

Notes for

GENETICS

Biology
Semester “B”
2018
Mr. Johnson

I. Genetics – the study of heredity.



How characteristics are
passed from one
generation to the next.

A. Genetic information structures:

1. DNA – the basic information-carrying molecule.
– very long (6 feet) double-strand.
2. Chromatin – DNA wrapped around histone proteins.
– how DNA is kept neatly.
3. Chromosomes – chromatin folded into bundles.
– humans have 46 chromosomes
(*that's **23 identical pair** per cell*)

Illustrated summary:



DNA $\xrightarrow[\text{histone proteins}]{\text{wrapped around}}$ chromatin $\xrightarrow{\text{folded into}}$ chromosome

4. Detail of a chromosome:

a. structure



bands are called “**genes**”

*DNA sequences that code for a single, specific trait.

Ex) eye color, nose shape, etc.

*about 3,000 genes per chromosome.

b. Remember, chromosomes come in pairs.

*we have **23 pairs**.

*one from mom and one from dad.

B. Some genetics vocabulary: [Speaking the Language](#)

1. **Alleles** – different forms of a gene.

a. Dominant allele – the one that shows.

b. Recessive allele – the one that is covered up.

2. **Trait** – a physical characteristic, determined by a combination of two alleles.

3. **Generations** – age groups

a. P generation = parents

b. F_1 = offspring of P generation

c. F_2 = offspring of F_1 generation

4. Genotype & Phenotype

(genetic makeup)

(physical appearance)

5. Homozygous & Heterozygous

"Purebred"

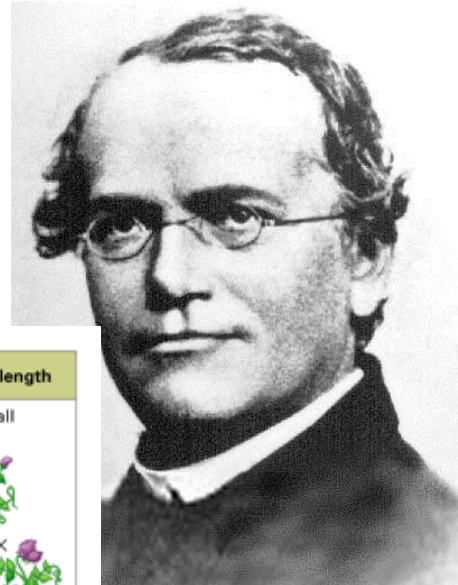
"Hybrid"

(Both alleles are the same) (Both alleles are different)

C. [Gregor Mendel](#) (start-2:00)

1. European monk & teacher from 1800's.
2. Crossed pea plants and studied their offspring.
3. Compared several physical traits in pea plants:

- *pea color
- *plant height
- *wrinkly / smooth peas
- *flower color
- *etc.

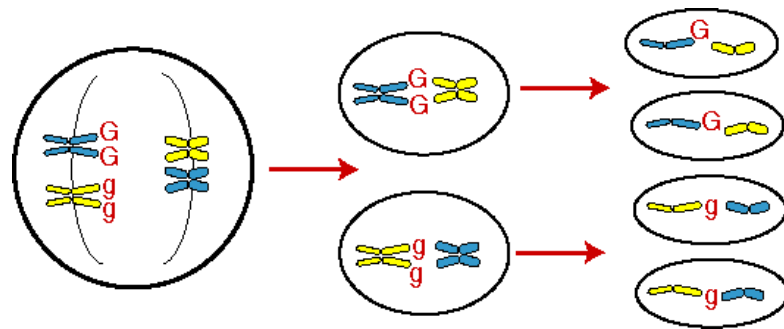


	Flower color	Flower position	Seed color	Seed shape	Pod shape	Pod color	Stem length
P	Purple X White	Axial X Terminal	Yellow X Green	Round X Wrinkled	Inflated X Constricted	Green X Yellow	Tall X Dwarf
F ₁	Purple	Axial	Yellow	Round	Inflated	Green	Tall

2. Concluded 4 things about heredity:

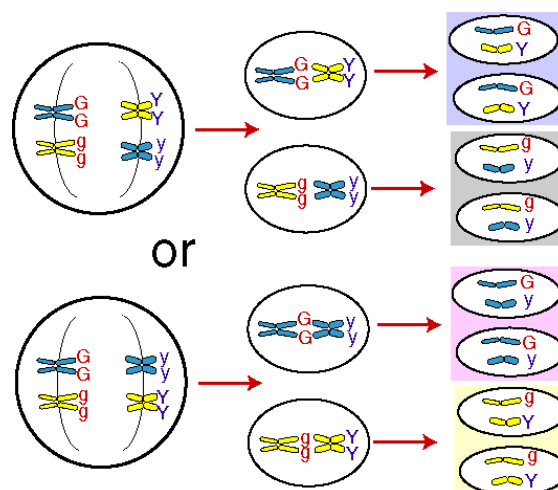
- a. characteristics (traits) are determined and passed down by genes.
- b. Some genes may be dominant over others.
- c. We have 2 copies of each gene (one from each parent) which are separated during **meiosis** in a process called

SEGREGATION. (5:20-6:30)



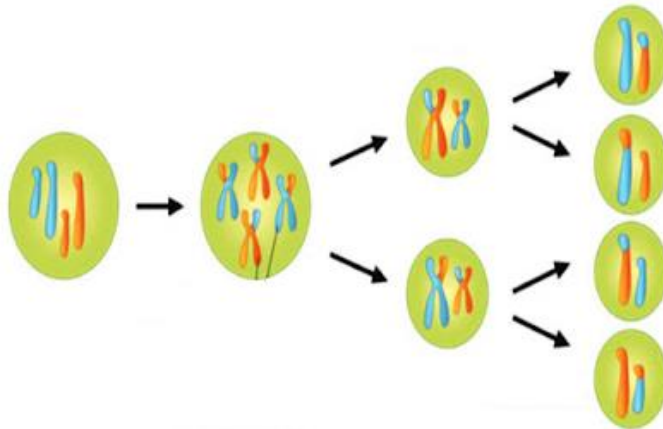
- d. Genes are passed down **separately** from each other in a process called

INDEPENDENT ASSORTMENT (6:30-7:15)



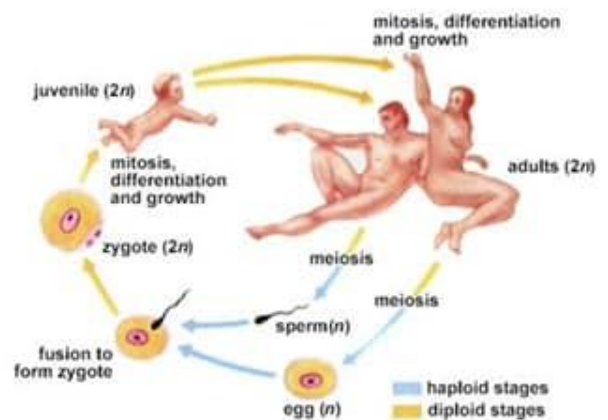
D. Meiosis (start-1:50)

1. Cell division that cuts the chromosomes in half.



2. Makes cells called “gametes” (egg and sperm cells).

3. Gametes have only half the chromosomes so when they combine there is just one complete set of chromosomes.



4. **Somatic** cells - all cells in body except gametes
 - full set of 46 chromosomes (that's 23 pairs from each parent).
 - called a “**diploid**” number of chromosomes, or ‘**2n**’.
5. **Gamete** cells
 - half-set of chromosomes (that's 23 **total**).
 - called a “**haploid**” number of chromosomes, or just ‘**n**’.

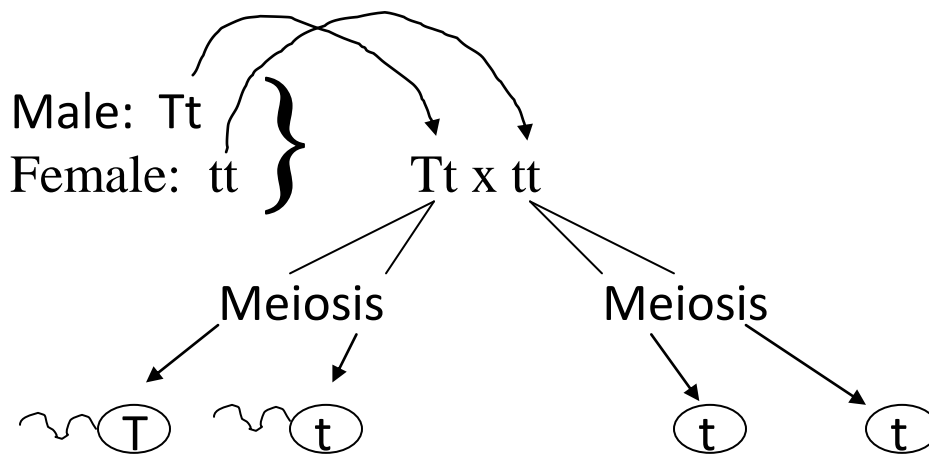
E. Predicting offspring using the Punnett Square

Ex) Tallness is a single trait controlled by 2 alleles:

T is the dominant allele for tall and

t is the recessive allele for short.

Let's cross a **heterozygous** tall male with a **homozygous recessive** female:



Genotypic Ratio -

Phenotypic Ratio -

E. Predicting offspring using the Punnett Square

Ex) Tallness is a single trait controlled by 2 alleles:

T is the dominant allele for tall and

t is the recessive allele for short.

Let's cross a **heterozygous** tall male with a **homozygous recessive** female:

(same alleles)

(lowercase)

tt

Male: Tt

Female: tt

Tt x tt

Meiosis

Meiosis

T t

t t

Put these alleles
on top of the
Punnett Square

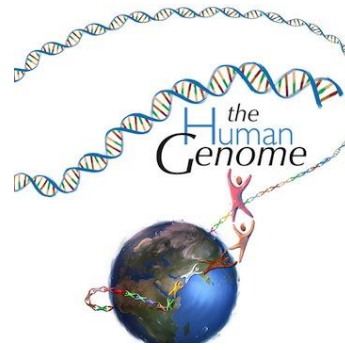
put these alleles
on the side of
the Punnett Square

	T	t
t	Tt	tt
t	Tt	tt

Genotypic Ratio - (possible gene combinations) $2Tt : 2tt$

Phenotypic Ratio - (How they actually look) $2 \text{ tall} : 2 \text{ short}$

II. Human Genome Project



A. An attempt to analyze and map all human genes.
(which genes on which chromosomes do what?)

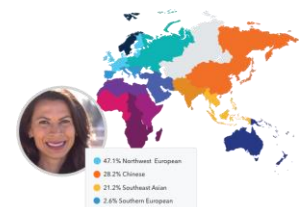
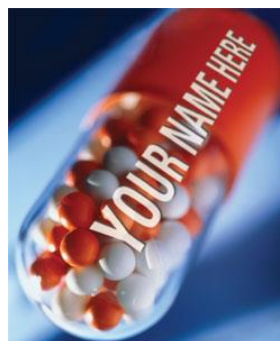
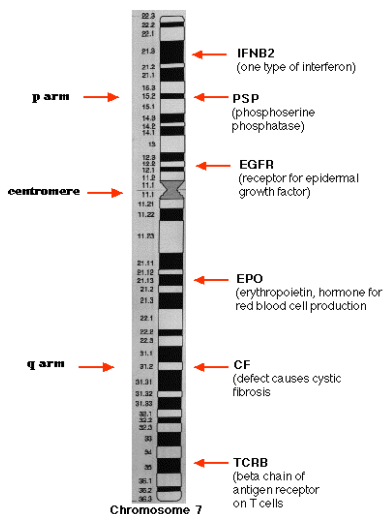


B. Started in 1990, basically finished by 2000.



C. What's it good for?

Gene Therapy disease prevention personalized medicine because it's just plain cool!



III. Karyotypes

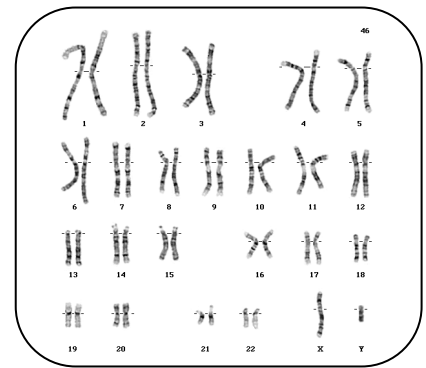
A. Full set of chromosomes arranged into a meaningful pattern.

B. Arranged by **size** and **gene banding pattern**

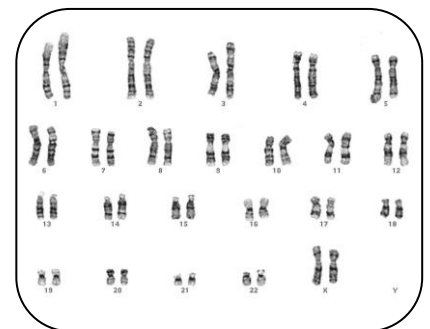
from largest to smallest with sex pair last.

C. Pairs #1-22 called “**autosomes**” or “**somatic chromosomes**”.

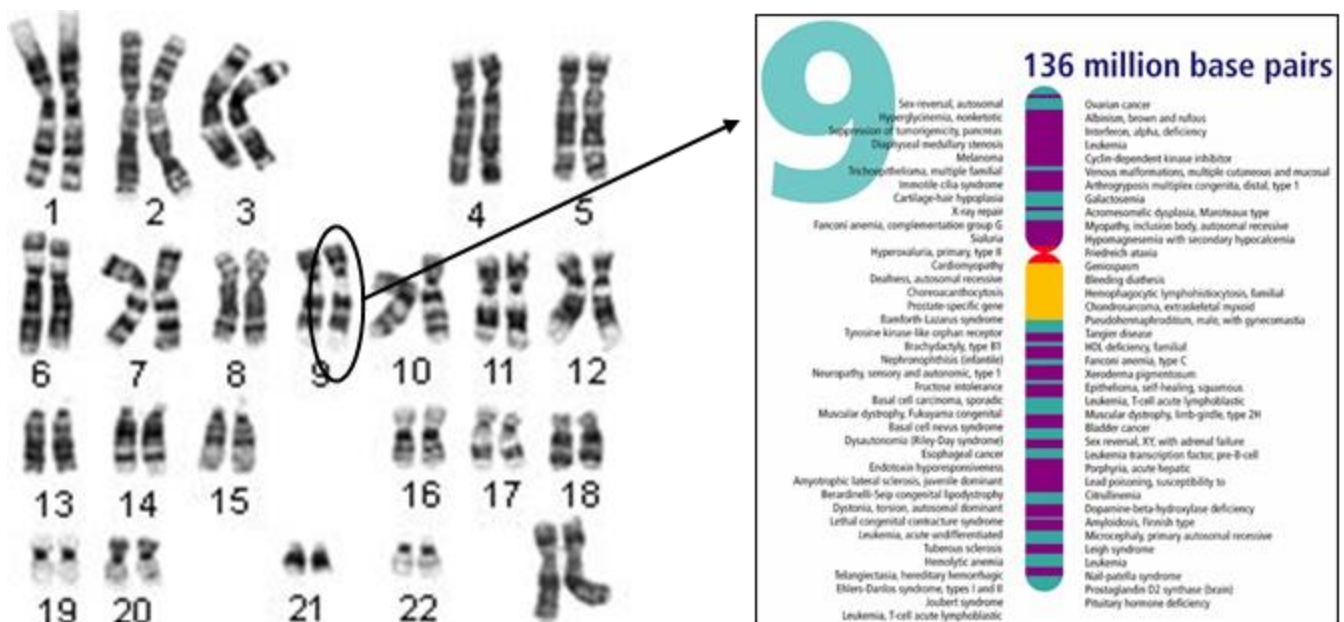
male (46XY)



female (46XX)



D. Closer look at a chromosome shows the genes (*bands*):



E. Karyotypes can show many forms of genetic disease:

1. **Extra** chromosome (47) = **trisomy** disease
2. **Missing** chromosome (45) = **monosomy** disease
3. **Part** of a chromosome missing = **deletion** disease



4. **Changed** chromosome
*some genes are switched around in one of these ways:

a. Translocations

– two chromosomes trade sections



b. Inversions

– section of a chromosome flips around



c. Duplications

- section of chromosome is doubled



F. Some Genetic Disorders you may have heard of...

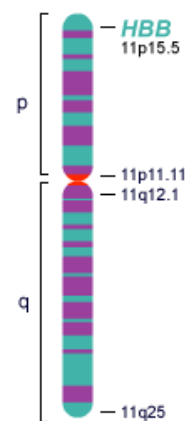
Autosomal Disorders (mutations of genes on chromosomes #1-22)

1) Sickle-Cell Anemia <https://the1a.org/shows/2018-02-05/why-you-dont-hear-much-about-sickle-cell-anymore>

- recessive gene (called HBB) on chromosome #11
- moon-shaped RBC cannot carry oxygen
- tired, cold, sick, painful

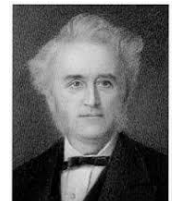


Chromosome 11

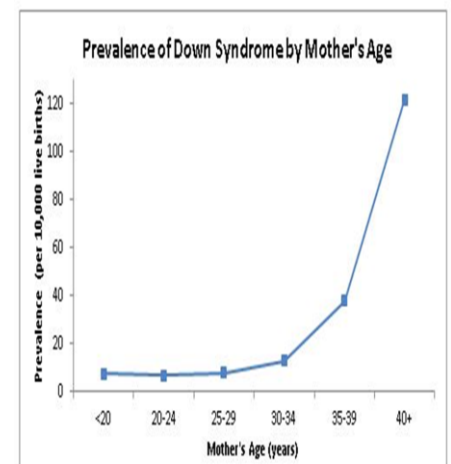
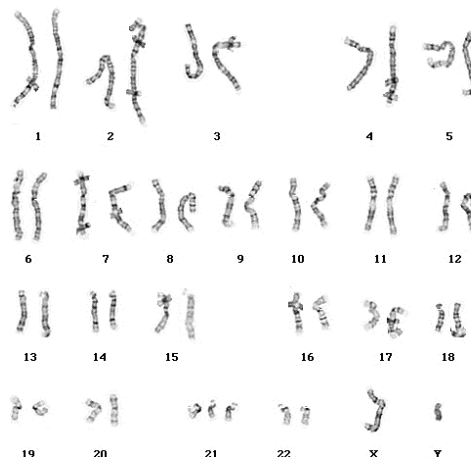


2) Down's syndrome (*trisomy 21*)

- extra chromosome #21
- flattened faces, large tongue, low-set ears
- mental impairment (IQ 25-50)
- heart defects, 50% mortality by 30 years old
- chances increase with the age of the mother



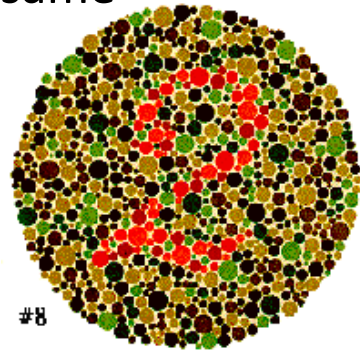
Dr. John Langdon Down
Great Britain, 1899



Sex-Linked Disorders (mutations of genes on chromosomes #23)

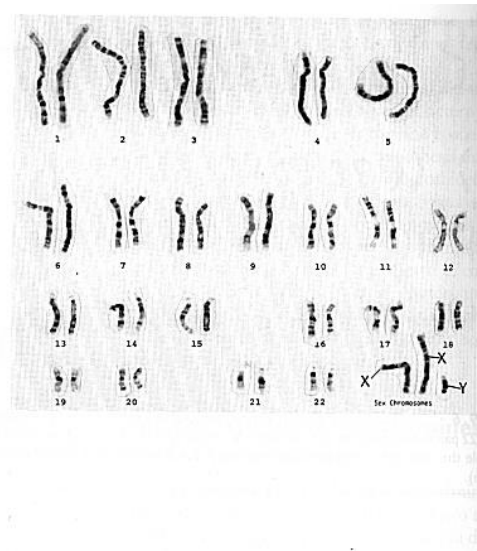
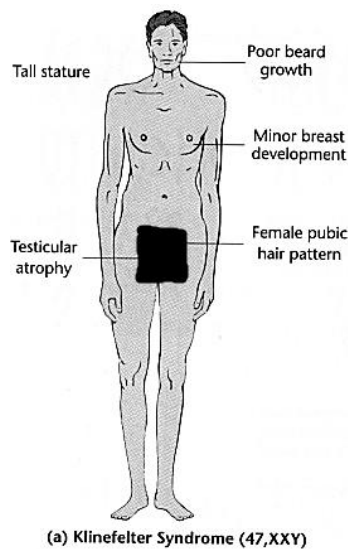
1) [colorblindness](#) - two different colors appear the same (red/green is most common)

- caused by a recessive gene on the X chr.
- 5-10% of males, less than 2% of females



2) Klinefelter's syndrome

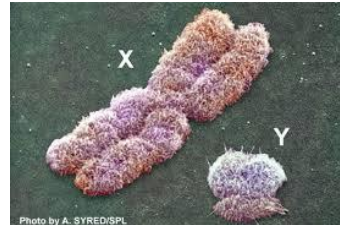
- males with extra X's (XXY or $XXXY$)
- Male (*remember: if it has a Y, it's a guy*) with exaggerated female characteristics
 - wide hips
 - some breast development
 - little facial hair



IV. Predicting Sex-Linked Traits

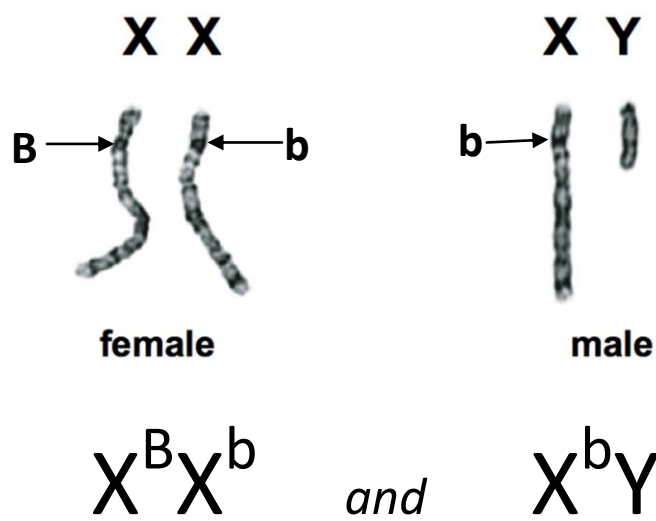
- a. Recall that chromosome **pair #23** determines our sex.
- b. Most sex-linked genes are on the X-chromosome.

**it's just so much bigger
with more genes!*



- c. Genes are written as “exponents” on the chromosome.
**remember: the genes are found on the chromosomes*

So, if we are talking about the gene for colorblindness
(let's call it **B** for **normal vision** and **b** for **colorblindness**)
and these genes are located on the X chromosome like
the picture below shows, we would write the genotypes as...



EX) Colorblindness is a sex-linked recessive allele found on the X chromosome.

B=normal vision / b=colorblind.

$X^B X^B$ = female with normal vision

$X^B X^b$ = female with normal vision

$X^b X^b$ = female with colorblind vision

$X^B Y$ = male with normal vision

$X^b Y$ = male with colorblind vision

Problem: Cross a heterozygous normal-vision female with a colorblind male, giving the GR and PR.

Genotypic Ratio -

Phenotypic Ratio -

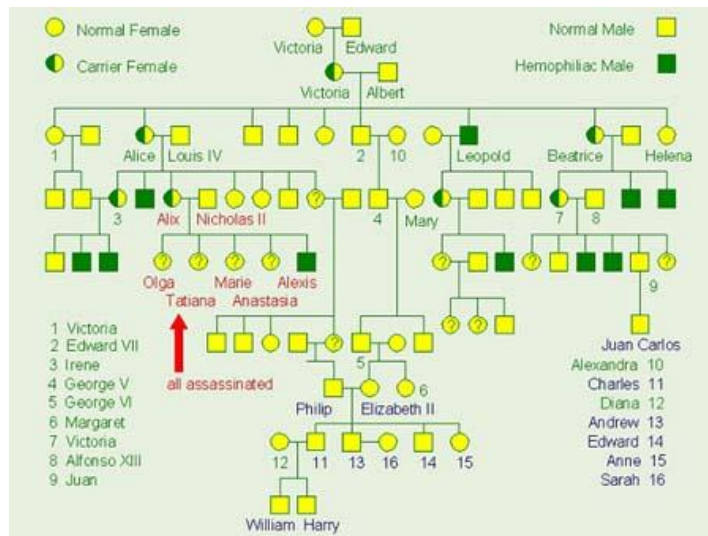
V. Pedigrees

A. Chart which shows genetic relationships within a family.



B. Symbols used in a pedigree:

1. \square = male and \bigcirc = female
2. Shaded = homozygous dominant
3. Unshaded = homozygous recessive
4. Half-shaded = heterozygous ("carrier")
5. Roman numerals = generations
6. Numbers = birth order within a generation



C. Let's build an example of a pedigree!!!

Albinism – a recessive trait

(A=normal / a=albino)

The Adam's family has a history of albinism. Tom Adams, who appears normal but is a carrier for albinism, marries a woman named Mary who has albinism. They have five children named Steven, Katie, Laura, Kim, and Nate. Katie, Laura, and Nate are carriers of albinism, while Steven and Kim are true albinos. Steven, the oldest, gets married to Meagan who has no genes for albinism. They have three children: Kelli, Cassi, and Mike (you figure out their genotypes). Laura gets married to an albino named Carl. They have two albino children named Britney and Tom. Nate, the youngest, gets married to Colleen who has no albinism genes. They travel the world doing good things and have decided to have no children.

*How many generations are in the pedigree?

*How many offspring did Generation I have?

*How many marriages in Generation II?

*What's the genotype and phenotype of the second oldest child in Generation II?

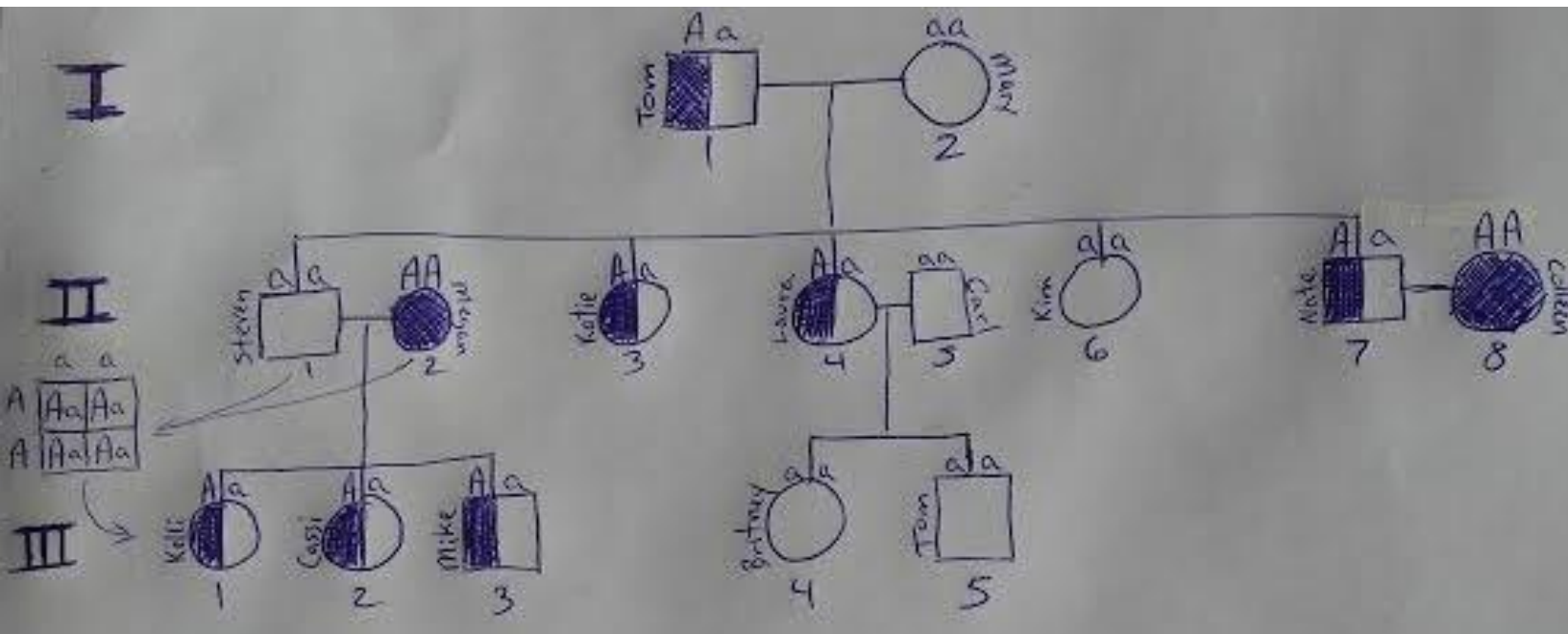
*If **II-3** had an affair with **II-4's husband** (ooh-la-la), what would their children look like?(yup, Punnet Square time!)

C. Let's build an example of a pedigree!!!

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*How many generations are in the pedigree? 3

*How many offspring did Generation I have? 5

*How many marriages in Generation II? 3

*What's the genotype and phenotype of the second oldest child in Generation II?

↓ Aa ↓ normal

*If II-3 had an affair with II-4's husband (ooh-la-la), what would their children look like? (yup, Punnet Square time!)

Katie
(Aa)

Carl
(aa)

	A	a
a	Aa	aa
a	Aa	aa

GR - 2Aa : 2aa

PR - 2Normal : 2albino

VI. Human Blood Types

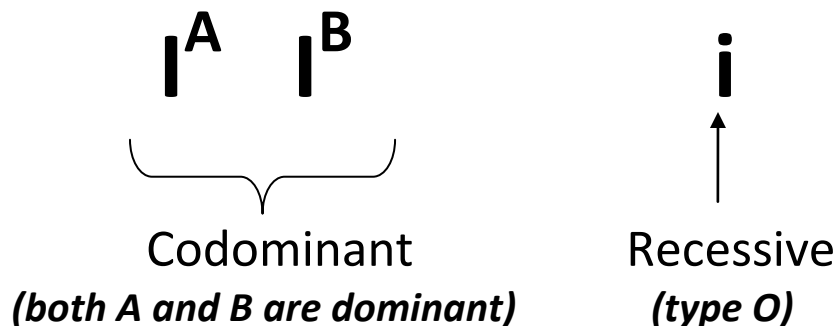
- A. Variety of genetically determined types.
- B. Using the wrong blood type in a transfusion can be fatal!
- C. Blood type is a **polygenetic** trait.
(it is determined by two or more genes working together)

In blood, the **two genes** are:

the **ABO gene** and the **Rh gene**.

1. ABO gene:

- a. There are **3 alleles** for the ABO gene:



- b. $I^A I^A$ and $I^A i$ = type **A** blood
- c. $I^B I^B$ and $I^B i$ = type **B** blood
- d. $I^A I^B$ = type **AB** blood
- e. ii = type **O** blood

2. Rh gene

a. **Two** alleles for this gene: Rh^+ and Rh^-

b. Rh^+ is dominant, Rh^- is recessive.

- Rh^+/Rh^+ is __POSITIVE__
- Rh^+/Rh^- is __POSITIVE__
- Rh^-/Rh^- is __NEGATIVE__

D. Antigens

1. “Name tags” on red blood cells which are recognized by antibodies in our immune system.

2. The **A**’s and **B**’s indicate the antigen types:

- Type **A** has **A** antigens
- Type **B** has **B** antigens
- Type **AB** has both **A** antigens and **B** antigens
- Type **O** has **no** antigens

E. Antibodies

1. Part of the immune system in the blood.

2. Kills blood cells with matching antigen.

a. Type **A** blood has **B**-antibodies

b. Type **B** blood has **A**-antibodies

c. Type **AB** blood has **no** antibodies

d. Type **O** blood has **A**-antibodies and **B**-antibodies

3. **Universal Donor:** Type **O**

*can be given to anyone

(no antigens for antibodies to attack)

4. **Universal Recipient:** Type **AB**

*can accept blood from anyone

(no antibodies to attack blood cells)

*Questions:

1. If someone has the genotype $I^A I^A Rh^+ Rh^-$ then what is their blood phenotype?
2. Someone with type AB blood ($I^A I^B$) has a child with a person with type A blood ($I^A i$), what blood types can their children have? (Show with a Punnett Square & Ratios)
3. Can a child from parents with type A blood ($I^A i$) and type B blood ($I^B i$) have type O blood? (Show with a Punnett square)

*Questions:

1. If someone has the genotype $I^A I^A Rh^+ Rh^-$ then what is their blood phenotype?

type A

positive is dominant, so...

A-positive

2. Someone with type AB blood ($I^A I^B$) has a child with a person with type A blood ($I^A i$), what blood types can their children have? (Show with a Punnett Square & Ratios)

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

GR - $1 I^A I^A : 1 I^A I^B : 1 I^A i : 1 I^B i$

PR - 2 type A : 1 type AB : 1 type B

3. Can a child from parents with type A blood ($I^A i$) and type B blood ($I^B i$) have type O blood? (Show with a Punnett square)

YES

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

GR - $1 I^A I^B : 1 I^B i : 1 I^A i : 1 ii$

PR - 1 type AB : 1 type B : 1 type A : 1 type O

