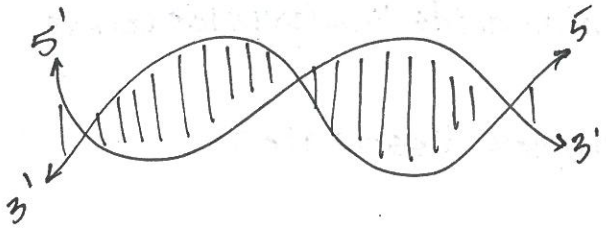
Genetics Unit:

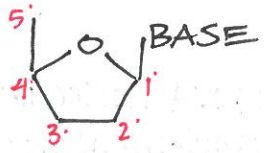
Part I: "The Stuff"

DNA: Deoxyribonucleic Acid



(PHOSPHATE)

POA



(SUGAR)

- DNA is an antiparallel double helix molecule of linked nucleotides.
- Wrapped around histone proteins, that complex wraps around itself multiple times to form the chromosome structure.
- **Replication: DNA to DNA**
  - Helicase & Topoisomerase: unzip + stabilize
  - RNA Primase: adds RNA primer on 5' end of new strand
  - DNA Polymerase: adds complementary DNA nucleotides (5'→3')
  - Leading Strand: 5'→3' into the replication fork
  - Lagging Strand: 5'→3' away from repl. fork (must jump back)
  - Ligase: seals nucleotides (phosphodiester bonds)
  - Nuclease: checks/cuts out mistakes

Transcription (DNA to RNA):

- Initiation: RNA polymerase finds the TATA box on the DNA
- Elongation: RNA poly. adds complementary RNA nucleotides to the DNA strand (5'→3' copy)
- Termination: Termination sequence is read and mRNA is released
- mRNA modifications post-transcription: 5' cap, poly-A tail (3' end), spliceosomes cut out introns leaving only the exons

Translation (RNA to Protein):

- Initiation: Ribosome reads the mRNA (AUG = start codon)
- Elongation: tRNA molecules (anticodon and amino acid) bind with complementary mRNA codons. Amino acids link (peptide bonds) forming a polypeptide chain.
- Termination: Stop codon is read and the complex dissociates
- Polypeptide modifications post-translation:

1°: ~~~~~

2°: ∿∿∿ and ⌇⌇⌇⌇

3/4°:  = Protein

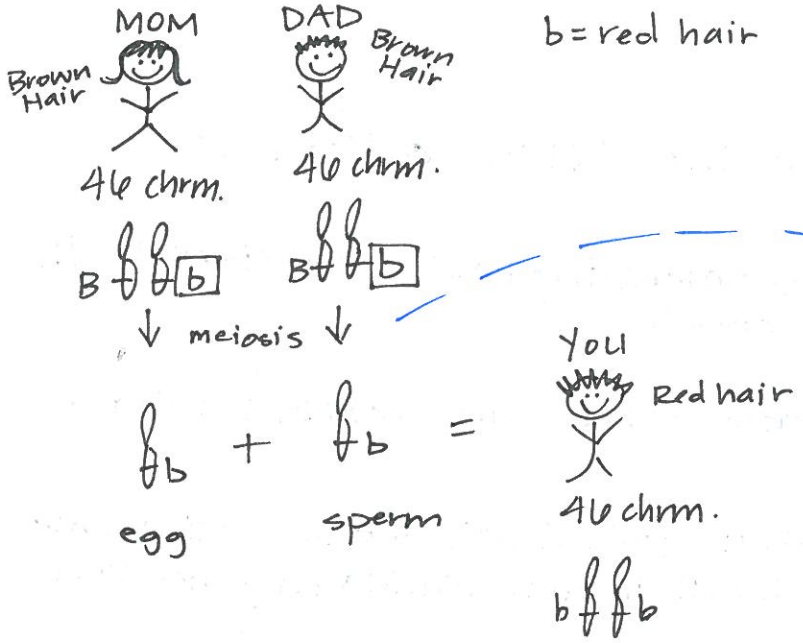
Part II: "How You Get the Stuff"

Inheritance

Humans (you) are diploid. You have 2 copies of each chromosome, meaning you have two copies of each gene. So of your 46 chromosomes, 23 came from mom and 23 came from dad. These matching sets of chromosomes are homologous meaning they are the same size and code for the same genes. However, they are not perfect copies. Different versions of the genes (called alleles) can be found on homologous chromosomes. Think about genes, alleles and diploid cells as you work through the problem below.

QUESTION: You mom has brown hair. Your dad has brown hair. You have red hair. Is this even possible? Justify your answer.

B = brown hair
b = red hair



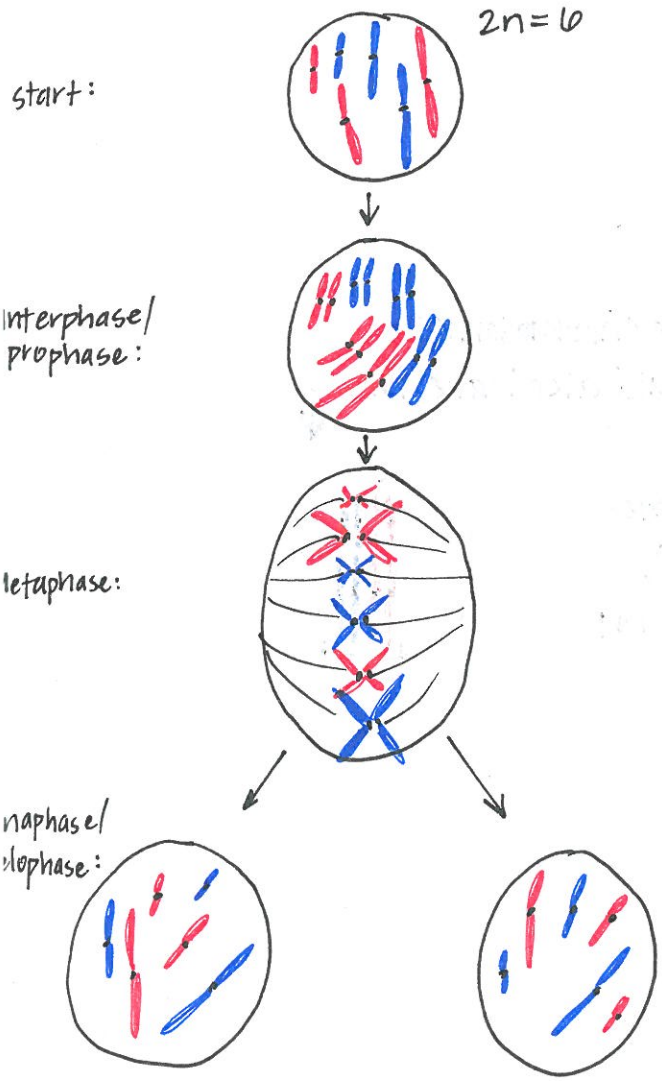
Law of Segregation

→ You have 2 copies of each gene

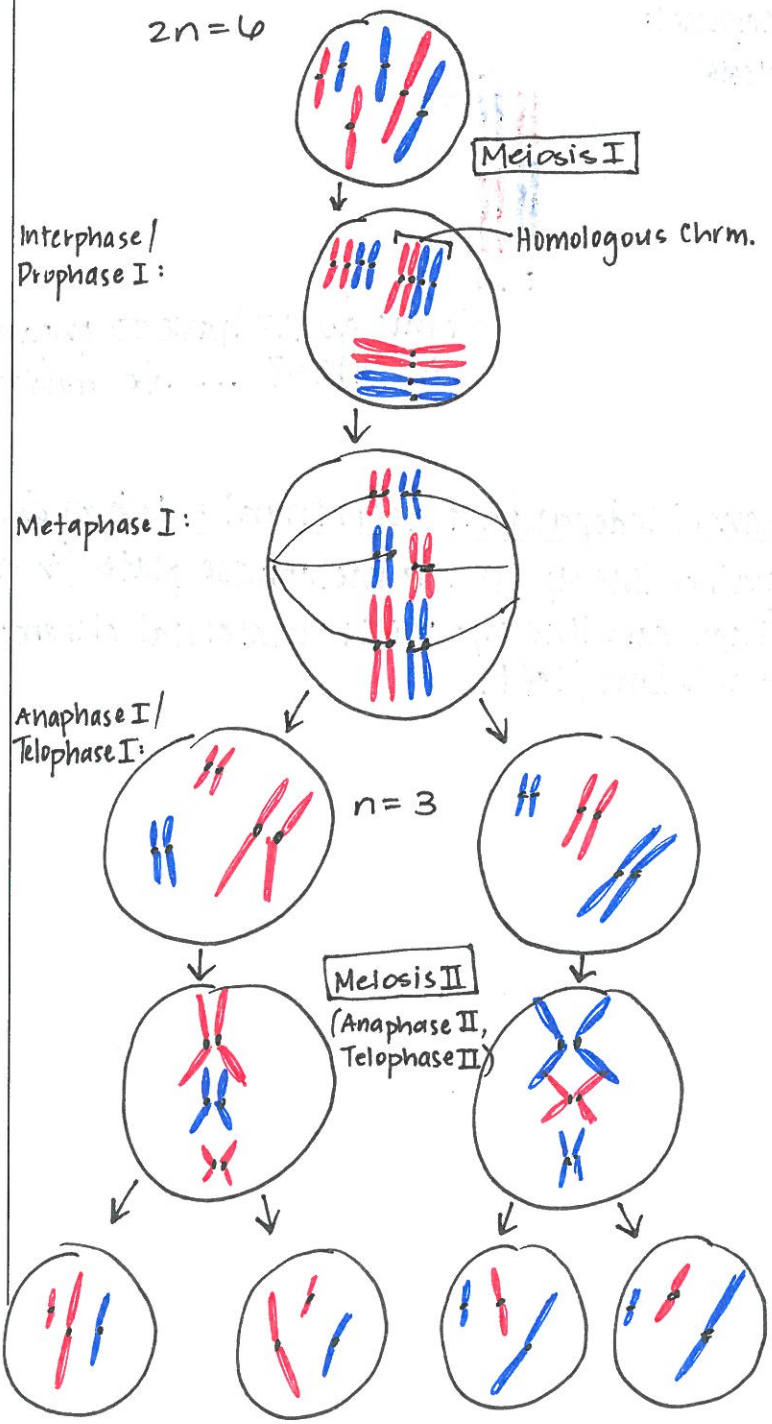
→ In meiosis, one copy goes into one gamete; the other in the other gamete.

→ 50/50 chance you will get either allele in your gamete (sperm/egg)

Mitosis



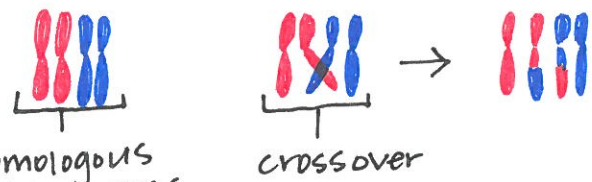
Meiosis

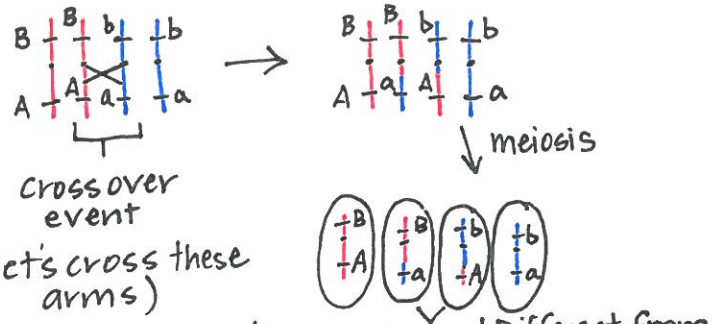


Recombinant Chromosomes:

~~Law of Independent Assortment~~

During meiosis I (prophase/metaphase), the chromosome arms of homologous chrms can switch.

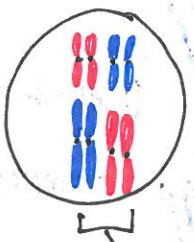




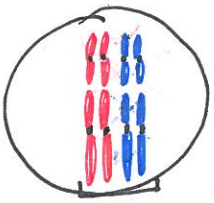
Law of Independent Assortment:

~~Meiosis: Recombinant Chromosomes/Crossing Over~~

Metaphase I:
Meiosis)



Why do we have to organize the chromosomes like this? ... We don't. We could also have this ↓



Law of Independent Assortment = When chromosome pairs line up on the metaphase plate (in meiosis), they can line up with maternal chromosomes on the right/left.

Inheritance Patterns: *(complete, incomplete, co-dominance, multiple alleles, linked genes, sex-linked genes, multiple genes)*

Complete Dominance: one allele overrides the other

Example: *Tongue Rolling*

(an allele is an alternative version of a gene - you have 2 copies of each gene - so you have 2 alleles for each gene AA or Aa or aa)

T = can roll
t = cannot roll

↑ We use uppercase and lowercase letters to represent alleles.
(*Notes: We have to use the same letter when talking about the different alleles of one gene.)

TT = can roll
Tt = can roll
tt = cannot roll

↑ Genotype (the alleles or letters)

↑ Phenotype (the physical appearance of the gene)

Homozygous = two of the same allele (TT, tt)
Heterozygous = two different alleles (Tt)

EX. Mate a homozygous tongue roller with a non-tongue roller

(TT) can only give T → 100% will be Tt ← (tt) can only give t

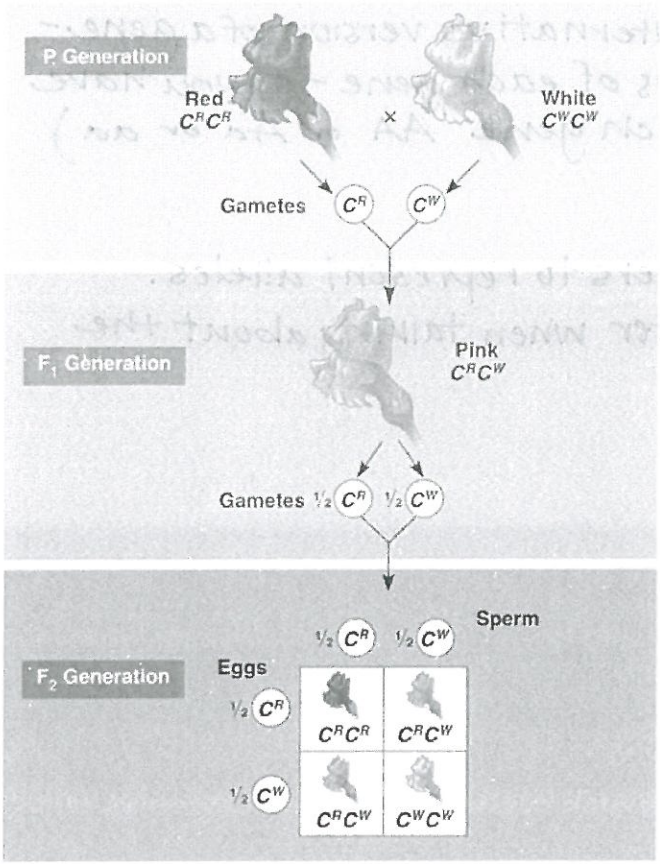
Punnett Square:

	T	T
t	Tt	Tt
t	Tt	Tt

= 100% Tt (genotype)
Tongue rollers (phenotype)

Incomplete Dominance: neither allele overrides the other, instead an intermediate phenotype occurs

Example: *Snap Dragons*



RR = red

Rr = pink

rr = white

ex. BB = black skin
Bb = grey skin
bb = white skin

Whats the probability of producing a white skined alien from a grey and white alien cross?

Bb ↓b × bb ↓b

Q. prob. of bb

½ × ½ = ¼ or ½ 50% bb white alien

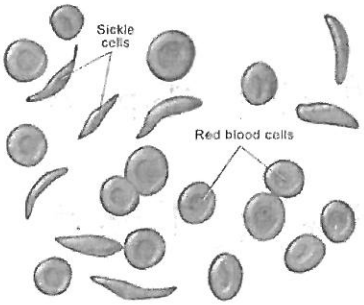
or P.□. way:

	B	b
b	Bb	bb
b	Bb	bb

¼ : 50% bb white alien

Co-Dominance: both alleles are expressed

Example: *Sickle Cell Anemia*



D = normal blood cells
d = sickled blood cells

Genotype	Phenotype
DD	All normal blood cells
Dd	Both normal and sickled cells
dd	All sickled cells

Heterozygote Advantage → Malaria Defense

ex. FF = stripes
Ff = stripes + dots
ff = dots

A gardener has flowers with stripes and dots and flowers that only have dots. The customer only wants striped flowers. How does the gardener do that?

Has → Ff and ff

Ff x ff

	F	f
f	Ff	ff
f	Ff	ff

* mate two flowers that have both stripes and dots

Ff x Ff

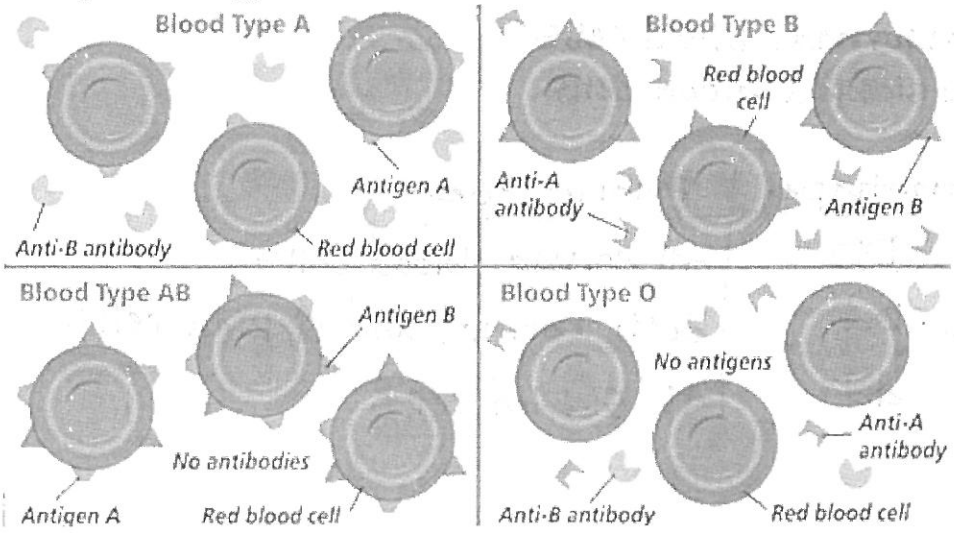
	F	f
F	FF	Ff
f	Ff	ff

25% of the flowers will be pure striped.

* You can then mate these together to get only striped flowers.

Multiple Alleles: one gene has multiple alleles

Example: *Blood Types*



Genotype	Phenotype
$I^A I^A$ or $I^A i$	Type A
$I^B I^B$ or $I^B i$	Type B
$I^A I^B$	Type AB
ii	Type O

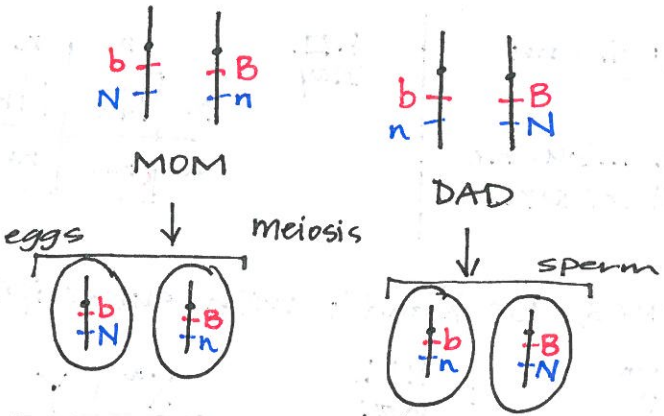
* (+)(-) is not in this gene.

Linked Genes: genes located very close to one another on the same chromosome.

Let's look at an example with a possible linked genes: red hair + freckles
(assume complete dominance)

IF 2 GENES ARE LINKED:

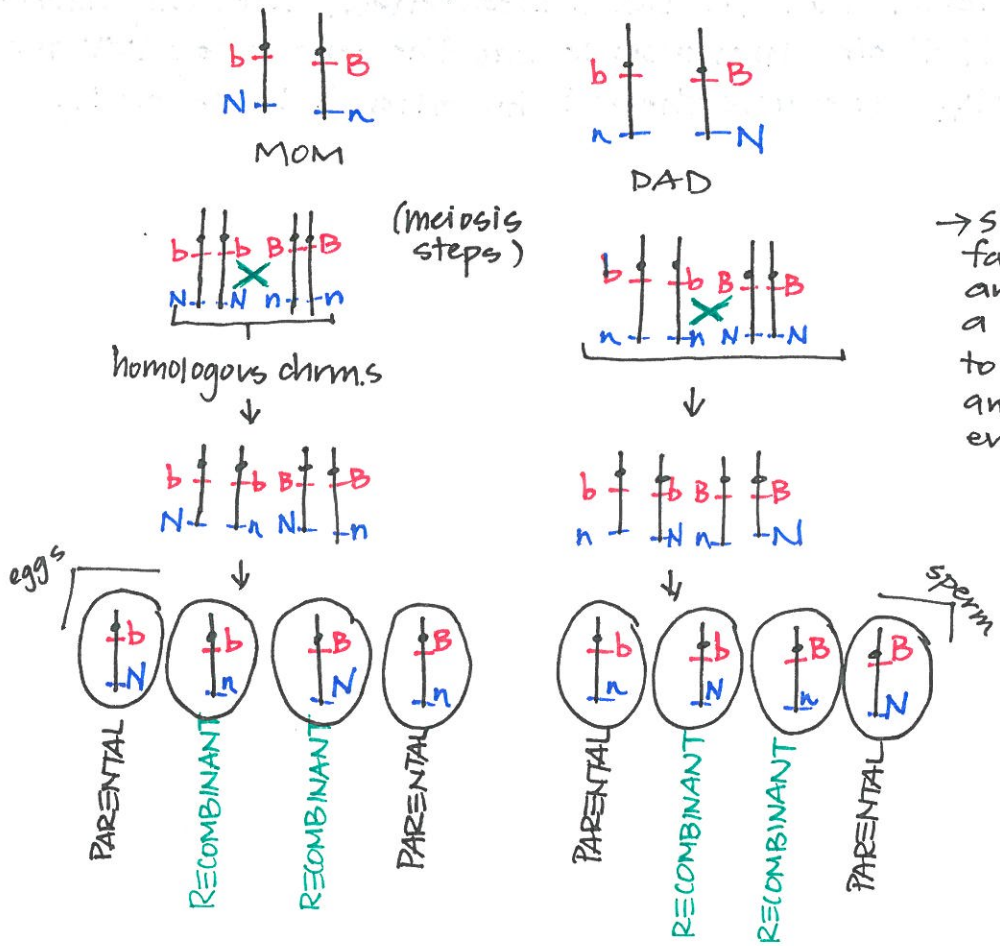
B = brown hair
b = red hair
N = normal skin
n = freckled skin



→ all gametes produced will be parental genotypes (no new combinations are produced)

→ the possibility of a cross over occurring between two linked genes (located close together) is VERY low.

IF 2 GENES ARE NOT LINKED:



→ since the genes are far apart from one another, they have a greater probability to be split from one another in a crossover event.

Sex-Linked Genes: genes located on the sex chromosomes (X, Y)

Example: *Colorblindness*

N = normal vision

n = colorblind

Mate a colorblind man with a heterozygous woman. What's the prob. they get a colorblind girl?

nn x Nn Q: girl nn

1 ↘ ↙ ½

nn ⇒ 50%. ...but we can't tell sex...

P.□. Way

	n	n
N	Nn	Nn
n	nn	nn

→ 50%. but we have no way to tell sex.

→ For sex-linked genes, use X and Y with subscripts.

♂ X^nY x ♀ X^NX^n

½ ↘ ↙ ½

♀ X^nX^n = 1/4 or 25%.

P.□. Way

	X^n	Y
X^N	X^NX^n	X^NY
X^n	X^nX^n	X^nY

Q. ♀ X^nX^n = 1/4 or 25%.

→ Women who are heterozygotes are known as carriers... can Males be carriers? Not for sex-linked genes. They only have one X chromosome. If their chromosome has the gene they will for sure have that phenotype (because their is no other X to cover it.)

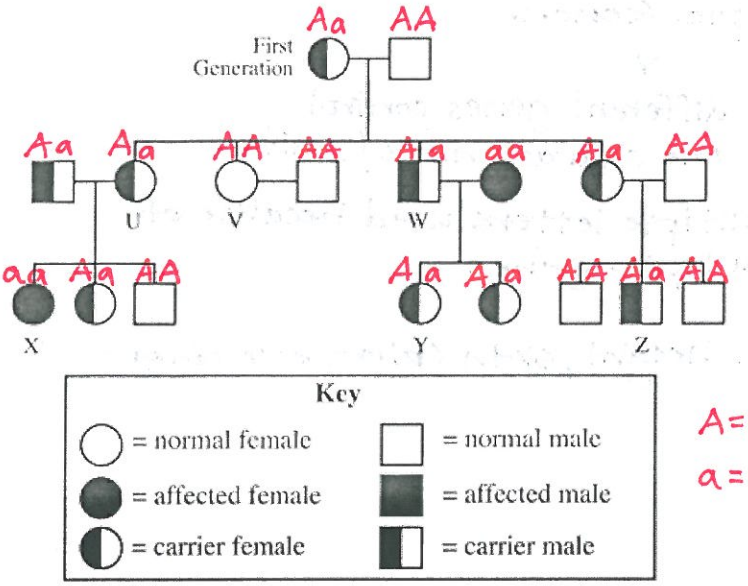
Multiple Genes: several genes control one trait

Example: *Height*

Multiple Alleles	vs.	Multiple Genes
↓		↓
different versions of one gene		different genes control one characteristic / trait
• one letter used		• multiple letters used because of multiple genes
• one gene		
• ex. Blood Type		• ex. height, skin color, eye color

Visual View of Inheritance:

Pedigrees: Family trees that show genetic inheritance



Fill in the genotypes for the pedigree provided. Assume it shows a family with Chloupek's disease, an autosomal recessive disease of the brain.

↳ We know it's not sex-linked.

*males can be carriers of autosomal diseases.

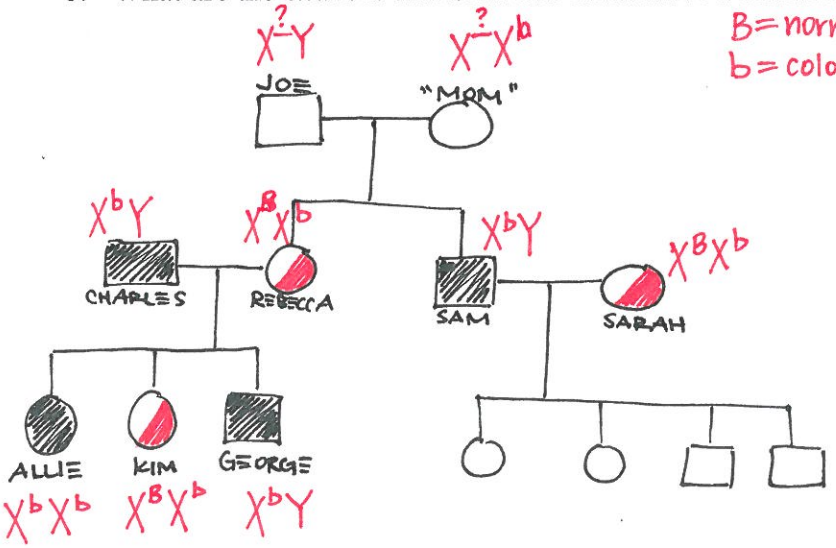
A = normal
a = diseased

Create a pedigree of the Smith family. Joe is the dad of Rebecca and Sam. Sam is colorblind (a sex-linked recessive disease of the eye). Rebecca married Charles, who is colorblind. Rebecca and Charles have three kids (Allie, Kim, and George). George and Allie are both colorblind. Sam marries Sarah, who is a carrier of colorblindness, and they have four kids, two boys and two girls.

* use X & Y,
* men cannot be carriers.

a. Who are the possible carriers of colorblindness? Rebecca, Sarah, Kim
b. What are the chances that Sam and Sarah have a colorblind girl? A colorblind boy?

B = normal color vision
b = colorblind



(b) $X^bY \times X^BX^b$

P.□ way:

	X^b	Y
X^B	X^BX^b	X^BY
X^b	X^bX^b	X^bY

colorblind girl = 50%. (or 25% total)
colorblind boy = 50%. (or 25% total)

Genetic Problems:

Monohybrid: heterozygous for only one trait

Dihybrid: heterozygous for two traits

Monohybrid Problems:

HH

In rhinos, a long horn (H) is dominant over a small horn (h). If a homozygous long horned rhino is crossed with a small horn rhino, what would the offspring look like (phenotype)? What if you mixed two of their offspring? What is the percentage of offspring that are small horned?

hh

H = long horn
h = small horn

HH x hh → All (100%.) long horned.

(b.) mix Hh x Hh → ?: % offspring hh

½ ½ → ¼ hh = 25%. hh

or

P.□. way

	H	h
H	HH	Hh
h	Hh	hh

25%. or ¼ hh

$I^A I^A$ or $I^A i$

$I^A I^B$

A baby was stolen from the hospital. The parents were type A and type AB. A baby was found 16 hours later and when blood tested, the results showed type O. Is this the stolen baby? Justify your answer.

ii

* For the baby to be ii, it would have gotten a little i from each parent.

* Type AB is incapable of donating a "i" so this is not the parent

or

P.□ way:

	I^A	I^A
I^A	$I^A I^A$	$I^A I^A$
I^B	$I^A I^B$	$I^A I^B$

or

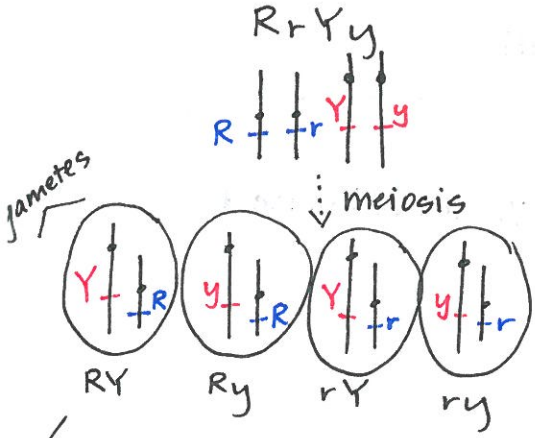
	I^A	i
I^A	$I^A I^A$	$I^A i$
I^B	$I^A I^B$	$I^B i$

No "ii" possible. Not their baby.

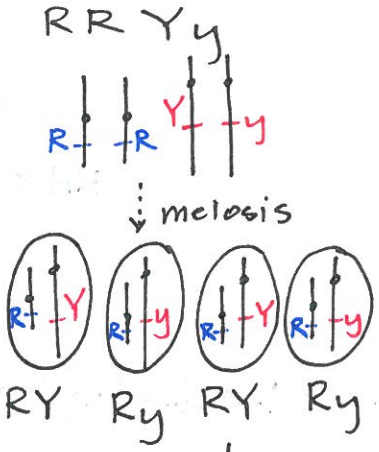
Dihybrid Problems: assume each gene is not linked and located on a different chromosome.

What is the probability that a heterozygous round yellow pea (RrYy) crossed with a homozygous round heterozygous yellow pea (RRYy) would produce a round yellow pea with the genotype of RrYy?

RrYy x RRYy → Q: prob. of RrYy?



x



P.S. way:

	RY	Ry	RY	Ry
RY	RRYY	RRYy	RRYY	RRYy
Ry	RRYy	RRyy	RRYy	RRyy
rY	RrYY	RrYy	RrYY	RrYy
ry	RrYy	Rryy	RrYy	Rryy

Q: prob. of RrYy

4/16 → 1/4 → 25% prob. of genotype RrYy

(!) *** **Law of Multiplication:**

* No need for large complex punnett squares.

Q. prob. of RrYy offspring

split in 3 steps:

1.) Prob to get Rr (Rr x RR) = 1/2
2.) Prob to get Yy (Yy x Yy) = 1/2
3.) Multiply steps 1+2

Rr x Yy
1/2 1/2 = 1/4 (25%)

same answer, but a lot less work

Part III: "What You Do with the Stuff Once You've Got It"

Gene Expression