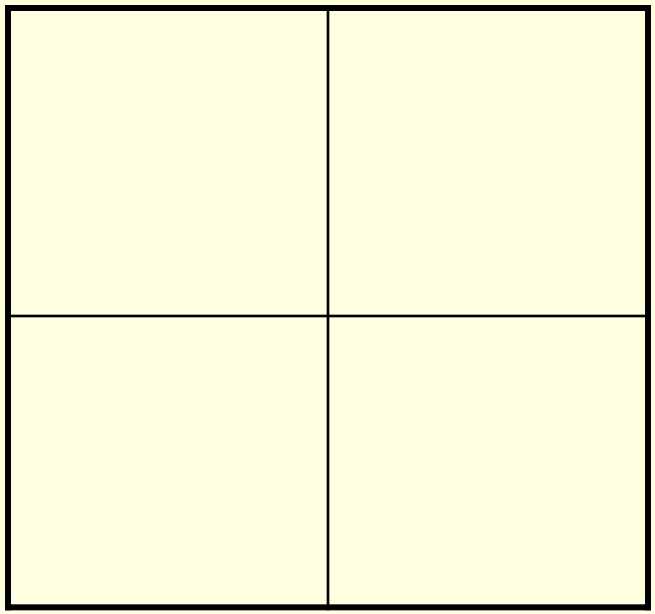
Ss x ss

S = smooth, s = rough



Example monohybrid cross

- Purple flowers are dominant over white flowers in pea plants. Cross a heterozygous purple plant with a white plant.
- Parent genotypes?
  - Pp
  - pp



Predictions for two traits

- Like tossing two coins at once (outcome of one doesn't affect outcome of the other)
- Involves two separate traits
  - How many letters will we use this time?
  - 2 different ones!!
- Called a dihybrid cross

Rules for dihybrid crosses

- Figure out all possible allele combinations
- Place combinations for both male and female on top and side of 16 square Punnett
- Combine parent alleles inside boxes
- Determine phenotypic ratios (no need to go through and do genotypes)

Practicing allele combinations

- AaBb —>
  - AB, Ab, aB, ab
- AABB —>
  - AB, AB, AB, AB (or just AB)
- aabb —>
  - ab, ab, ab, ab (or just ab)
- AAbb —>
  - Ab, Ab, Ab, Ab (or just Ab)

- aaBB —>
  - aB, aB, aB, aB (or just aB)
- Aabb —>
  - Ab, Ab, ab, ab (or just Ab, ab)
- aaBb —>
  - aB, aB, ab, ab (or just aB, ab)

Practice Dihybrid

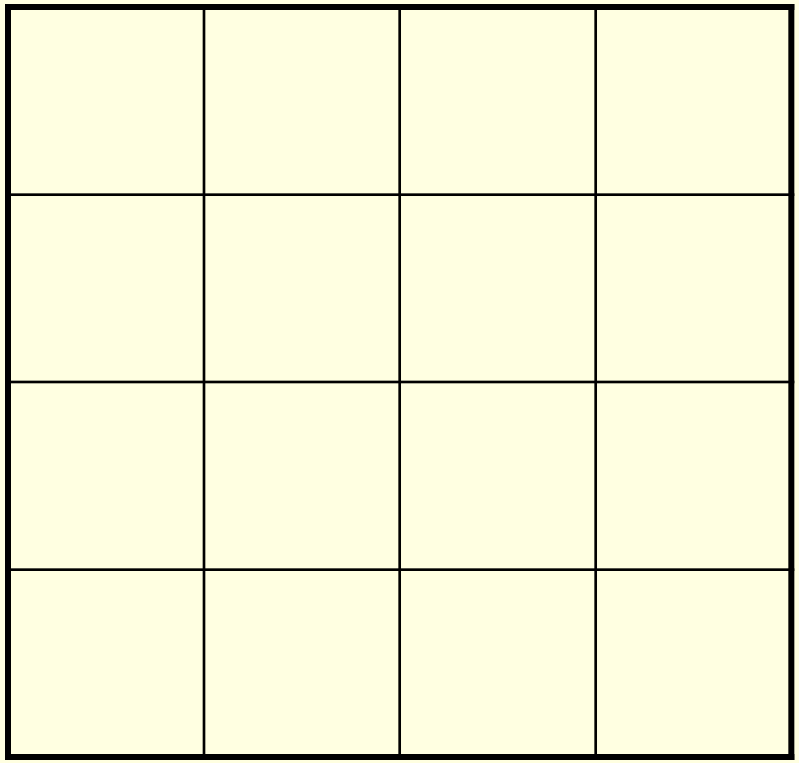
- RrYy x RrYy
- R = round, r = wrinkled
- Y = yellow, y = green

1. Figure out allele combinations
   - 1st plant?
     - RY, Ry, rY, ry
   - 2nd plant?
     - RY, Ry, rY, ry

Practice Dihybrid

2. Place on top and side of Punnett square/Fill in boxes

3. Figure out phenotypic ratios



Other patterns of inheritance

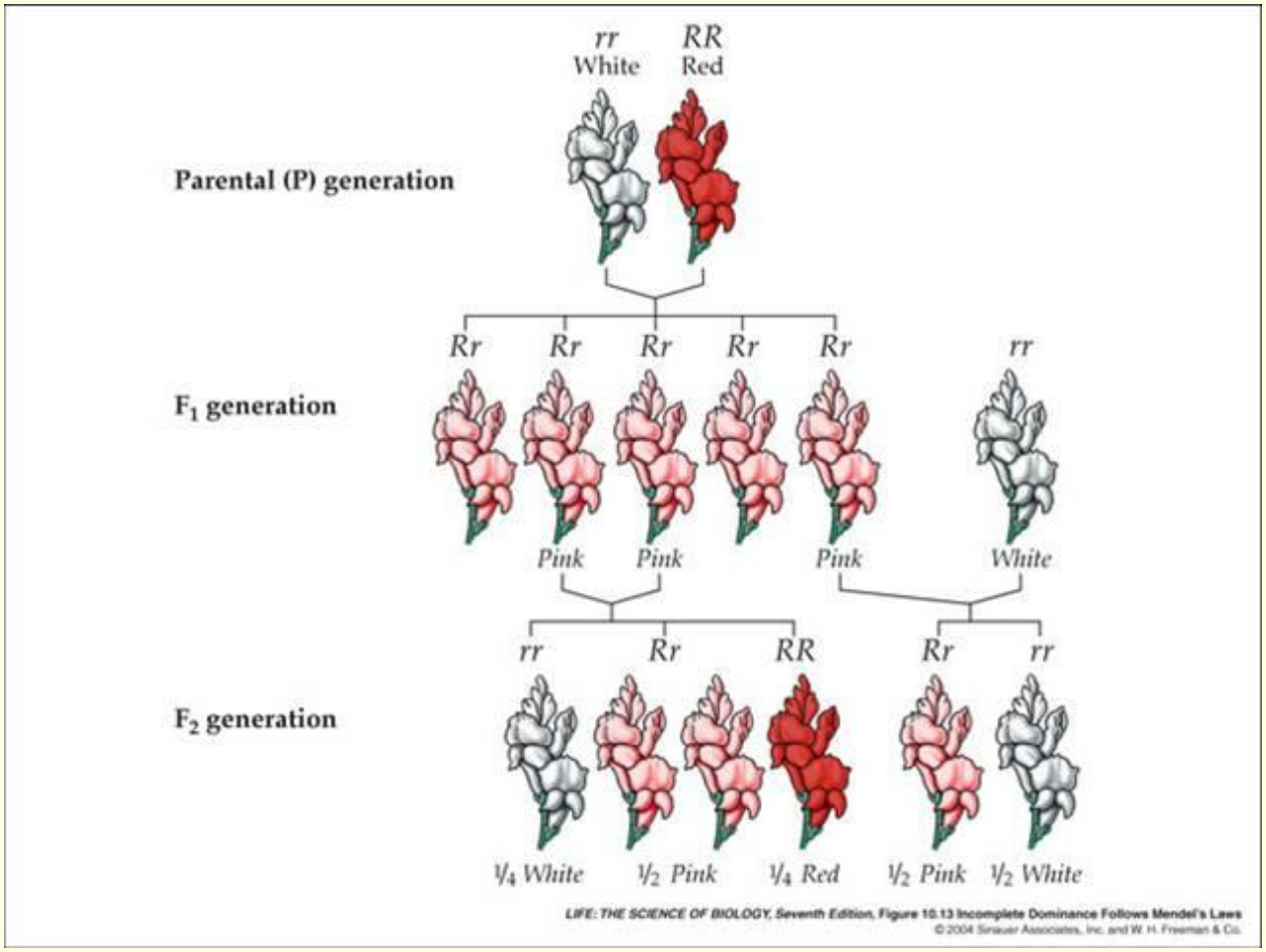
- It's not all as simple as I've made it out to be!
- Not all traits are completely dominant
- Several other patterns of inheritance
  - Incomplete dominance
  - Codominance
  - Sex-linked
  - Polygenic traits
  - Multiple alleles
  - Pleiotropy

Incomplete dominance

- Pattern of inheritance in which heterozygous offspring show a phenotype between the phenotypes of the parents (somewhere in the middle)
- Neither allele expressed fully
- Ex. Flower color in snapdragons
  - Red flower + white flower = pink flower
- Ex. Cow color
  - Red (brown) bull + white cow = roan (pink) cow

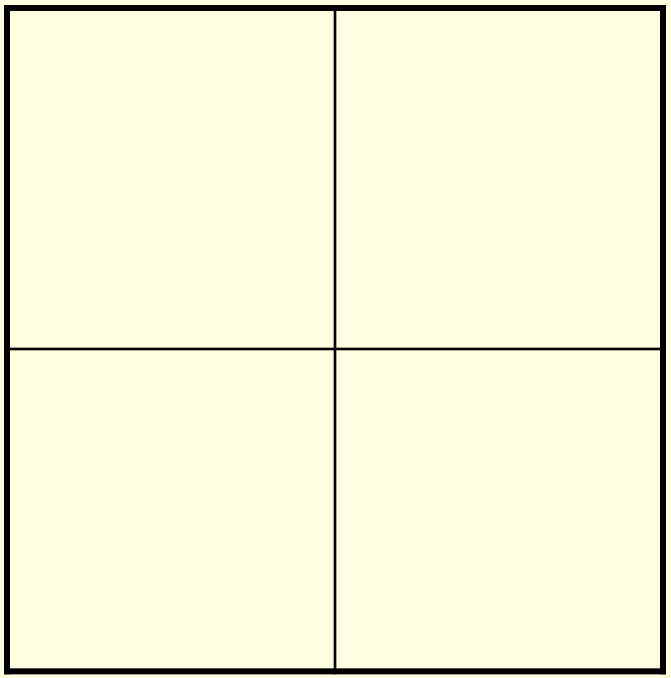


Incomplete dominance example



Example Punnett- Incomplete Dominance

- Knowing that four o'clock flowers show a pattern of incomplete dominance, create a Punnett showing a cross of a two pink four o'clocks (r= red, w= white).

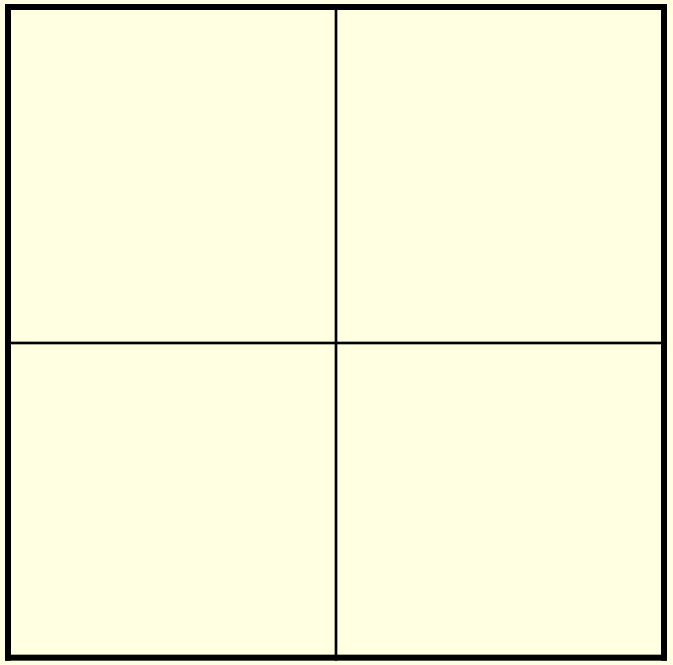


Codominance

- Pattern of inheritance where both alleles in the heterozygous offspring are fully expressed
- Ex. Human blood type
  - Type = Phenotype, Letters = genotype
  - Type A: AA, AO (homo and heterozygous)
  - Type B: BB, BO (homo and heterozygous)
  - Type AB: AB (Only heterozygous)
  - Type O: OO (Only homozygous)

Example Punnett- Codominance

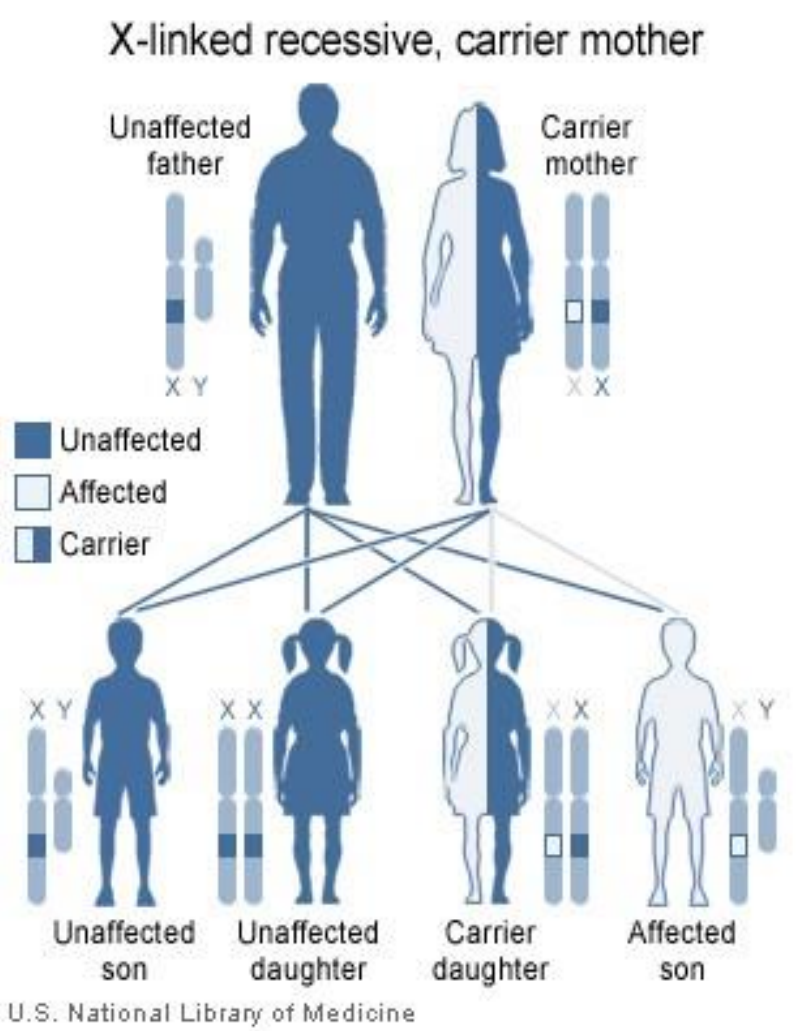
- Knowing that blood type shows a pattern of codominance, cross a person with type O blood and one with type AB blood.



Sex-Linked Pattern of Inheritance

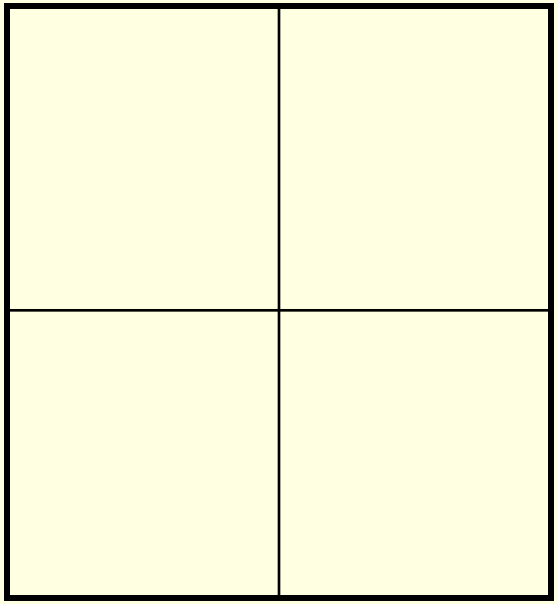
- Phenotypic expression of an allele that is dependent on the gender of the individual
- Carried on either sex chromosome (X or Y)
  - Remember: Men = XY, Woman = XX
  - Many more genes carried on the X chromosome, so many more X-linked traits than Y-linked traits
    - Ex. Hemophilia, color-blindness
    - If have one healthy X, it dominates over the infected X (in females)
    - If have only one infected X, Y can't dominate over it

Sex-Linked Inheritance Pattern



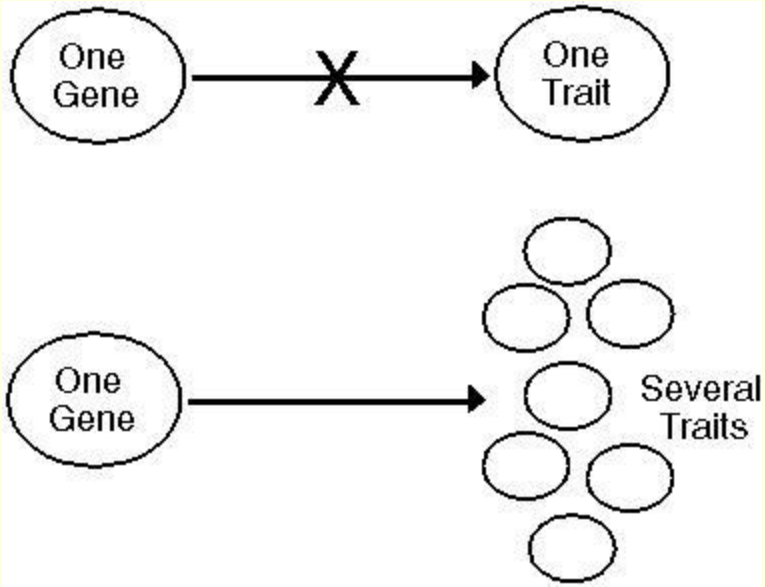
Sex-linked Example

- Knowing that color blindness is a sex-linked trait, cross a carrier female with a non-infected male and determine the probability of this couple having a color blind child.



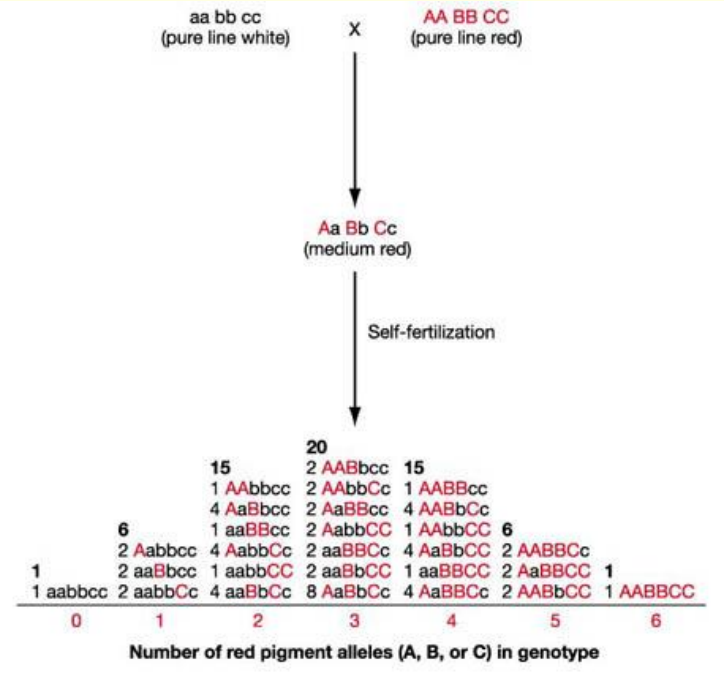
Pleiotropy

- Single gene affects more than one trait
- Ex. Sickle cell disease, Marfan's syndrome



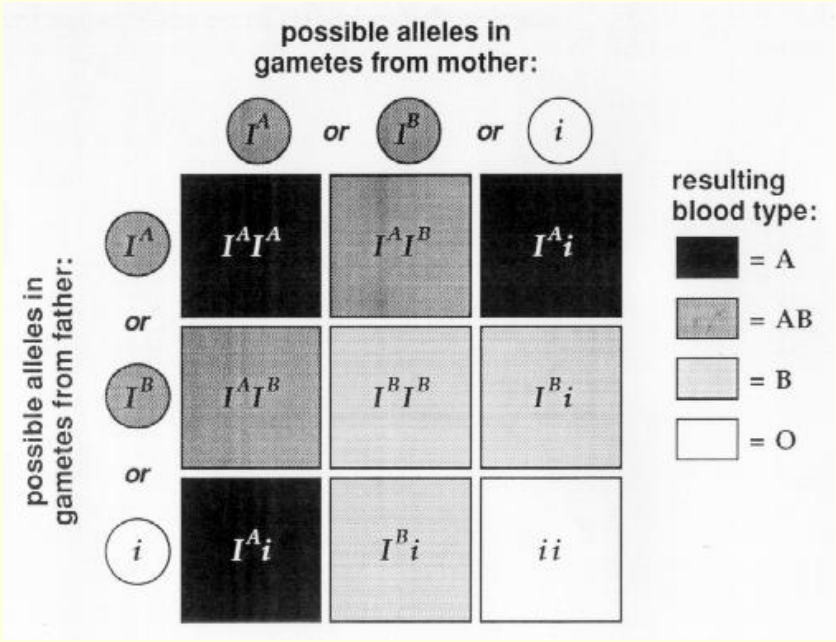
Polygenic traits

- One trait controlled by two or more genes
- Ex. Human skin colors



Multiple alleles

- More than two alleles for the same gene
- Ex. Human blood type (phenotypes produced by 3 different alleles)

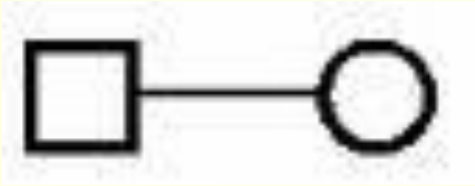


Pedigree

- Most knowledge of human genetics comes from studying patterns of heredity in populations and families
- The best way to do this is by creating a pedigree
  - Chart that shows how a trait and the genes that control it are passed through a family

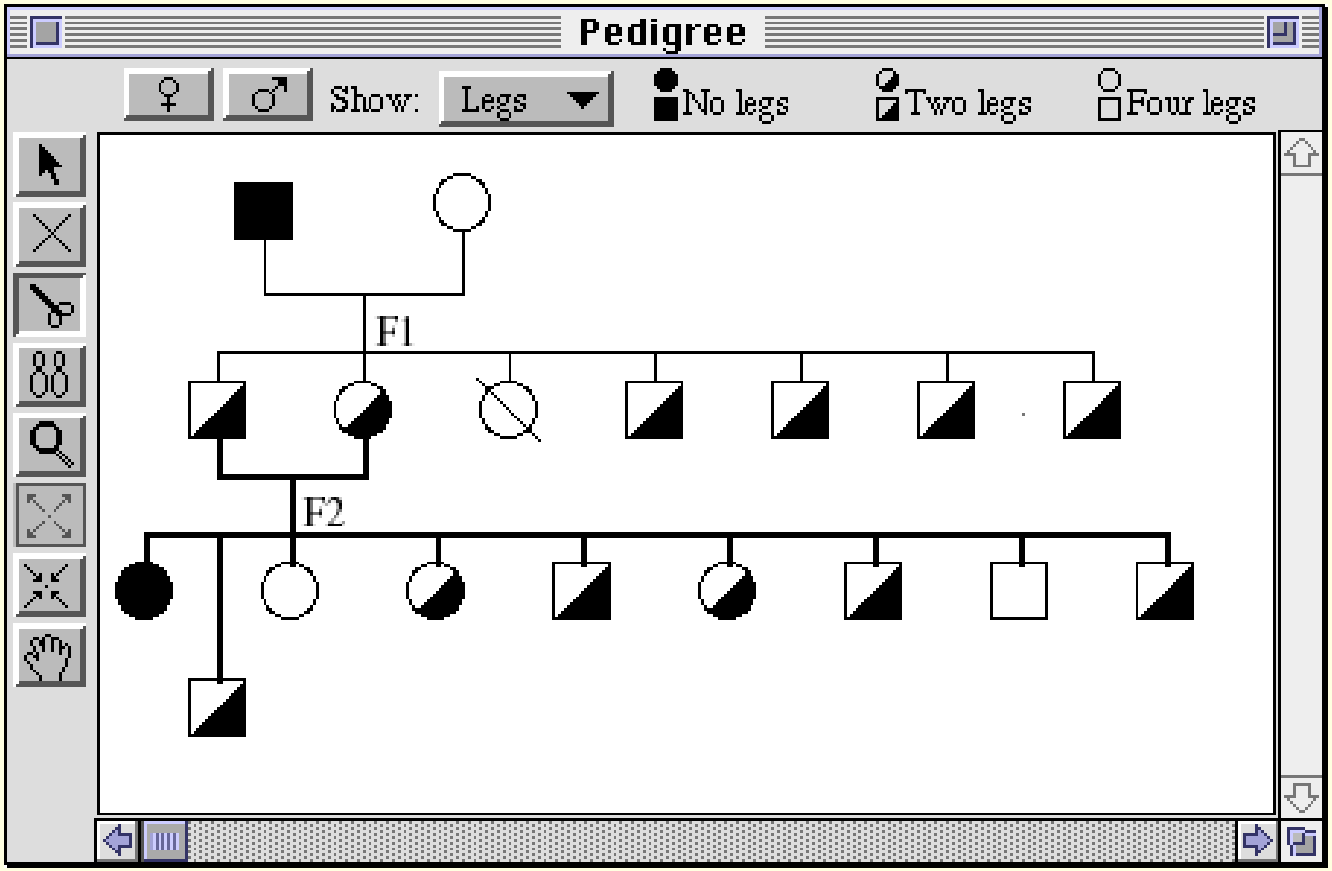
Pedigree (cont.)

- Circle = female
- Square = male

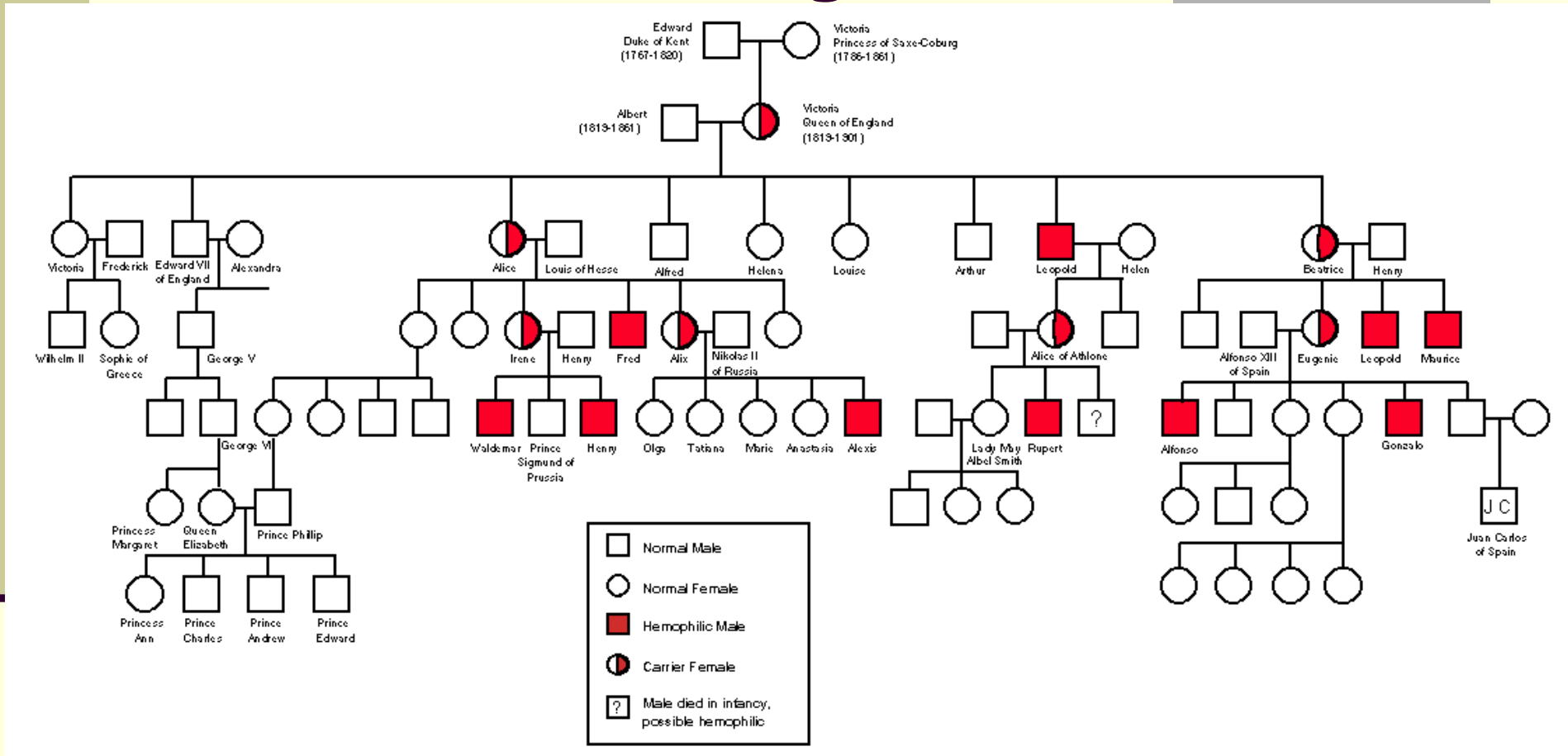


  - Empty circle: doesn't have trait (non-carrier)
  - Half shaded: carrier of trait (heterozygous individual that carries a recessive trait but doesn't express it- not shown)
  - Completely shaded: shows trait

Pedigree example



British Royal Family- Hemophilia Pedigree



Recessive Traits

- Carrier's not affected by recessive trait
- Can pass trait on to offspring
- In case of diseases, doesn't have the disease but can give disease to their children (How would this happen?)

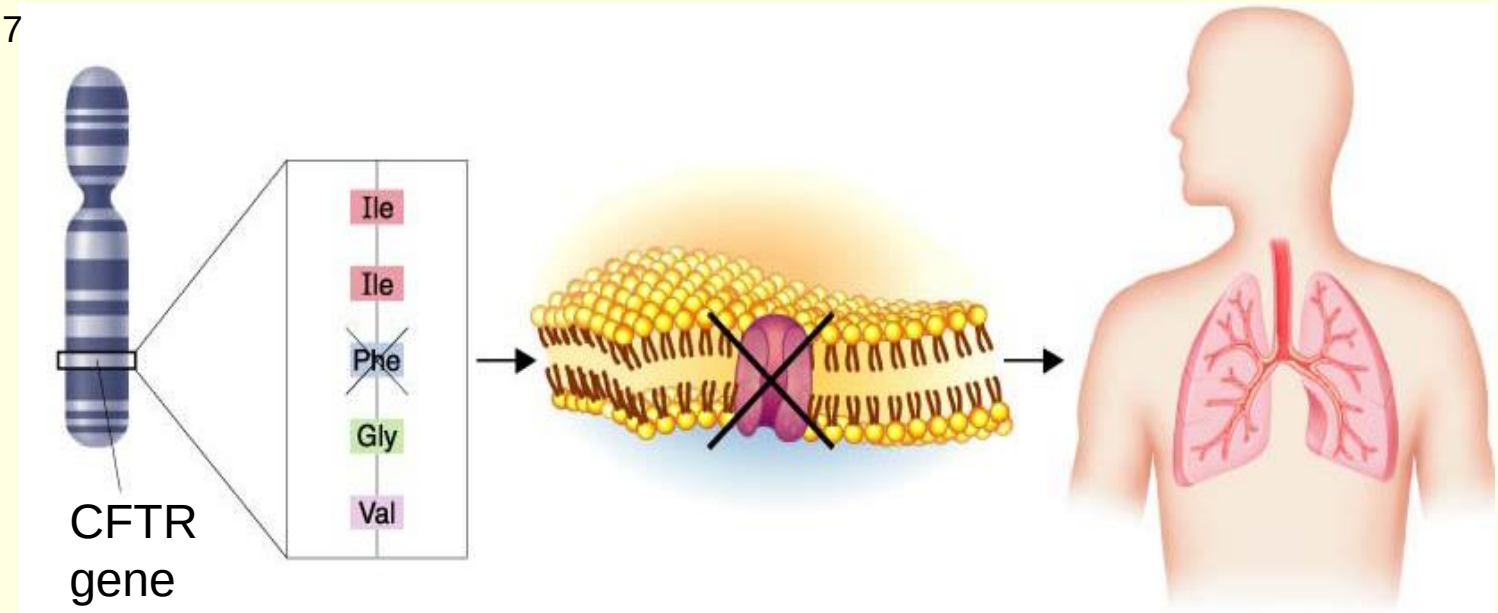
Tay-Sachs

- Recessive
- Fatal genetic disorder
- Causes fat build-up in the brain
- Found mostly in the Jewish community (Why is that?)

Cystic Fibrosis

- Recessive
- Fatal disease (mostly in chilo
- Causes excessive secretion of thick mucus (can't break it down)
- Found mostly in Caucasian community

Chromosome # 7



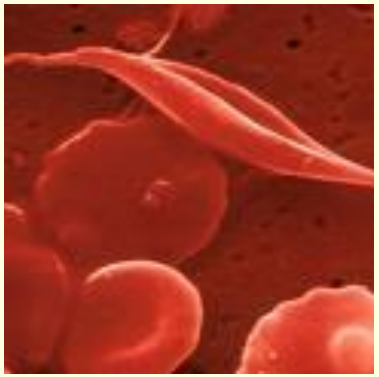
A The most common allele that causes cystic fibrosis is missing 3 DNA bases. As a result, the amino acid phenylalanine is missing from the CFTR protein.

B Normal CFTR is a chloride ion channel in cell membranes. Abnormal CFTR cannot be transported to the cell membrane.

C The cells in the person's airways are unable to transport chloride ions. As a result, the airways become clogged with a thick mucus.

Sickle Cell Anemia

- Recessive
- Causes red blood cells to form into sickle shape (half moon)
- Can't carry oxygen well
- Found mostly in the African-American community



Albinism

- Recessive
- True albinos have no skin pigm
- Can't produce melanin
- Can affect people of all races

Dominant Traits

- Always expressed in person's genotype
- Freckles, widow's peak, brown eyes
- Dominant diseases less common than recessive because person usually dies before passing on the trait

Polydactyly

- Dominant
- Extra fingers and/or toes
- Usually removed at birth

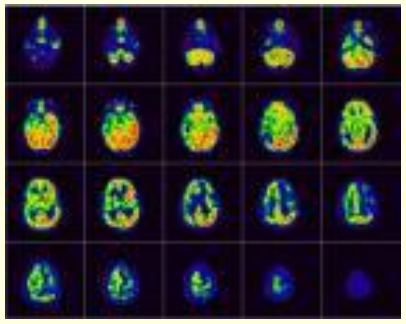

19/03/201

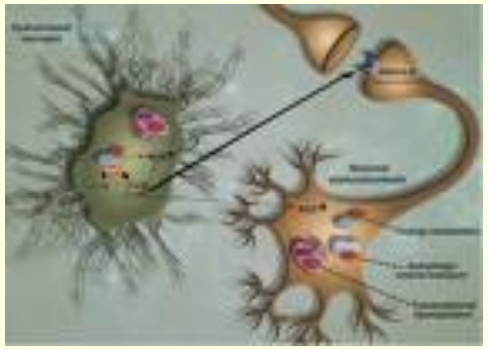

19/03/2010


19/03/2

Huntington's Disease

- Dominant
- Results in deterioration of the nervous system
- Don't usually develop symptoms until 30's or 40's





Achondroplasia

- Dominant
- Bone growth development disease
- Results in dwarfism

Genetic Counseling

- More people aware of genetic problems
- Can have DNA tested to determine if they are carriers for most diseases
- VERY EXPENSIVE!!!!!!!!!!