**Note**

- This lecture is given during laboratory time but the information will be covered on the lecture exams.
- You will be given a homework assignment which will have genetics problems. The homework problems will be on the laboratory exam.

**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.

**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. This was more common then.
- 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.

**Plant Characteristics**

- Pea shape – smooth or wrinkled
- Seed Color – yellow or green
- Pod Shape – smooth or wrinkled
- Pod Color – yellow or green
- Flower Color – purple or white
- Flower Position – low on stem or at tip
- Stem Length – tall or short
**Terminology**

- **Traits**: The 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.

**Mendel’s Experiments**

- Mendel took plants that produced only yellow peas and crossed them with plants that produced only green peas. These parent plants are P (parental generation).
- The result was all plants that had yellow peas. These plants are the F₁ (first filial generation).

**Result from First Experiment**

- **P generation**: All yellow or all green peas.
- The plants produced pea pods with all yellow peas.
- This would not be true if the genetic material were "blended". The fact that they were all yellow could not be by chance – he repeated the experiment many times.

**Mendel’s next experiment**

- Mendel planted the pea pods and grew up the plants – these are F₂ plants.
- He allowed these F₁ plants to self pollinate themselves.
- The pollen from the plants fertilized the same plants’ eggs.
- The plants produced pea pods, with some yellow peas and some green peas (F₂).

**Results from both experiments**

- P generation were all green bred to all yellow.
- F₁ generation were all yellow peas.
- F₂ generation had both yellow and greens (3:1 ratio).
- Some how the green trait had "hidden" for a generation.

**Table 11.2** Ratios of Dominant to Recessive in Mendel’s Plants

<table>
<thead>
<tr>
<th>Phenotypic trait</th>
<th>Genotypic trait</th>
<th>Ratio of dominant to recessive in F₂ generation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Smooth seed</td>
<td>wrinkled seed</td>
<td>2:1</td>
</tr>
<tr>
<td>White seed</td>
<td>Green seed</td>
<td>3:1</td>
</tr>
<tr>
<td>Yellow pod</td>
<td>White pod</td>
<td>2:1</td>
</tr>
<tr>
<td>Purple flower</td>
<td>White flower</td>
<td>3:1</td>
</tr>
<tr>
<td>Flower color:</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yellow</td>
<td>White</td>
<td>1:1</td>
</tr>
<tr>
<td>Tall</td>
<td>short</td>
<td>3:1</td>
</tr>
</tbody>
</table>

Average ratio, all traits: 3:1.
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.

Review of Meiosis

- Remember that at the end of meiosis, the gametes have halved the number of chromosomes (23 in humans), one of each in a pair of chromosomes.
- The chromosomes are not in the duplicated form.

DNA and Genes

- Gene: the part of DNA that codes for a protein.

Allele

- There are pairs of chromosomes. Pairs of homologous chromosomes will contain the code for the same proteins.
- In a pair of homologous chromosomes, each one contains the same genes. These different forms of the same genes are called alleles.

Genotype and Phenotype

- Dominant alleles are written with a capital letter.
- Recessive alleles are written with a small letter, always use the same letter.
- In Mendel's peas, the allele for yellow peas are written as Y and allele for green peas are written as y.

- 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.
**Punnett Sq - Pea Example**

- Remember that 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 only produced the same kind of plant.
- So the plants with yellow peas are YY and the plants with green peas are yy.

**F, Generation of Experiment**

- When P generation were cross fertilized (yellow pea plants with green pea plants) they produced only yellow plants.

<table>
<thead>
<tr>
<th>Y</th>
<th>Yy</th>
<th>Yy</th>
</tr>
</thead>
<tbody>
<tr>
<td>Y</td>
<td>Yy</td>
<td>Yy</td>
</tr>
<tr>
<td>Y</td>
<td>Yy</td>
<td>Yy</td>
</tr>
</tbody>
</table>

**F2 generation**

- 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.

**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 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.

- 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?
  A. Purple
  B. White

**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.
Can you see all the shades of colors in the picture?
A. Yes
B. No

X-linked traits
- When drawing Punnett Squares:
  - Normal X = X
  - X carrying the trait = X<sup>b</sup>
  - Normal Female: XX
  - Female Carrier: X<sup>b</sup>X
  - Color blind Female: X<sup>b</sup>X<sup>b</sup>
  - Normal Male: XY
  - Color blind male: X<sup>b</sup>Y

What is the chance the couple could have a color blind daughter?
A. 1/4
B. 2/4
C. 3/4
D. 0/4 (none)

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

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

Sickle Cell Anemia
- This is an autosomal recessive disorder.
- The protein in blood cells that carries the oxygen in hemoglobin.
- Sickle-cell disorder is due to a point mutation in the DNA that controls the formation of the hemoglobin protein – causing a misshaped red blood cells
- What is the chance that two carriers will produce a child with sickle cell anemia?
- A carrier is a person that does not express a recessive disorder but carries the allele for the disorder (heterozygous)
- To determine the chances that two carriers will pass on the disorder — do a Punnett's square

If two people that are heterozygous for Sickle Cell Anemia have children, what is the chance they will have a child with Sickle Cell Anemia?
- A. 0% (0/4)
- B. 25% (1/4)
- C. 50% (2/4)
- D. 75% (3/4)

- The child will have:
  - A 25% or 1 in four chance of having the disorder
  - A 50% chance of being a carrier
  - A 25% chance of being “normal”

The Benefits of Sickle-Cell
- There is an overlap of where sickle-cell anemia is found in the highest numbers and where malaria is found.
- People who are homozygous for the disorder experience severe symptoms, but people who are heterozygous (carriers) show few if any symptoms.
- But carriers of sickle-cell anemia have greater protection from malaria symptoms

Sickle-cell anemia is considered to be a recessive disorder since people with heterozygous alleles do not express the disorder, but they do produce some of the abnormal hemoglobin.
- People who are heterozygous are able to produce both the normal and abnormal hemoglobin proteins
- The abnormal protein protects against the affects of malaria

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.
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.

Would you want to know if you have the Huntington gene

A. Yes
B. No

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 RR = red, rr = white, but Rr = pink.

Blood Type Co-dominance

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

- Type A has only A proteins
- Type B has only B proteins
- Type AB has A and B proteins
- O Type has no proteins
### Genotype, Phenotype, and Blood Type

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype</th>
<th>Blood Type</th>
</tr>
</thead>
<tbody>
<tr>
<td>AB</td>
<td>A &amp; B proteins</td>
<td>AB</td>
</tr>
<tr>
<td>AA</td>
<td>A proteins</td>
<td>A</td>
</tr>
<tr>
<td>BB</td>
<td>B proteins</td>
<td>B</td>
</tr>
<tr>
<td>OO</td>
<td>No protein</td>
<td>O</td>
</tr>
<tr>
<td>BO</td>
<td>B proteins</td>
<td>B</td>
</tr>
<tr>
<td>AO</td>
<td>A proteins</td>
<td>A</td>
</tr>
</tbody>
</table>

### If the parents have type A (homozygous) and type O blood, what are the phenotypes of the offspring?

- **A.** Type A
- **B.** Type B
- **C.** Type AB
- **D.** Type O

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

- **A.** Type A
- **B.** Type B
- **C.** Type AB
- **D.** Type O
- **E.** Type A and B
- **F.** Type AB, B, and A

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

- **A.** Down’s syndrome
- **B.** Color blindness
- **C.** Huntington’s
- **D.** Cri-du-chat

### Aneuploidy

- Aneuploidy are disorders where a person has an abnormal number of chromosomes.
- Everyone should have 23 pairs of chromosomes or 46 chromosomes.
- Gametes should have 23 total chromosomes so when two gamete meet the offspring will have 46 chromosomes.
Down Syndrome

- Down syndrome is caused by aneuploidy
- People with down syndrome have three of the 21st chromosome instead of the normal two chromosomes.
- Down syndrome results in mental retardation

<table>
<thead>
<tr>
<th>Mother's age</th>
<th>Chances of giving birth to a child with Down syndrome</th>
</tr>
</thead>
<tbody>
<tr>
<td>20</td>
<td>1 in 1925</td>
</tr>
<tr>
<td>25</td>
<td>1 in 1205</td>
</tr>
<tr>
<td>30</td>
<td>1 in 885</td>
</tr>
<tr>
<td>35</td>
<td>1 in 365</td>
</tr>
<tr>
<td>40</td>
<td>1 in 110</td>
</tr>
<tr>
<td>45</td>
<td>1 in 32</td>
</tr>
</tbody>
</table>

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

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

Genetic Counseling

- What options do couples have to try to prevent having a baby with a birth defect?
- The first option is going to a genetic counselor who will help them draw a family tree of genetic traits
- The counselor may recommend getting tested to see if you are carriers for certain traits if the test is available
### Genetic screening
- Genetic screening is a process of analyzing blood or skin to search for a particular genotype.
- In order to locate this faulty gene, scientists search for variations in pieces of DNA called "markers".
- With such markers it becomes theoretically possible to screen individuals of every age, from infants to adults, even babies before birth.

### Genetic Screening Cont
- Markers have already been found for various diseases such as:
  - Huntington's disease, sickle cell anemia, cystic fibrosis, Tay-Sachs disease, Duchenne muscular dystrophy, hemophilia and thalassaemia, and some rare cancers.

### Types of Genetic Screening
- Carrier Screening
- Prenatal screening
- Newborn screening
- Pre-implantation Screening

### Carrier Screening
- Carrier screening is the analysis of adults to determine if they have a gene or a chromosome abnormality that may cause problems either for offspring or the person screened.
- What do you do if you find out you are a carrier?

### Prenatal Screening
- Prenatal screening is done when a fetus is at risk for various identifiable genetic diseases or traits.
- There are numerous tests that can be used:
  - maternal serum alpha-fetoprotein (MSAFP) screening
  - amniocentesis
  - chorionic villus sampling (CVS)

### maternal serum alpha-fetoprotein screening
- MSAFP is a prenatal blood-screening test performed at the 16-18 week gestation date and tests for spina bifida. Enhanced MSAFP is also a blood-screening test that measures levels of certain biochemical markers to test for the presence of Down's syndrome.
- This test only has an accuracy of 60-65%.
- Benefits: it is not invasive to the fetus.

### Amniocentesis
- Amniocentesis is a prenatal test where a sample of the amniotic fluid is removed from the mother using a needle.
- Amniocentesis performed at the 16-18 week of gestation uses amniotic fluid to test up to 180 genetic disorders.
- the risk of miscarriage due to amniocentesis is between one in 200 and one in 400, depending on the skill and experience of the doctor performing it.

### Amniocentesis cont
- Amniocentesis can identify several hundred genetic disorders, including some of the most common: including Down syndrome, cystic fibrosis, Sickle cell anemia, Tay-Sachs disease, or Huntington disorder. Neural tube defects such as spina bifida and anencephaly.
- The test is more than 99 percent accurate in diagnosing these conditions.
- Amniocentesis doesn't detect every birth defect.

### Amniocentesis
- Who should have this test?
- What if you know you will not abort the fetus no matter what the result?
Chorionic villus sampling

- Chorionic villus sampling (CVS) is a prenatal test done by analyzing cells taken from the chorionic villi — tiny fingerlike projections on the placenta.
- Its primary advantage over amniocentesis is that you can take it earlier — generally between 10 and 12 weeks of pregnancy.

CVS Cont

- CVS is better than 99 percent accurate at detecting hundreds of genetic disorders and chromosomal abnormalities.
- The test does not detect neural tube defects, such as spina bifida.
- There is a 1 percent chance of getting a false positive result in which some of the cells cultured from the placenta contain abnormal chromosomes but the fetus is normal.

Newborn Screening

- Newborn screening analyzes the blood or tissue samples taken in early infancy in order to detect genetic disorders for which early intervention can avert serious health problems or death.
- This started in 1960 with the ability to test newborns for a rare metabolic disease, phenylketonuria (PKU).
- Two other examples of newborn screening are the testing for sickle cell anemia and Tay-Sachs disease.

Newborn Screening cont

- Should all newborns be screened for all the known diseases?
- Who should pay for the tests?
- Would it be worth it to test all babies in order to save a few babies?
- Would the health care money be better spend on pre-natal care?

Pre-implantation Genetic Screening

- Many couples use in vitro fertilization techniques to become pregnant.
- This is where the woman is given hormone injections so she will produce more eggs than just one.
- These eggs are collected and sperm is added to the eggs outside the woman’s body — in a lab.
- The developing embryos are then placed into the woman’s uterus to develop.

Pre-implantation Genetic Diagnosis

- You can test the embryos for many of the disorders including Down Syndrome.
Things to consider:

- Not all the embryos produced in the clinics will be implanted, what do you do with the surplus?
- If you have multiple embryos implant, would you keep them all? Where would you draw the line?
- What do you do with the embryos you don’t use – are these "alive"?
- What factors do you use to “select” your offspring – gender?

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.

Important Concepts for Lecture Exam

- 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
- Autosomal disorders – differences between recessive and dominant autosomal disorders, examples of recessive autosomal disorders
- The benefits of sickle cell anemia

Important Concepts for Lecture Exam

- Huntington as an example of a dominant autosomal disorder, age of onset of Huntington and what does that mean passing the trait on to your offspring
- The basics of how aneuploidy can happen and that it can happen during Meiosis I or II
- Downs syndrome as an example of Aneuploidy

Important Concepts for Lecture Exam

- Cri-du-chat syndrome as an example of changes to chromosomal structure due to deletion of part of the chromosome.

Definitions

- For both lecture and lab exams:
  - Phenotype, genotype, parental (P) generation, First filial (F1) generation, X-linked, Autosomal disorders, autosomal chromosomes, heterozygous, homozygous, recessive, dominant
- For lecture exam only: Aneuploidy, Genetic screening.

Important Concepts for 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, dominant autosomal, and co-dominant (this will be on the lab practical only).
- Be able to solve genetic problems similar to the homework problems (these problems will be tested on the lab practical only).