

## Ivan's Chapter 6 Study Guide

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### Body Cells and Gametes

- **Somatic Cells** (also called body cells) make up your tissues and organs
  - The DNA in these cells is not passed down to your children
- **Germ Cells** are cells which reside in your reproductive organs
  - Testes in men and ovaries in women
  - Develop into **Gametes**
- Gametes are sex cells
  - Ova/eggs in women
  - Spermatozoa in men
  - The DNA from these cells can be passed down to your children
- Besides mutations, your somatic cells are genetically identical

### Chromosomes

- Each species has a different number of chromosomes per cell
  - Humans have 46 chromosomes which come in 23 pairs
    - Can be divided into two sets, 23 of which came from your mother and 23 of which came from your father
- **Homologous chromosomes** are two chromosomes which have the same appearance, length, and genes (although each copy may differ)
- Chromosomes 1 - 22 are called autosomes
  - These contain genes which are not related to an organism's sex
- The 23rd pair of chromosomes are sex chromosomes
  - **Sex chromosomes** directly control sex characteristics
  - Are represented as an "X" and "Y"
    - Males have XY chromosomes and females have XX chromosomes
- The X chromosome is larger than the Y chromosome
  - Contains many genes unrelated to sex, as well as genes which are related
- The Y chromosome is the smallest chromosome and has the fewest genes
  - Contains genes which relate to the development of male traits

### Reproduction

- *Sexual Reproduction* requires the fusion of two gametes
  - This fusion of egg and sperm is known as *fertilization*
- The nuclei of the egg and sperm fuse
  - The unified nucleus contains a full set of chromosomes
    - This means that the egg and sperm only have 23 chromosomes each

### Diploid and Haploid Cells

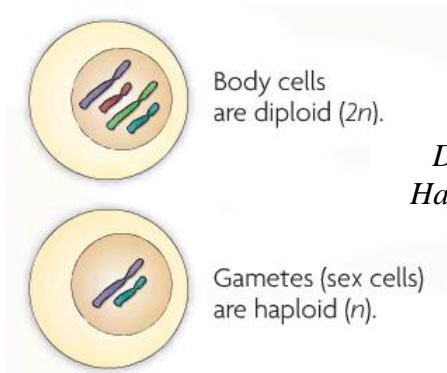
- **Diploid cells** have two copies of each chromosome (One copy from mom, one copy from dad)
  - Are represented as " $2n$ "
  - Body cells are diploid since they contain a full set of 46 chromosomes
- Haploid cells only have one copy of each chromosome
  - Are represented as " $n$ "
- Each sperm or egg cell has 22 autosomes and 1 sex chromosome
  - Eggs always contain the X chromosome
  - Sperm can either contain an X or a Y
- Although a change in chromosome number is usually harmful, sometimes it can result in a new species
  - The fertilization of nonhaploid gametes has created many plant species with more than two sets of chromosomes
  - Some plants have 4 copy of each chromosome
    - Tetraploidy ( $4n$ )

### What is Meiosis?

- **Meiosis** is a form of nuclear division that divides diploid cells into haploid cells
  - Undergone by germ cells to produce gametes
  - Called "Reduction division" because it halves the chromosome number
  - Results in genetically unique haploid cells from diploid cells

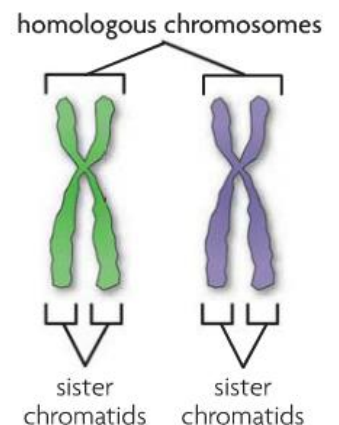
### Homologous chromosomes vs Sister chromatids

- **Homologous chromosomes** are two separate chromosomes
  - One from your mother, one from your father
  - Divided during meiosis I
- Very similar to each other
  - Have the same length and same genes
  - However, they aren't genetically identical
- **Sister chromatids** are two halves of a duplicated chromosome joined at a centromere
  - Divided during meiosis II



### *Diploid vs Haploid Cells*

### *Homologous chromosomes and sister chromatids*



### Meiosis I

- DNA has already been copied
- Divides homologous chromosomes
- Produces two haploid cells with duplicated chromosomes

#### 1. Prophase I

- The nuclear membrane breaks down
- Centrosomes and centrioles move to opposite poles of the cell
- Spindle fibers assemble
- Duplicated chromosomes condense
- Homologous chromosomes separate into pairs

#### 2. Metaphase I

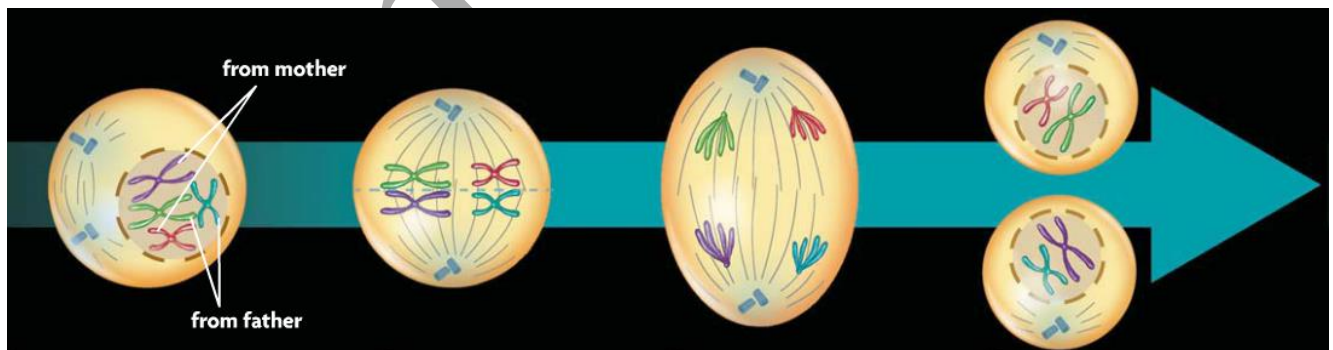
- The 23 chromosomes line up across the cell equator
- Some from the father, some from the mother
- Mixes up chromosomal combinations
- Meiosis may result in 8,388,608 possible chromosomal combinations

#### 3. Anaphase I

- Paired homologous chromosomes separate
- Move to opposite sides of the cell
- Sister chromatids remain together and throughout meiosis I

#### 4. Telophase I

- In some species, the nuclear membrane reforms
- Spindle fibers disassemble
- The cell undergoes cytokinesis
- Results in two cells with unique combinations of 23 chromosomes sourced from each part



*The process of  
Meiosis I*

### Meiosis II

- Divides sister chromatids
- Results in undoubled chromosomes
- Applies to cells produced in meiosis I
- DNA is not copied again

#### 5. Prophase II

- The nuclear membrane breaks down
- Centrosomes and centrioles move to opposite sides of the cell
- Spindle fibers assemble

#### 6. Metaphase II

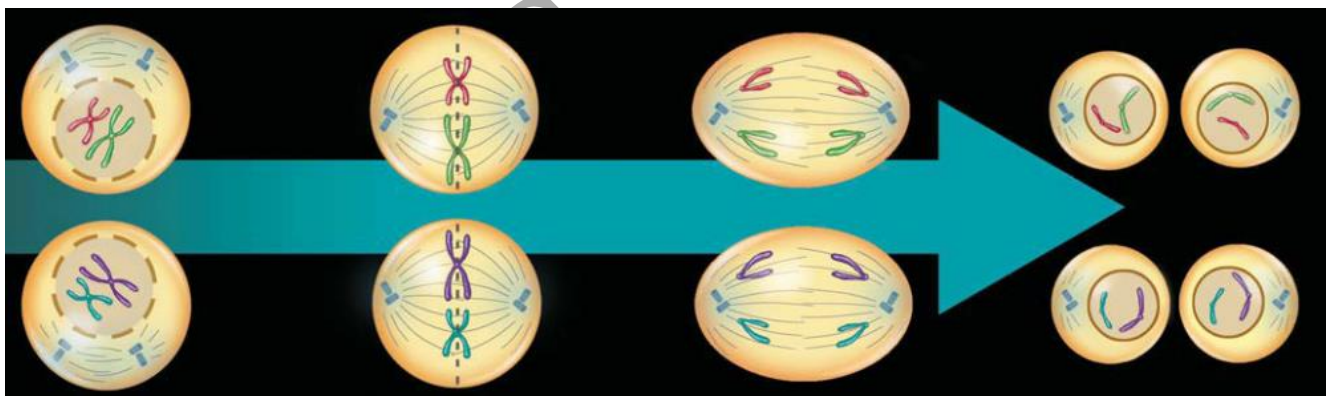
- Spindle fibers align the 23 chromosomes at the equator
  - Each chromosome still has two sister chromatids

#### 7. Anaphase II

- Sister chromatids are pulled apart
  - Move to opposite sides of the cell

#### 8. Telophase II

- The nuclear membrane reforms
- Spindle fibers disassemble
- The cell undergoes cytokinesis
- Results in four haploid cells with a combination of chromosomes from the mother and father.



*The process of  
Meiosis II*

### Gametogenesis

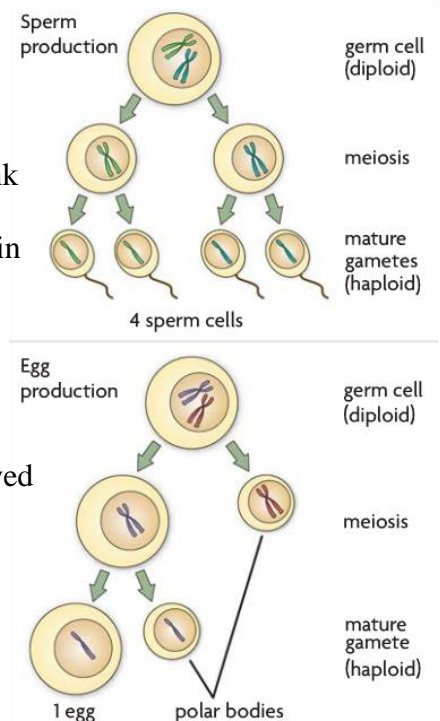
- **Gametogenesis** is the production of gametes
  - Includes meiosis and other changes which produce a pure gamete
  - The final stages differ between males and females
- **Sperm cells** = The male gamete
- **Egg cells** = The female gamete, larger than sperm cells
- The sperm cell's main contribution is DNA
  - Must swim to the egg to fertilize it
- Sperm start off as round cells and end up as streamlined cells with flagella for locomotion
- Egg cells are formed before birth
  - Includes periods of active development and inactivity
- Egg cells contribute DNA, organelles, molecular building blocks, and other materials needed to support life
- Only one cell out of the four produced by meiosis makes an egg
  - This cell receives most of the materials
- The rest of the molecules and materials are distributed into **polar bodies**
- **Polar bodies** are small cells with little more than DNA which are broken down and not used
- In many species such as humans, polar bodies produced by meiosis I do not undergo meiosis II

### Mendelian Genetics

- **Traits** = Distinguishing characteristics which are inherited
  - Eye color, skin tone, flower color, etc.
- **Genetics** = The study of biological inheritance and genes
- One of the first people to study genetics was an Austrian monk named Gregor Mendel
  - Discovered the ruling principles and laws of genetics in the mid 1800s
- Mendel recognized that traits are inherited as discrete units from the parent
- Organisms inherit two copies of each discrete unit (genes)
  - Describes how traits are passed through generations
- Mendel controlled breeding, used purebred plants, and observed traits that only appeared in 2 alternate forms

### Experimentation and Fertilization

- **Purebred** plants = A line of genetically uniform plants resulting from repeated self-pollination
  - Offspring inherit all of their parents' characteristics
  - Chosen so Mendel could know that variations in the offspring a result of his experiments
- Controlled fertilization by removing the stamen (male) and fertilizing the pistil (female) with pollen from a specific plant



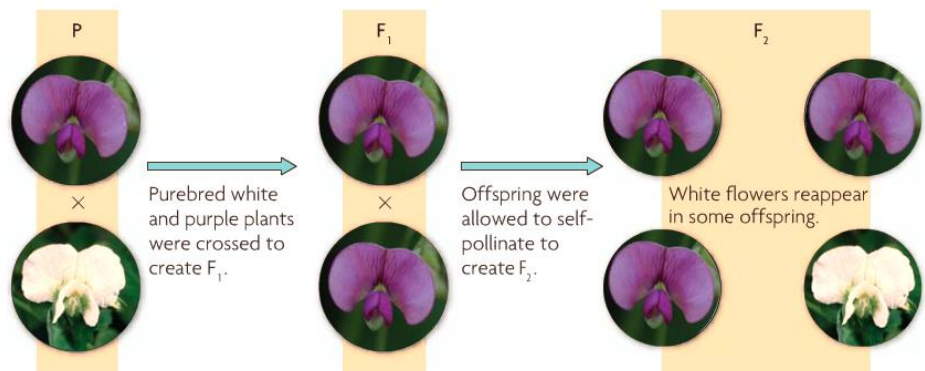
*The process of Gametogenesis*

### Results and Crosses

- **Cross** = The mating of two organisms
  - Purebred white-flowered plant with a purebred purple-flowered plant
    - The parental (P) generation
    - Traits that were hidden after crossing the P generation reappeared after the F<sub>1</sub> generation self-pollinated
- The offspring of the P generation are called the first filial (F<sub>1</sub>) generation
- The offspring of the F<sub>1</sub> generation are called the second filial (F<sub>2</sub>) and so on
- Each cross of Mendel's yielded similar F<sub>2</sub> ratios of around 3:1

RESULTS OF MENDEL'S MONOHYBRID CROSSES OF PEA PLANTS			
F <sub>2</sub> Traits	Dominant	Recessive	Ratio
Pea shape	5474 round	1850 wrinkled	2.96:1
Pea color	6022 yellow	2001 green	3.01:1
Flower color	705 purple	224 white	3.15:1
Pod shape	882 smooth	299 constricted	2.95:1
Pod color	428 green	152 yellow	2:82:1
Flower position	651 axial	207 terminal	3.14:1
Plant height	787 tall	277 short	2.84:1

- Mendel concluded that...
  - Traits are inherited as discrete units
    - Explained why traits persisted without blending together
  - Also concluded the **law of segregation**
- The **law of segregation** (Mendel's first law) states that...
  - Organisms inherit two copies of each gene, one from each parent
  - Organisms give only one copy of each gene to their gametes
    - The two copies of each gene segregate/separate during gamete formation

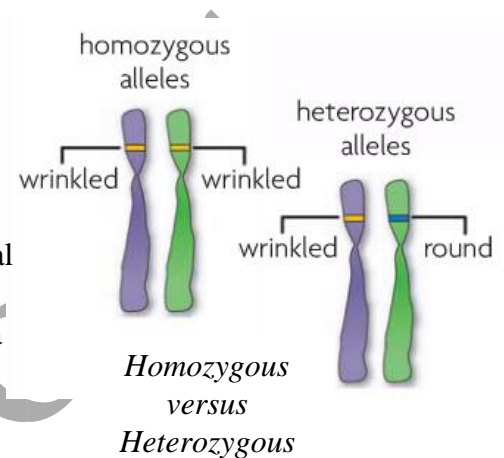


*Hidden traits reappeared in the F<sub>2</sub> generation*



### Alleles and Genes

- **Genes** = Pieces of DNA which provide instructions for making certain proteins
- Each gene has a **locus**, which is a specific position on a pair of homologous chromosomes
  - Tells where a gene is located on a chromosome
- **Alleles** are different forms of a gene located at a specific locus
  - Each gene has two alleles, one on each of your homologous chromosomes
- During fertilization, each parent gives one allele
- **Homozygous** = Two of the same alleles at the same locus
  - Ex: CC (Dominant) or cc (Recessive)
- **Heterozygous** = Two different alleles are the same locus
  - Ex: Cc
- All of an organism's genetic data is known as a **genome**
  - Everybody except for identical twins has an identical genome
- The genetic makeup of a specific set of genes is known as a **genotype**
  - Ex: All of the genes (even masked ones) which code for flower color
- The physical characteristics produced by the genotype are known as the **phenotype**
  - Ex: A flower having purple petals despite the presence of a hidden gene for white petals



### Dominant and Recessive Alleles

- **Dominant** alleles are expressed in the phenotype even when only one is present
  - Represented by uppercase letters
  - Ex: Cc, CC
- **Recessive** alleles are expressed in the phenotype only when two copies are present
  - Represented by lowercase letters
  - Ex: Cc, cc
- Despite overpowering recessive alleles, dominant alleles aren't necessarily present in more of the population or better than recessive alleles
  - For example, the allele polydactyly is dominant even though it isn't present in most of the population
- If the dominant allele P represents purple petals and p represents white petals, then P must code for pigment while p codes for nothing (the absence of pigment)
  - Even if the genotype is Pp, the dominant allele is still telling the cell to produce pigment
- Not all genotype are just dominant and recessive alleles
  - Codominance
  - Incomplete dominance

Punnett Squares and Monohybrid Crosses

- **Punnett Squares** are grids which are used to predict all possible genotypes which result from a cross
  - Developed by R. C. Punnett
- The ratio of genotypes in a generation can be found by counting the number of squares with a specific combination of alleles
- During meiosis, the alleles are separated
- Each gamete gets one allele
- **Monohybrid crosses** examine the inheritance of a single specific trait
- A **test cross** is a cross between an organism with an unknown genotype and a purebred organism with a recessive phenotype
  - The purebred organism must be homo. Recessive
  - Offspring will show the genotype of the organism in question

		Alleles from Father	
		A	a
Alleles from Mother	A	AA	Aa
	a	Aa	aa
		Pheno. Ratio – 3:1 Geno. Ratio – 1:2:1	

*Hetero x Hetero  
Monohybrid cross*

Dihybrid Crosses

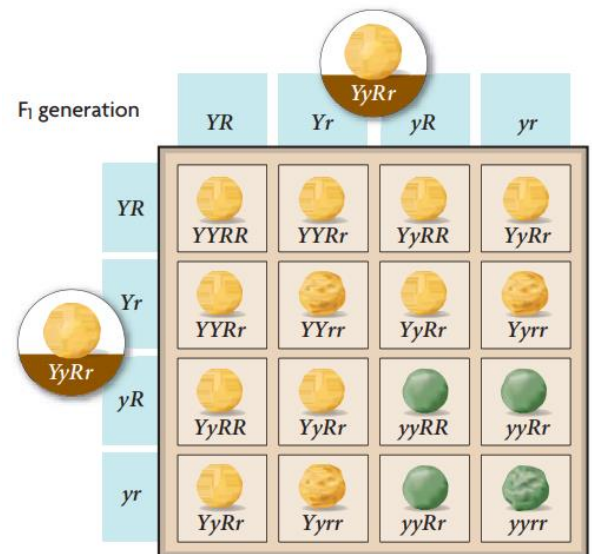
- **Dihybrid crosses** examine the inheritance of two different traits
- After finding a 9:3:3:1 phenotypic ratio in the F<sub>2</sub> generation when using purebred plants for the P generation, Mendel formed the...
- **Law of Independent Assortment** (Also known as Mendel's second law)
  - States that allele pairs separate independently of each other during meiosis, and different traits are inherited separately

Heredity and Probability

- The likelihood of an event occurring is **probability**
- $Probability = \frac{\text{number of ways that an event can occur}}{\text{number of total possible outcomes}}$



*Punnett  
Probability*



*F<sub>2</sub> generation  
A dihybrid cross*

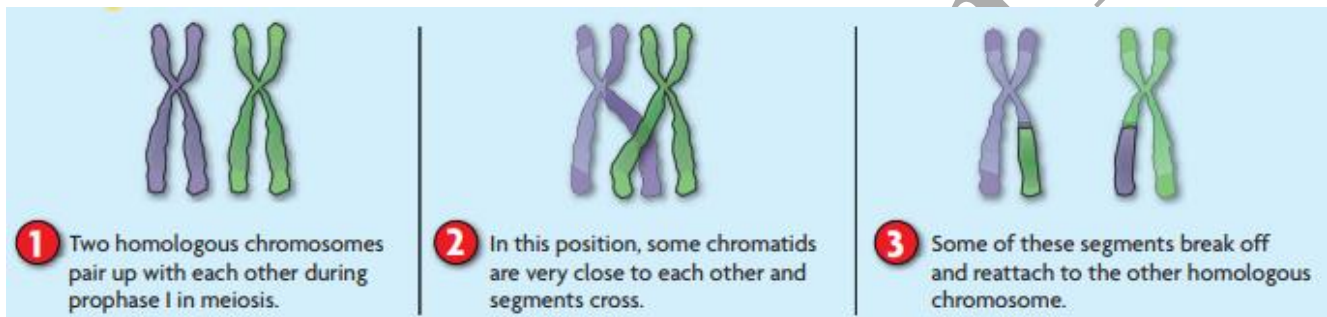


### Sexual reproduction and genetic variation

- Egg and sperm cells have  $2^{23}$  (around 8 million) combinations
- The total number of possible combinations formed from fertilization is around 70 trillion
- Sexual reproduction creates unique combinations of genes

### Crossing Over and Recombinant Chromosomes

- **Crossing over** is the exchange of gene segments between homologous chromosomes
- Since the chromatids are very close, part of each chromatid breaks off and reattaches to the other one
  - Known as **genetic linkage**
  - Results in recombinant chromosomes
- Crossing over happens whenever germ cells divide



### *The process of Crossing over*

- **Recombination** refers to the mixing of parental alleles
  - Also includes events other than crossing over
- The farther apart that genes are located, the more likely that they will separate when crossing over
  - More likely to assort independently
- Genes which are close together are inherited together, which is known as **genetic linkage**

### Incomplete Dominance

- The phenotype is in-between
- Red flower x white flower = 100% Pink Flowers

	R	R
W	RW	RW
W	RW	RW

### Incomplete vs. Codominance

Codominance: Both equally expressed  
Red flowers + White flowers =



Incomplete Dominance: alleles blend  
Red flowers + White flowers =



### *Incomplete vs Codominance*

Codominance

- Both phenotypes are equally present
- Homo. red bull x homo. white cow = 100% Roan Cows

	C <sup>r</sup>	C <sup>r</sup>
C <sup>w</sup>	C <sup>r</sup> C <sup>w</sup>	C <sup>r</sup> C <sup>w</sup>
C <sup>w</sup>	C <sup>r</sup> C <sup>w</sup>	C <sup>r</sup> C <sup>w</sup>

Blood Typing

- Blood is codominant
- Four blood types expressed by antigens (glycoproteins) on red blood cells
  - A, B, AB and O
- There are three alleles; A, B and O
  - However, you can only have two
  - Expressed as I<sup>A</sup>, I<sup>B</sup>, and i
- If you give somebody the wrong blood type, it will coagulate (clump)

**Cross a Homozygous B male with a heterozygous A female**

	I <sup>B</sup>	I <sup>B</sup>
I <sup>A</sup>	I <sup>A</sup> I <sup>B</sup>	I <sup>A</sup> I <sup>B</sup>
i	I <sup>B</sup> i	I <sup>B</sup> i

Phenotype:  
AB = 50%  
B = 50%

	Type A	Type B	Type AB	Type O
Genotype	I <sup>A</sup> I <sup>A</sup> or I <sup>A</sup> i	I <sup>B</sup> I <sup>B</sup> or I <sup>B</sup> i	I <sup>A</sup> I <sup>B</sup>	i i
Antigens	A antigen	B antigen	A & B antigens	None
Antibodies	Anti-B	Anti-A	None	Anti-A & Anti-B

*Blood Types, Genotypes, Antigens and Antibodies*

- There is more than one blood type due to mutation, natural selection, illness, and cultural isolation

Rh Factor

- Another protein on red blood cells
  - Named after the Rhesus Monkey
- 85% of humans have the Rh factor proteins
- Rh+ signifies that you have the protein
- Rh- signifies that you don't have it

[Dihybrid] Hetero. A+ father x Hetero AB+ mother

	A+	O-	Phenotype 50% A+ 25% AB+ 25% B-
A+	A+A+	A+O-	
B-	A+B-	B-O-	

Rh and Pregnancy

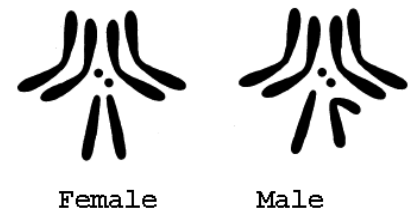
- When a Rh- mother is pregnant with an Rh+ child, the fetal blood crosses the placenta
- The mother's immune system sees the antigens as foreign invaders and begins to build up anti-Rh antibodies
  - Nothing happens to the first baby, since the antibodies haven't had enough time to build up
- During the next pregnancies, the antibodies can enter the fetus' immune system and cause blood coagulation
  - This is known as Rh disease

TYPE	CAN GIVE TO	CAN RECEIVE FROM
A+	A+, AB+	A+, A-, O+, O-
O+	O+, A+, B+, AB+	O+, O-
B+	B+, AB+	B+, B-, O+, O-
AB+	AB+	EVERYONE
A-	A+, A-, AB+, AB-	A-, O-
O-	EVERYONE	O-
B-	B+, B-, AB+, AB-	B-, O-
AB-	AB+, AB-	AB-, A-, B-, O-

*Blood Donor compatibility*

Sex Chromosomes

- Thomas Morgan (USA) studied genetics in the 1900s
  - Studied the Drosophila Melanogaster (fruit fly)
  - Discovered the X and Y chromosomes
- Fruit flies have 4 pairs of homologous chromosomes
  - 3 pairs of autosomes and 1 pair of sex chromosomes
- Mendel didn't study sex chromosomes since pea plants don't have sex chromosomes



Female

Male

*The chromosomes of a drosophila melanogaster*

**Cross a male and a female**

	X	Y	
X	X X	X Y	50% Male 50% Female
X	X X	X Y	

- Morgan observed eye color in fruit flies
  - Red and white
- The typical phenotype is known as the **wild type**
- Morgan also found that the genes for the fruit flies' eye color are stored in the "X" sex chromosome
  - Not true for humans

**Cross a white eyed male with a red eyed female**

	X <sup>r</sup>	Y	
X <sup>R</sup>	X <sup>R</sup> X <sup>r</sup>	X <sup>R</sup> Y	50% red eyed 50% white eyed
X <sup>R</sup>	X <sup>R</sup> X <sup>r</sup>	X <sup>R</sup> Y	

Sex-Linked Genes

- The X chromosome has more influence than the Y chromosome since it's longer and contains more genes
- It has been shown that over the years, bits of the Y chromosome have been absorbed by the X chromosome
- Men, who have only one X chromosome, are more susceptible to X-linked disease