Chapter 14 and 15 - Genetics

History – Mendel and His Peas

- When Mendel lived no one knew about DNA or meiosis.
- It was known that offspring inherited traits from the parents but no one knew how.
- It was thought that whatever the genetic material was, it would be blended to produce the offspring.
- But nature did not seem to follow this rule.

- If the genetic material was blending to produce offspring then why isn’t the offspring grey?
- Even though people could see this in nature and in agricultural breeding programs, they still believed in the blending theory.

Charles Darwin

- Charles Darwin was one of the few scientists who did not believe in the blending theory.
- Darwin believed that individuals in a population show variation.
- The traits that give you an edge to survive will be passed on to your offspring.
- The traits are not blended, instead the traits will be seen more or less often depending on how advantageous they are for the individual.

Mendel

- Just before Darwin presented his theory, Mendel started to work on his experiments with peas.
- Mendel was a monk and a scientist.
- He was raised on a farm and was aware of agricultural practices and research.
- He was well known for breeding new varieties of fruits and vegetables.
- Mendel attended the University of Vienna and studied both math and botany.
- Mendel believed that sperm and eggs contained "units" of information or traits.
- He used pea plants to prove his theory.
- Pea plants usually self-fertilize.

Some Terminology

- **Traits** – The inherited characteristics that vary between individuals.
- **Phenotype** – Physical features of the organism.
- **Genotype** – The genetic makeup that determines the phenotype.
- Think of the XX and XY of the sex chromosomes. This is the genotype that produces either male or female offspring.
- **Parental generation (P):** The parents in Mendel’s experiments. These plants are “pure” “true-breeding” = they only produce plants with their own phenotype when they are self-pollinated

- **First filial generation (F₁):** The offspring of the parental generation

- **Second filial generation (F₂):** The offspring of the first filial generation

- **Hybridization or Reciprocal cross:** Breeding between two different types of true breeding plants

- **Hybrid:** offspring of two genetically different parents

- **Monohybrid cross:** only follows one trait in the breeding – purple vs white flowers

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**Mendel’s Experiments**

- Mendel did a reciprocal cross: he took true-breeding purple flower plants and crossed them with true-breeding white flower plants = (P generation)

- The result was all plants that had purple flowers = F₁

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**Mendel’s next experiment**

- Mendel planted the peas and grew up the plants – these are F₁ plants

- He allowed these F₁ plants to self-pollinate themselves

- The pollen from the plants fertilized the same plants’ eggs

- The plants produced some plants with purple flowers and some plants had white flowers (F₂).
### Results from second experiment

- He counted all the plants from the experiment and he got about a 3:1 ratio of plants with purple flowers:white flowers.
- He did these same experiments over again with other traits and got the same results. Always a 3:1 ratio.

### Dominant and Recessive

- The trait that is expressed is the dominant trait.
- The trait that is hidden, or not expressed is recessive.
- In this example the yellow trait is the dominant trait over green which is recessive.

### DNA and Genes

- **Gene**: the part of DNA that codes for a protein.
- **Locus**: the location of a gene on the chromosome.
- **Alleles**: the alternate forms of a gene, for example the dominant allele is purple flowers, and the recessive allele is white.
Principle of Segregation

- Before sexual reproduction can take place, the alleles in a parent must separate – this takes place during Anaphase I of Meiosis.
- So an egg or sperm only has one of the two alleles.
- The alleles are joined together during fertilization.

Principle of Independent Assortment

- When the Alleles segregate, the alleles will segregate randomly from other alleles.
- Think back to Metaphase I, when the chromosomes lined up and segregated.

Homozygous and heterozygous

- If both the chromosomes in the pair have the same allele form, then they are homozygous for that trait – just for that trait, other alleles on the DNA can be different.
- If the chromosomes in the pair have different alleles they are heterozygous for this trait.
- (Note: this is different than homologous chromosomes – the alleles are on homologous chromosomes.)

Monohybrid Cross

- This is the simplest cross between two alleles of the same locus.
- For example if the gene to code for green peas is the same gene that codes for yellow, then breeding two plants would be a monohybrid cross for pea color.

Punnett Squares

- Dominant alleles are written with a capitol letter.
- Recessive alleles are written with a small letter.
- Always use the same letter for a trait.
- Give a key for the symbols you are using.
- In Mendel's peas, the allele for yellow peas are written as Y and allele for green peas are written as y.

A yellow pea can be YY or Yy.
A green pea is yy.
**Genotype and Phenotype**

- **Phenotype** – physical features of the organism
- **Genotype** – the genetic makeup that determines the phenotype

- The phenotype for a yellow pea is yellow color, and the genotype is YY or Yy
- The phenotype for a green pea is green color, and the genotype is yy

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**Figure 14.5-1**

- **P Generation**
  - Appearance: Purple flowers
  - Genetic makeup: PP
  - Gametes: P

- **P Generation**
  - Appearance: White flowers
  - Genetic makeup: pp
  - Gametes: p

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**Figure 14.5-2**

- **P Generation**
  - Appearance: Purple flowers
  - Genetic makeup: PP
  - Gametes: P

- **F₁ Generation**
  - Appearance: Purple flowers
  - Genetic makeup: Pp
  - Gametes: 1/2 P, 1/2 p

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**Punnett Sq - Pea Example**

- The yellow allele is dominant (Y) and green allele is recessive (y)
- In Mendel’s experiment the parental generation (P) when they were self fertilized they only produced the same kind of plant.
- So if you hybridize true breeding plants with yellow peas (YY) and the plants with green peas (yy)
- When P generation were cross fertilized (yellow pea plants with green pea plants) they produced only yellow plants

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<th>Green</th>
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<tbody>
<tr>
<td>Yellow</td>
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<tr>
<td>Yellow</td>
<td>Yy</td>
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- The offspring of the P generation experiment were all Yy genotype and yellow phenotype

- These peas were planted, the plants grew up and were self fertilized to produce the F₂ generation

- F₂ generation

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- Phenotype of the offspring:
  - On average, 3 peas should be yellow (YY or Yy) and 1 pea should be green (yy)

- Test Cross

- It is not always possible to tell the genotype of an organism

- Yellow pea plants may be YY or Yy

- To determine the genotype you can do a test cross between the unknown genotype and a homozygous recessive individual

- If you cross pollinated homozygous purple plants with homozygous white plants (P generation) and the result was offspring that were all purple (F₁). Which allele is dominant?

1. Purple
2. White
What are the genotypes of the P generation?

1. PP and pp
2. PP and Pp
3. Pp and Pp
4. Pp and pp

What are the genotypes of the F1 generation?

1. PP and pp
2. PP and Pp
3. Pp and Pp
4. Pp and pp

Monohybrid Cross

- Mendel derived the law of segregation by following a single character
- The F₁ offspring produced in this cross were monohybrids, individuals that are heterozygous for one character
- A cross between such heterozygotes is called a monohybrid cross

Dihybrid Crosses

- This is a cross between individuals with different alleles at two loci
- If the two pairs of alleles are on nonhomologous, then each pair will be inherited independently

Dihybrid Cross Example

- In pea plants, round peas are dominant over wrinkled and yellow peas are dominant over green
  - Y = yellow
  - y = green
  - R = round
  - r = wrinkled

Parental Generation Dihybrid Cross

- True breeding yellow round pea producing plants are crossed with true breeding green wrinkled pea producing plants:
  - RRYY x ryy
  - The gametes will be:
    - RY for the yellow round and
    - ry for the green wrinkled plants
  - All the offspring will be RrYy
**F1 Generation Cross**

- RrYy x RrYy

- The gametes can be:
  - RY, Ry, rY, ry

**Dihybrid Cross Example**

- In guinea pigs, coat color and coat length are inherited from alleles on non-homologous chromosomes, and black (B) is dominant to brown (b) and short coat (S) is dominant to long coat (s)

- When a homozygous black, homozygous short coated individual is bred to a brown, long coated individual (BBSS x bbss) the resulting genotypes are:
  - All the offspring will have BbSs genotype and will be black short coated

**Dihybrid Cross Example Cont**

- If the F1 generation are bred together then each F1 guinea pig will have an equal probability of producing the following gametes:
  - BS, Bb, bS, and bs
Gametes formed by segregation and independent assortment of alleles

- F₂ generation (cont’d next slide)

- The laws of probability govern Mendelian inheritance
  - Mendel’s laws of segregation and independent assortment reflect the rules of probability
  - When tossing a coin, the outcome of one toss has no impact on the outcome of the next toss
  - In the same way, the alleles of one gene segregate into gametes independently of another gene’s alleles

- Probability
  - Probabilities are calculated as fractions (1/4) or decimals (0.25)
  - The Punnett square lets you calculate the probabilities of offspring from a cross

- The Multiplication or Product Rule
  - The multiplication or product rule: predicts the combined probability of independent events by multiplying the individual probabilities.
  - What is the probability that Pp x Pp will produce pp?
  - Each parent has ½ probability of contribution p so ½ x ½ = ¼
The Addition or Sum Rule

- The addition or sum rule: predicts the probability of mutually exclusive events by adding the probabilities
- Example: What is the probability that two Bb parents will have a Bb child?
  - Either a B sperm combines with a b egg OR a B egg combines with a b sperm: $\frac{1}{4} + \frac{1}{4} = \frac{1}{2}$

Dihybrid probabilities

- R = round
- r = wrinkled
- Y = yellow
- y = green

- Example: RrYy x RrYy
  - You can do them separately then multiply:
  - What is the probability of getting a rryy offspring?

- The probability of getting a wrinkled pea = $\frac{1}{4}$
- The probability of getting a green pea = $\frac{1}{4}$
- What is the probability of getting a rryy offspring? $\frac{1}{4} \times \frac{1}{4} = \frac{1}{16}$

Linked Genes

- If different genes have loci on the same chromosome then they do not follow the rules of independent assortment
- There still can be variation due to crossing over – creating recombinant types of gametes

Animation: Crossing Over

Right-click slide / select "Play"
The allele for short coat (S) is dominant to the allele for long coat (s) flowers. At another chromosome, the allele for black color (B) is dominant over brown (b). A animal that is heterozygous short coat and brown, is crossed with an animal that has a long coat and is brown, what proportion of their offspring will be black with long coats?

1. 0
2. 1/4
3. 1/2
4. 1

The allele for short coat (S) is dominant to the allele for long coat (s) flowers. At another chromosome, the allele for black color (B) is dominant over brown (b). A animal that is heterozygous short coat and brown, is crossed with an animal that has a long coat and is brown, what proportion of their offspring will be black with long coats?

Exceptions to Mendel’s Genetics

- Mendel’s peas showed dominance – if there was an allele for yellow the plant produced yellow peas. You only saw green peas if there were two green alleles
- This is not the case for all organisms

Extending Mendelian Genetics for a Single Gene

- Inheritance of characters by a single gene may deviate from simple Mendelian patterns in the following situations:
  - When alleles are not completely dominant or recessive
  - When a gene has more than two alleles
  - When a gene produces multiple phenotypes

Degrees of Dominance

- **Complete dominance** occurs when phenotypes of the heterozygote and dominant homozygote are identical
- In **incomplete dominance**, the phenotype of F₁ hybrids is somewhere between the phenotypes of the two parental varieties
- In **codominance**, two dominant alleles affect the phenotype in separate, distinguishable ways
Incomplete Dominance

- In peas everything worked out neatly but in other plants like snapdragons it does not work like this. If you cross a red snapdragon with a white snapdragon you get all pink snapdragons.

- Red is dominant, but not completely dominant so $C^R = \text{red}$, $C^W = \text{white}$.

Co-Dominance and Multiple alleles

- Another type of dominance is co-dominance

- Here both the alleles are expressed in heterozygous organisms

Blood Type Co-dominance

- What does your blood type refer to?
- It is the type of glycoproteins on the surface of the blood cells.
- The 9th pair of chromosome in humans contains the gene that codes for the enzyme that attaches the carbohydrate to the protein. Each chromosome in the pair will have a different gene or allele for the enzyme.
- There are two main glycoproteins on blood cells: A and B, neither type is dominant over the other
Type A has only A glycoproteins
Type B has only B glycoproteins
Type AB has A and B glycoproteins
O Type has neither glycoproteins

(a) The three alleles for the ABO blood groups and their carbohydrates

<table>
<thead>
<tr>
<th>Allele</th>
<th>( I^A )</th>
<th>( I^B )</th>
<th>( i )</th>
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<tbody>
<tr>
<td>Carbohydrate</td>
<td>A △</td>
<td>B ○</td>
<td>none</td>
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(b) Blood group genotypes and phenotypes

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Red blood cell appearance</th>
<th>Phenotype (blood group)</th>
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<tbody>
<tr>
<td>( I^A I^A ) or ( I^A i )</td>
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The Relation Between Dominance and Phenotype

- A dominant allele does not subdue a recessive allele; alleles don’t interact that way
- Alleles are simply variations in a gene’s nucleotide sequence
- For any character, dominance/recessiveness relationships of alleles depend on the level at which we examine the phenotype

Level of Examination

- **Tay-Sachs disease** is fatal; a dysfunctional enzyme causes an accumulation of lipids in the brain
  - At the *organismal* level, the allele is recessive
  - At the *biochemical* level, the phenotype (i.e., the enzyme activity level) is incompletely dominant
  - At the *molecular* level, the alleles are codominant

If one parent has heterozygous type A and the other has type O blood, what are the phenotypes of the offspring

1. Type A
2. Type B
3. Type AB
4. Type O
5. Type A or O
If the parents have type A (homozygous) and type B (homozygous) blood, what are the phenotypes of the offspring?

1. Type A
2. Type B
3. Type AB
4. Type O

25% 25% 25% 25%

If the parents have type AB and type O blood, what are the phenotypes of the offspring?

1. Type A
2. Type B
3. Type AB
4. Type O
5. Type A and B
6. Type AB, B and A

17% 17% 17% 17% 17%

Multiple Alleles

- In Blood Types there are three possible alleles: A, B or O
- Each person can only possess two, but there exists more that two alleles in a population.

Mendelian Genetics for Two or More Genes

- Some traits may be determined by two or more genes

Interaction between genes

- Several pairs of genes may interact to affect a single phenotype

Interaction between genes:
Interaction between genes

- **Epistasis** – when an allele at one locus can prevent the expression of an allele at a different locus.
- **Labrador coat color:**
  - \( B = \) black, \( b = \) brown
  - Allele for depositing the color is \( E = \) expresses the color, and \( e = \) suppresses the color
  - Yellow: \( BBee, Bbee, bbee \)
  - Chocolate: \( bbEE, bbee \)
  - Black: \( BBEE, BbEE, BbEe \)

Polygenes

- Polygenes act additively to produce a phenotype
- Certain traits like height and skin color are controlled by multiple genes acting and different loci
- Skin color is controlled by as many as 60 genes at different loci

Genes and the Environment

- The phenotype is not completely controlled by genes. Think about the height of a person. Part of what determines the height of that person is genetic, but diet also plays a roll.
- The color of Hydrangea flowers depends on the soil. Acidic soil produces blue flowers, basic soil produces pink flowers
Integrating a Mendelian View of Heredity and Variation

- An organism’s phenotype includes its physical appearance, internal anatomy, physiology, and behavior
- An organism’s phenotype reflects its overall genotype and unique environmental history

Pedigree Analysis

- A pedigree is a family tree that describes the interrelationships of parents and children across generations
- Inheritance patterns of particular traits can be traced and described using pedigrees

Autosomal Genetic Disorders

- Sex-linked disorders are only those disorders that are controlled by the X or Y genes
- All other chromosomes are autosomal chromosomes – they control the autosomal disorders
- There are recessive autosomal disorders, and dominant autosomal disorder

Autosomal Recessive Disorders

- Autosomal recessive disorders: These are disorders that are controlled by DNA on any of the non-sex chromosomes (22 homologous autosomal chromosomes)
- These are recessive disorders so they are only expressed if the person is homozygous for the allele
- Examples include: sickle-cell anemia, cystic fibrosis, albinism, phenylketonuria

Cystic Fibrosis

- Cystic fibrosis is the most common lethal genetic disease in the United States, striking one out of every 2,500 people of European descent
- The cystic fibrosis allele results in defective or absent chloride transport channels in plasma membranes leading to a buildup of chloride ions outside the cell
- Symptoms include mucus buildup in some internal organs and abnormal absorption of nutrients in the small intestine

Figure 14.15

Key

- Male  Female  Affected male  Affected female  Mating  Offspring

1st generation

2nd generation

3rd generation

Widow’s peak

No widow’s peak

Attached earlobe

Free earlobe

(a) Is a widow’s peak a dominant or recessive trait?

(b) Is an attached earlobe a dominant or recessive trait?
Phenylketonuria

- Autosomal recessive disorder
- Lack of a certain enzyme causes a defect in amino acid metabolism
- Individuals can not convert phenylalanine into tyrosine, so phenylalanine builds up in the body
- Phenylalanine converted into phenylketones, which damage the nervous system of developing children
- Simple blood test at birth detects disorder, then restrictive diet prevents mental retardation

Tay-Sachs Disease

- Autosomal recessive disease
- Affects the central nervous system
- Results in blindness and mental retardation, early death.
- People with this disease lack an enzyme normally found in lysosomes in brain cells. This enzyme normally breaks down a membrane lipid.

Sickle-Cell Disease: A Genetic Disorder with Evolutionary Implications

- **Sickle-cell disease** affects one out of 400 African-Americans
- The disease is caused by the substitution of a single amino acid in the hemoglobin protein in red blood cells
- In homozygous individuals, all hemoglobin is abnormal (sickle-cell)
- Symptoms include physical weakness, pain, organ damage, and even paralysis

Sickle-Cell Disease: A Genetic Disorder with Evolutionary Implications

- Heterozygotes (said to have sickle-cell trait) are usually healthy but may suffer some symptoms
- About one out of ten African Americans has sickle cell trait, an unusually high frequency of an allele with detrimental effects in homozygotes
- Heterozygotes are less susceptible to the malaria parasite, so there is an advantage to being heterozygous

If two people that are heterozygous for cystic fibrosis have children, what is the chance they will have a child with cystic fibrosis?

1. 0
2. 1/4
3. 1/2
4. 1
**Dominant Autosomal Disorders**

- Dominant Autosomal disorders are those disorders control by the non-sex chromosomes.
- These disorders will be expressed when the person has one or two alleles for the disorder. The allele for the disorder is dominant over the normal allele.
- Examples include: Huntington disorder, cholesterolemia, achoo syndrome, achondroplasia.

**Huntington Disorder**

- Huntington disorder is a dominant autosomal disorder so in a Punnetts square the Huntington allele is written H and the normal allele is h.
- A person with the genotype Hh or HH will develop Huntington disorder. People with hh will not develop the disorder.
- People with Huntington disorder do not usually show symptoms until after they have reproduced.

**Purple flowers are dominant, a cross between homozygous purple plants with white plants, what is the probability of the F1 having purple flowers**

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**Phenylketonuria is a recessive autosomal disorder, if a woman, whose father had PKU, marries a man with PKU, what is the probability of their child will have PKU**

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<td>4.</td>
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Two people are both heterozygous for Huntington's, a dominant disorder, what is the probability that their next child will have the disease?

1. 0
2. 1/4
3. 3/4
4. 1

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**X-Linked Inheritance in Humans**

- Remember that XX and XY are the sex chromosomes.
- Genes that are located on the X chromosomes are called X-linked or sex-linked.
- X-linked recessive traits are mainly seen in males – Why?

**X-Linked Inheritance in Humans**

- Males only have one X chromosome, so if there is a trait on the X chromosome and they inherit the recessive allele, then they will express the trait.
- If females inherit one recessive allele, they have a chance to also inherit the dominant allele.
- There are many disorders that are X-linked: color blindness, hemophilia, muscular dystrophy

**X-Linked Color Blindness**

- Color blind people can not distinguish between certain shades of greens and reds
- In color blindness the proteins in pigments that absorb green and red light are controlled by DNA on the X chromosome
- There are two alleles for this – the dominant one produces the proteins, the recessive allele does not.
- This disorder is a recessive disorder which means that it will only express if it is homozygous for the recessive allele
Can you see all the shades of colors in the picture?

1. Yes
2. No

X-linked traits

- When drawing Punnett Squares:
  - Normal X = X^B
  - X carrying the trait = X^b
  - Normal Female: X^B X^B
  - Female Carrier: X^B X^b
  - Color blind Female: X^b X^b
  - Normal Male: X^B Y
  - Color blind male: X^b Y

Question

- Red-green color blindness is due to a sex-linked recessive allele on the X chromosome. Two normal visioned parents produce a color-blind son.

- Draw a punnett square to show how this happened

What is the chance the couple could have a color blind daughter?

1. 1/4
2. 2/4
3. 3/4
4. 0/4 (none)

X chromosomes in Males and Females

- The X chromosome has genes needed by both sexes but females have two chromosomes, where as males have one X chromosome.
- In some species, the males compensate by having a more active X chromosome.
- In most mammals, one of the X chromosomes in females gets turned off in most cells. The inactive X is called a Barr body. It is random which X will be inactivated in each cell.

X Inactivation in Female Mammals

- In mammalian females, one of the two X chromosomes in each cell is randomly inactivated during embryonic development
- The inactive X condenses into a Barr body
- If a female is heterozygous for a particular gene located on the X chromosome, she will be a mosaic for that character

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Normal Chromosome Number

- Humans have 46 chromosomes, 23 pairs
- 22 pairs are homologous, autosomal
- 1 pair is non-homologous, sex chromosomes

Abnormal Chromosome Number

- In **nondisjunction**, pairs of homologous chromosomes do not separate normally during meiosis
- As a result, one gamete receives two of the same type of chromosome, and another gamete receives no copy
Abnormalities in Chromosome Number

- **Aneuploidy** results from the fertilization of gametes in which nondisjunction occurred.
  - Offspring with this condition have an abnormal number of a particular chromosome.

Aneuploidy

- A **monosomic** zygote has only one copy of a particular chromosome.
- A **trisomic** zygote has three copies of a particular chromosome.

Abnormalities in Chromosome Number

- **Polyplody** is a condition in which an organism has more than two complete sets of chromosomes.
  - Triploidy (3n) is three sets of chromosomes.
  - Tetraploidy (4n) is four sets of chromosomes.
  - Polyplody is common in plants, but not animals.

Abnormalities in Chromosome Number

- Remember back to meiosis when gametes are formed.
  - In meiosis I the pairs of chromosomes line up at the equator and one chromosome from each pair is pulled to the poles.
  - In meiosis II the 23 chromosomes line up at the equator and the chromatids are pulled to the poles.
  - If during either of these events the chromosomes or chromatids don’t separate properly aneuploidy will occur.

Abnormalities in Chromosome Number

- Polyplody – More likely to occur due to two sperm fertilizing an egg.
- Aneuploidy – More likely to occur due to a chromosome failing to separate properly during meiosis = nondisjunction. Can occur during meiosis I or II.
- So if a gamete does not go through meiosis properly and ends up with 22 or 24 chromosomes (instead of the normal 23)
- Then when the gametes combine the offspring will have 45 or 47 chromosomes (instead of 46)
- Remember that this can happen during either meiosis I or meiosis II
- Most of the time an embryo with aneuploidy will be miscarried

**Down Syndrome**

- Down syndrome is caused by aneuploidy
- People with down syndrome have three of the 21st chromosome instead of the normal two chromosomes = trisomy
- Down syndrome results in mental retardation, facial abnormalities, susceptible to certain cancers and Alzheimer’s

**Sex Chromosome Aneuploidies**

- Sex chromosome aneuploidies are usually less severe than autosomal aneuploidies.
- Remember Barr bodies form from one copy of X
Klinefelter Syndrome

- Sex chromosome aneuploidy
- Males who have 47 chromosomes, XXY
- Symptoms: small testes, sterile, tall, breast development, may have mental retardation
- Have Barr bodies in cells, therefore genetically test as female

Turner Syndrome

- Sex chromosome aneuploidy
- Females who have 45 chromosomes, XO
- Symptoms: internal and external genitalia underdeveloped, sterile
- Do not have Barr bodies in cells, therefore genetically test as male

XYY Karyotype

- Sex chromosome aneuploidy
- Men who have 47 chromosomes, XYY
- Symptoms: tall, acne

Changes in the Chromosome Structure

- During Meiosis I when there is crossing over or at other times during interphase or Meiosis the chromosomes may be damaged.
- Examples of types of damage include: deletion, inversion, translocation, and duplication
- Causes can include: radiation, chemicals or chance

Alterations of Chromosome Structure

- Breakage of a chromosome can lead to four types of changes in chromosome structure
  - Deletion removes a chromosomal segment
  - Duplication repeats a segment
  - Inversion reverses orientation of a segment within a chromosome
  - Translocation moves a segment from one chromosome to another
Cri-du-chat Syndrome

- Cri-du-chat syndrome is caused by a deletion in part of chromosome number 5
- Children with this disorder have a defect in their larynx that causes them to make cat sounds. This effect goes away by the age of 4
- Other effects of the syndrome are life-long including mental retardation

Fragile X Syndrome

- A fragile site is a place on a chromosome where part of the chromatid seems to be attached by a thin thread.
- When the fragile site is at the end of the X chromosome it results in Fragile X syndrome.
- The fragile site contains a repeat of bases
- More pronounced in males
- Mild to severe mental retardation

Genetic Testing and Counseling

- Genetic counselors can provide information to prospective parents concerned about a family history for a specific disease

Tests for Identifying Carriers

- For a growing number of diseases, tests are available that identify carriers and help define the odds more accurately

Types of Genetic Screening

- Carrier Screening
- Prenatal screening – amniocentesis and CVS
- Newborn screening
- Pre-implantation Screening – testing embryos before implanting them in the uterus
Inheritance of Organelle Genes

- Extranuclear genes (or cytoplasmic genes) are found in organelles in the cytoplasm.
- Mitochondria, chloroplasts, and other plant plastids carry small circular DNA molecules.
- Extranuclear genes are inherited maternally because the zygote’s cytoplasm comes from the egg.
- The first evidence of extranuclear genes came from studies on the inheritance of yellow or white patches on leaves of an otherwise green plant.

Some defects in mitochondrial genes prevent cells from making enough ATP and result in diseases that affect the muscular and nervous systems.
- For example, mitochondrial myopathy and Leber’s hereditary optic neuropathy.

Dominant Autosomal Disorders

- Dominant Autosomal disorders are those disorders control by the non-sex chromosomes.
- These disorders will be expressed when the person has one or two alleles for the disorder. The allele for the disorder is dominant over the normal allele.
- Examples include: Huntington disorder, cholesterolemia, Achoo syndrome.
**Huntington Disorder**

- Huntington’s disease – It is a degenerative disease that affects the cerebral cortex region of the brain.
- Initial symptoms are abrupt, jerky movements, these symptoms typically develop in middle age. Late in the disease dementia occurs.
- It is caused from a repeat of three bases of the DNA on chromosome 4. This part of the DNA protein important in the functioning of an enzyme important gene expression.

**Huntington Disorder**

- Huntington disorder is a dominant autosomal disorder so in a Punnett square the Huntington allele is written H and the normal allele is h.
- A person with the genotype Hh or HH will develop Huntington disorder. People with hh will not develop the disorder.
- People with Huntington disorder do not usually show symptoms until after they have reproduced.

**Would you want to know if you have the Huntington gene**

1. Yes
2. No

**This is a disorder resulting in a female with a XO genotype:**

1. Down’s syndrome
2. Turner syndrome
3. Huntington's
4. Klinefelter syndrome

**What is an example of a dominant autosomal genetic disorder?**

1. Down’s syndrome
2. Color blindness
3. Huntington's
4. Cri-du-chat
Ethical Dilemmas
- Science gives us the tools to diagnose certain disorders but it is up to us to decide the ethical use of these tools.
- Couples need to decide what risks they are willing to live with when conceiving and what to decide in the event of a bad outcome of a prenatal test.
- Would you want to know if you are a carrier for a recessive autosomal disorder?
- Would you want to have prenatal screening done even if you know you will not abort the fetus no matter what the result?
- What should the criteria be for deciding what children get newborn screening.
- Do you think that pre-implantation diagnosis is a good idea? What are some things you might want to consider before doing this procedure?

Important Concepts
- Know the terminology for genetics
- Understand the principles of segregation and independent assortment
- Know what monohybrid crosses, test crosses and dihybrid crosses are
- Understand the product and sum rules
- Know what linked genes are and how the effect inheritance of traits
- Understand how crossing over affects the inheritance of linked genes, be able to interpret the results of a genetics experiment – determining if the genes were linked and if crossing over occurred
- X-Linked Inheritance – how are traits passed to offspring on the X chromosome, why are males more likely to be effected, color blindness as an example
- Know how organisms compensate for the fact that females have two copies of X, and males have only one copy
- Understand the basics of incomplete dominance

Important Concepts
- Know the ABO blood types and codominance
- Know what epistasis is and how it effects inheritance
- Know what polygenes are
- Autosomal disorders – differences between recessive and dominant autosomal disorders, examples of each type
- Know all the genetic disorders discussed in this lecture, including the cause, effects, which are dominant autosomal, recessive autosomal, X linked disorders.
- Know disorders due to abnormal number or abnormal structure of chromosomes
- Know the basics of how aneuploidy can happen and that it can happen during Meiosis I or II
### Important Concepts

- Know the benefits of being heterozygous sickle cell anemia
- What is genetic counseling.
- What are the types of genetic screening (carrier, prenatal, newborn, and pre-implantation) and understand what each type of screening is.

### Lab Practical Important Concepts

- Be able to solve monohybrid and dihybrid cross problems (for the lab practical)
- Given the genotype (or phenotypes) of the parents, be able to determine the chance the offspring will inherit a disorder or will be a carrier or will not be either. You should be able to do this for X-linked, recessive autosomal, and dominant autosomal, co-dominant (blood types) (lab practical)
- Be able to solve genetic problems similar to the homework problems (these problems will be tested on the lab practical).