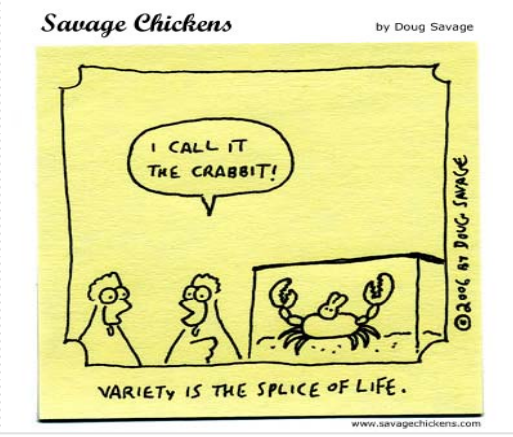
Unit 3
Chapter 16
Genetics & Heredity

Biology 3201

Intro to Genetics

- For centuries, people have known that certain physical characteristics are passed from one generation to the next.
- Using this knowledge, they learned to produce crops and livestock with desired characteristics.
- However, how these characteristics are passed from one generation to the next was unknown to them.



16.1 – Genetics of Inheritance

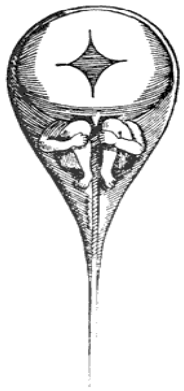
- **Traits** - Distinguishing or unique characteristics which make one organism different from other organisms.
  - Some traits are desirable while others are not.
  - Can you think of any undesirable traits? Desirable?
- It can be observed that traits can be passed down from one generation to the next (ie. Parents to offspring). This transmission of traits is called **heredity** and the traits which are passed on are said to be **inherited**.

What is Genetics?

- **Genetics** is a branch of Biology which is concerned with studying the inheritance of traits and the variations caused by them.
- By studying genetics we gain a better understanding of how we can determine the inheritance of certain traits and patterns of involved in their inheritance.
- The knowledge of genetics which we have today is a far cry from what we knew in the past.

Past Genetics

- **Hippocrates** (460 - 377 BC), a Greek philosopher, theorized that every part of the body was involved in the production of the "seeds" which the parent produced. The seeds of the male and female parent fused together to produce a new individual.
- In the 18th century, scientists believed that sperm contained pre-formed embryos. Thus it was the male who had a major contribution to the new individual which was being produced. The contribution of the female was small.
- In 1853, a monk named Gregor Mendel performed a number of experiments which involved pea plants. This study took place over an eight year period and the results of these experiments laid down a basis of inheritance from which other studies were done.



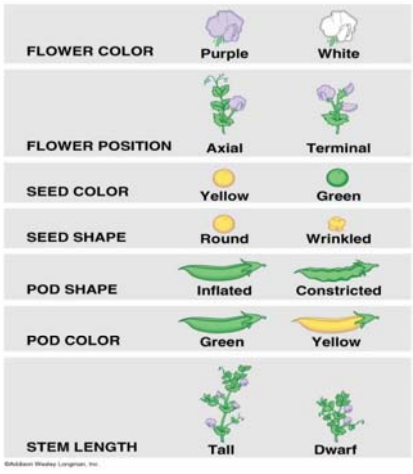
Mendel's Experiments I

Mendel chose the pea plants because:

1. Pea plants were commercially available throughout Europe at this time.
2. Pea plants are easy to grow and mature quickly.
3. The structure of the pea plants reproductive organs allowed Mendel control which plants reproduced.
4. He cross-pollinated and self-pollinated these plants.
5. Different varieties of the pea plant had different traits which could be observed easily from one generation to the next.

Mendel's Experiments II

- Mendel examined seven different traits in pea plants (shown to the right)
- Each trait had only two possible forms or **variations**.
- In order to perform his experiments, Mendel bred his pea plants until he obtained **purebred** plants. A purebred organism is similar to the parent or parents which produced it. These purebred plants were **true breeding** plants which produced plants with the desired features that Mendel was trying to obtain.
  - For example, a tall parent plant would only produce tall offspring plants.



Mendel's 1st Experiment
The Monohybrid Cross

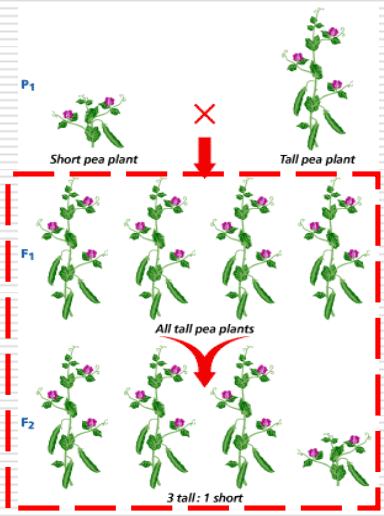
- Once he obtained purebred plants for each of the traits which he was using, he called these the **parent or P generation**.
- He crossed these parent plants to obtain a first generation of offspring which he called the **first filial generation or F1 generation**.
- The plants which were produced in the F1 generation were called **hybrids** because they were the result of a cross between two different purebred plants.
- When two plants from the F_1 generation were crossed, the offspring were called the **second filial generation or F_2 generation**
- Since only **one** trait was being considered in these crosses, they are called **monohybrid crosses**
- See Figure 16.5 on page 529 in your text

Monohybrid cross

- When Mendel performed his cross for the trait of plant height, he crossed a purebred tall plant with a purebred short plant.
  - Mendel expected the offspring to be medium height. What height would you expect the offspring plants to be?
- This was not the case, all the offspring were tall.
- From this observation he concluded that the trait for tall was **dominant** and the trait for short was **recessive**.
- Both forms of the trait were present in the F1 plants, but the short form could not be seen since it was being dominated by the tall form.
- A dominant trait is a characteristic which is always expressed or always appears in an individual.
- A recessive trait is a characteristic which is latent or inactive and usually does not appear in an individual.
- From this Mendel formed what he called the **principle of dominance**.
  - When individuals with contrasting traits are crossed, the offspring will express only the dominant trait.

Law of Segregation

- When Mendel crossed two F_1 offspring to obtain the F_2 offspring he obtained the following results every time
  - Dominant trait expressed in 75% of plants
  - Recessive trait expressed in 25% of plants
  - This 3:1 ratio is called the **Mendelian ratio**



Mendel's Conclusions

- Each parent in the F1 generation starts with two hereditary **factors**. These factors are either both dominant, both recessive, or a combination of dominant or recessive.
- Only one factor from each parent is contributed to the offspring.
- Each offspring inherits only one factor from each parent. If the dominant factor is inherited, it will be expressed. However, the recessive factor will only be expressed if the dominant trait is not present

16. 3 – Introduction

- When Mendel did his experiments with pea plants, he did not know that chromosomes existed in cells.
- In the early 1900s, chromosomes were discovered and observed in cells.

The Chromosome Theory of Inheritance

- In 1902, two scientists Walter Sutton and Theodor Boveri were studying meiosis and found that chromosomes behaved in a similar way to the factors (genes) which Mendel described.
- Sutton and Boveri made three observations
  1. Chromosomes occur in pairs and these pairs segregate during meiosis.
  2. Chromosomes align independently of each other along the equator of the cell during meiosis.
  3. Each gamete (sex cell) receives only one chromosome from each pair.

Chromosome Theory

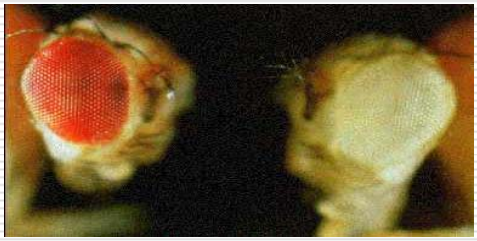
- From the above observations, they formed the **chromosome theory of inheritance**. This theory states
- Mendel's factors (genes) are carried on chromosomes
- The segregation and independent assortment of chromosomes during meiosis accounts for the pattern of inheritance in an organism.

Morgan's Discoveries

- In 1910, an American scientist called Thomas Morgan made a very important discovery from his work with fruit flies

Morgan and his Fruit Flies

- Normal fruit flies have red eyes
- Morgan crossed two red eyed parent flies and obtained a white eyed male. In other crosses, he obtained red eyed females, red eyed males and white eyed males.
- Since the white eye color was only present in the male flies, Morgan concluded that eye color was linked to an organisms sex.



Morgan & Linked Genes

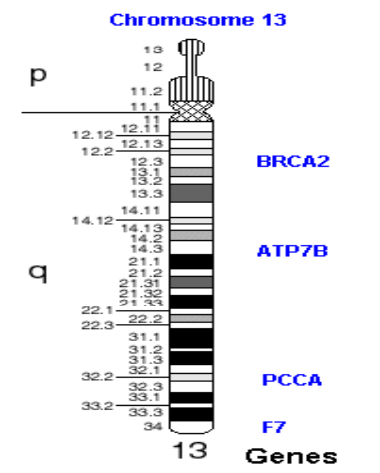
- The gene for eye color in fruit flies was located on the sex chromosome, in this case the X chromosome.
  Such genes are called **sex-linked genes**
- Morgan also stated that genes which are located on the same chromosomes are linked to each other and usually do not segregate (separate) when inherited. These are called **linked genes**

However…

- Morgan found that some genes do segregate
- Morgan created the **gene-chromosome theory** which states that genes exist at specific sites and are arranged in a linear fashion along chromosomes.

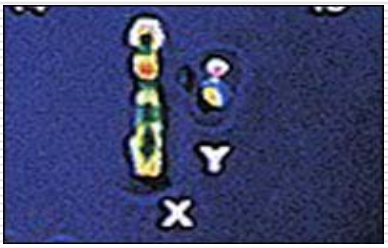
Chromosome 13 Gene Map

- Note that all genes are located in a linear fashion from one end of the chromosome to the other



Sex-Linked Inheritance

- Certain traits depend on the sex of the parent which carries the trait. The genes for these traits are located on the sex chromosomes, X or Y.



Sex-linkage

- transmission of genes which are located on the sex chromosomes is called **sex-linked inheritance**
- Genes which are located on the X chromosome are called X-linked while those on the Y chromosome are called Y-linked. Most sex linked genes are located on the X chromosome

Chromosomes & Gene Expression

Chromosome Inactivation

- Males and females produce the same amounts of proteins. This is coded by genes which are located on the X chromosome.
- Females have two X chromosomes in their cells while males have only one X chromosome.
- one of the two female X chromosomes is inactivated and this inactivated chromosome is called a **Barr body**

Polygenic Inheritance

- Most traits are controlled by one gene, however, some traits are controlled by more than one gene, this is called **polygenic inheritance**.
- Polygenic genes cause a range of variation in individuals called **continuous variation**.

Polygenic Traits in Humans

- Height
- Skin Colour
- Hair
- Eye Colour

Modifier Genes

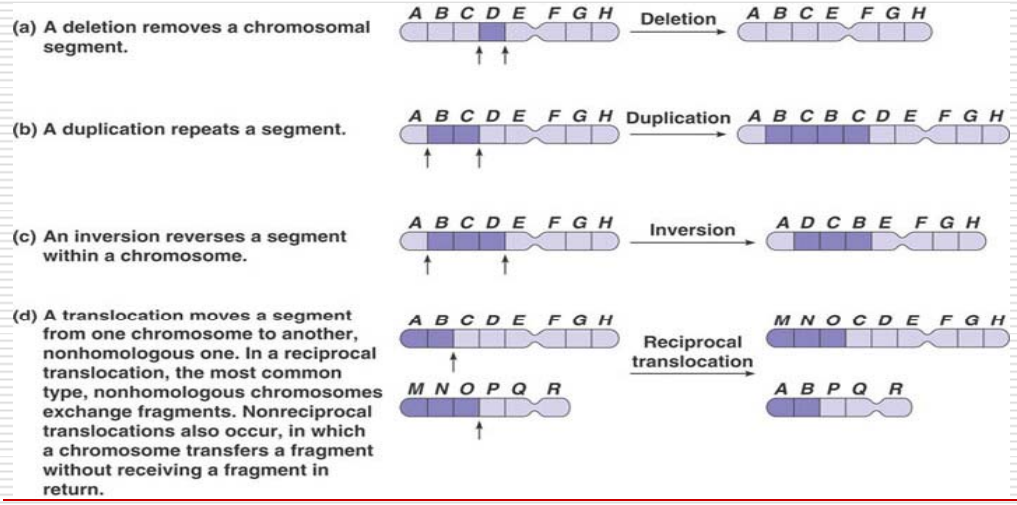
- **modifier genes** – Genes that work with other genes to control the expression of a particular trait.
- In humans, modifier genes help control the trait of eye color.
  - In this case, modifier genes influence the level of melanin present in the human eye to provide a range of eye colors from blue to brown.

Changes in Chromosomes

- **Changes In Chromosome Structure**
  - Changes in the physical structure of chromosomes can occur:

    1. Spontaneously
    2. As a result of irradiation
    3. After exposure to certain chemicals

Structural Changes in Chromosomes



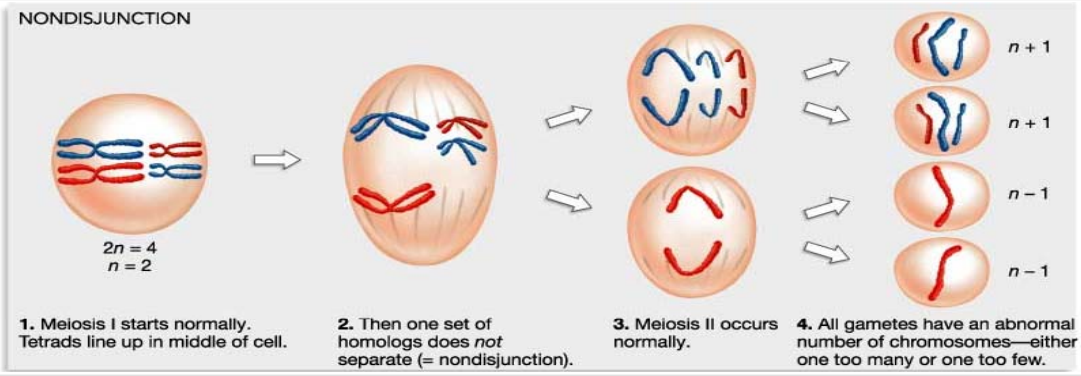
Structural Change & Disorders

- **Deletion**
  - Loss of a piece of chromosome #5
  - Cri-du-chat
  - Affects the larynx making cat sounds
- **Inversion**
  - Some forms of autism
- **Duplication**
  - Duplication in the X chromosome
  - Fragile X syndrome
- **Translocation**
  - Down Syndrome
    - # 14 and 21
  - Lukemia
    - #22 and 9

Nondisjunction

- Sometimes, chromosomes fail to separate from each other during meiosis. This produces gametes (eggs / sperm) which have either too many or too few chromosomes
- If a gamete which does not have the correct number of chromosomes is involved in fertilization, a zygote will be produced which has either too many or too few chromosomes
- This creates an embryo whose cells contain either more or less than 46 chromosomes. These embryos are usually aborted by the mother, but some survive and have genetic disorders

Nondisjunction



Pages 552 – 553 outlines genetic disorders which result from nondisjunction Monosomy, Down syndrome, Turner Syndrome

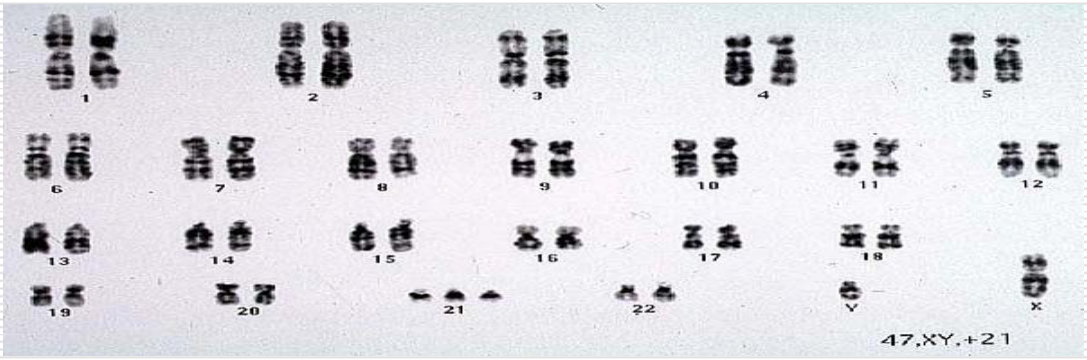
You need to know how each of these disorders arise in an individual for the test as well as the public exam.

Types of Nondisjunction

- **Trisomy** - When an individual inherits an extra chromosome.
- **Monosomy** - When an individual inherits one less chromosome.
- **Three disorders**
  - Down Syndrome
  - Turner Syndrome
  - Klinefelter Syndrome

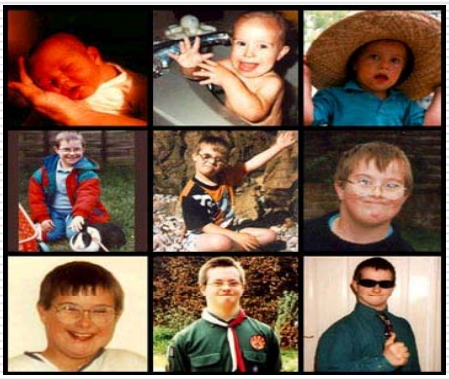
Down Syndrome (Trisomy 21)

- This occurs when an individual receives three copies of chromosome 21 instead of the normal two.



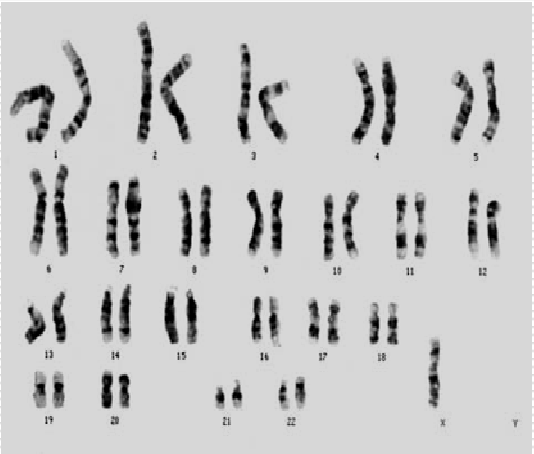
Symptoms of Down Syndrome

- Mild to moderate mental impairment
- A large, thick tongue
- Speech defects
- A poorly developed skeleton
- Short body structure
- Thick neck
- Abnormalities in one or more vital organs



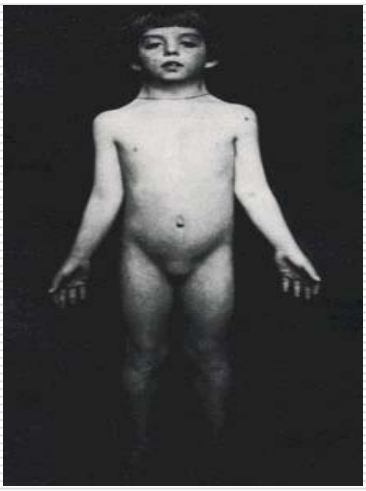
Turner Syndrome

- An individual inherits only a single X chromosome, as well the Y chromosome is missing.
- This results in a female with the genotype XO
  - O represents a missing chromosome



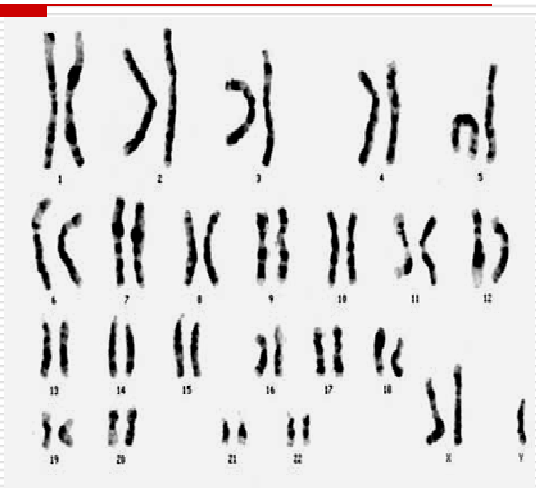
Turner Syndrome Symptoms

- Infertility
- External female genitalia, but no ovaries.
- Webbed neck
- Heart defects
- Kidney abnormalities
- Skeletal abnormalities
- Learning difficulties
- Thyroid dysfunction



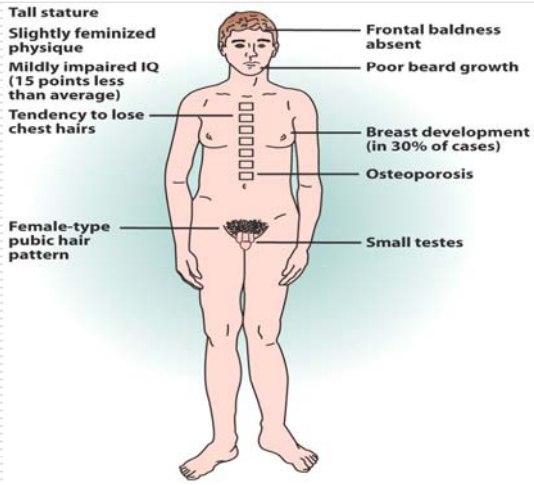
Klinefelter Syndrome

- A male who has an extra X chromosome.
- These individuals have the genotype XXY instead of XY



Klinefelter Symptoms

- Immature male sexual organs
- Lack of facial hair
- Some breast development



Jacobs Syndrome

- Males with an extra Y chromosome, having the genotype XYY
- Symptoms
  - Speech and reading problems
  - Delayed emotional maturity
  - Persistent acne
- Generally XYY males have normal potency and sexual libido, though in rare cases they may also have Klinefelter

Questions… Just a few

- Page 554 – Section Review
  - Numbers: 7, 8, 9, 10, 11

16.4 - Introduction

- The study of human genetics is a complicated field. This is due to a number of reasonsHumans have long life spans.
  1. We produce very few offspring.
  2. Most people do not keep very accurate records of their family history.

Patterns of Inheritance

- There are certain patterns of inheritance which scientists have determined for particular human genetic disorders. These include:
  - Autosomal Recessive Inheritance
  - Codominant Inheritance
  - Autosomal Dominant Inheritance
  - Incomplete Dominance
  - X-linked Recessive Inheritance

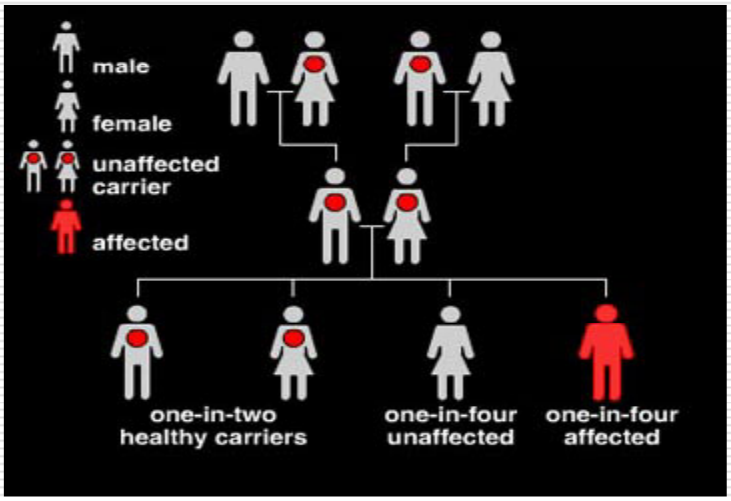
Autosomal Recessive Inheritance

- Disorder is carried on the autosomes (body chromosomes), not sex chromosomes
- Examples include:
  - Tay-Sachs disease
  - Phenylketonuria (PKU)
  - Albinism

Tay-Sachs Disease

- Individuals lack an enzyme in the lysosomes which are located in their brain cells.
  - The lysosomes are unable to break down specific lipids. Thus the lipids build up inside the lysosomes and eventually destroy the brain cells.
- Children appear normal at birth, but experience brain and spinal cord deterioration around 8 months old.
- By 1 year of age, children become blind, mentally handicapped, and have little muscular activity.
  - Most children with their disorder die before age 5.
- There is no treatment for this disorder.

Tay-Sachs



Phenylketonuria (PKU)

- A enzyme which converts a substance called phenylalanine to tyrosine is either absent or defective.
- Phenylalanine is an amino acid which is needed for regular growth and development and protein metabolism.
- Tyrosine is another amino acid which is used by the body to make the pigment melanin and certain hormones

PKU

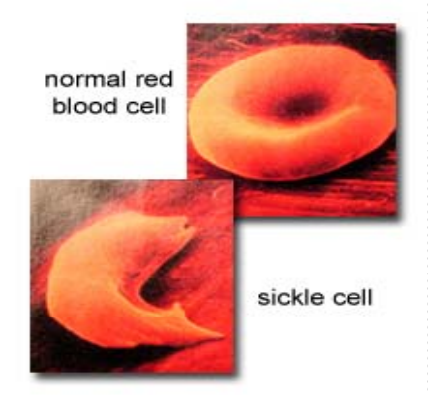
- When phenylalanine is not broken down normally, harmful products accumulate and cause damage to the individual's nervous system.
  - This results in PKU
- Babies who develop PKU appear normal at birth.
  - Can become mentally handicapped within a few months
- Today, testing and proper diet can prevent PKU from occurring in children

Albinism

- Genetic disorder in which the eyes, skin and hair have no pigment.
- People with this disorder either lack the enzyme necessary to produce the melanin pigment in their cells or lack the ability to get the enzyme to enter the pigmented cells.
- Albinos face a high risk of sunburns and eye damage from exposure to the Sun.

Co-dominant Inheritance

- Sickle-cell Anemia
  - Best example of a co-dominant disorder
- Symptoms
  - Defect in the hemoglobin and the red blood cells
  - Defect leads to clots and reduced blood flow to vital organs
  - Low energy, suffer from various illnesses and are in constant pain
  - May die prematurely

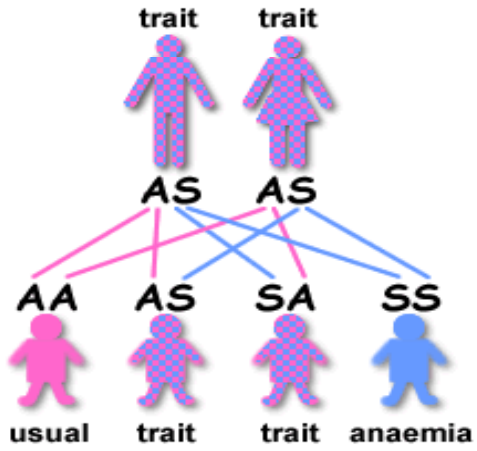


Both Parents as Carriers

- Cross:

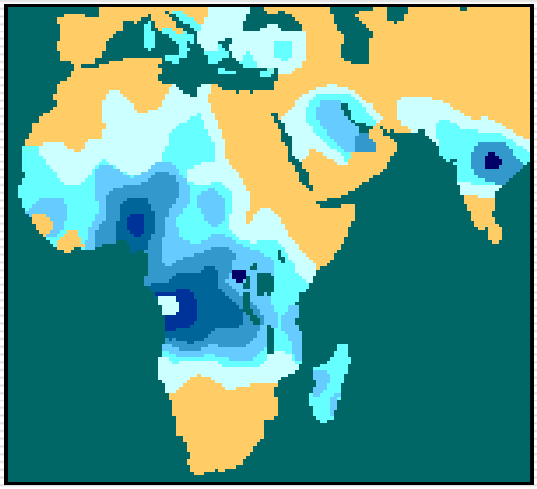
Hb^AHB^S x Hb^AHB^S

- Results:
  - 25% Normal
  - 50% Normal carriers
  - 25% Anemia



Heterozygous Advantage

- Sickle – Cell Anemia is largely predominant in Africa
- Malaria is the leading cause of death among young people
- Heterozygous individuals have been found to be less likely contract Malaria, and thus more likely to live and pass on the anemia allele
- Anemia alleles are normally lost from the population because the individuals rarely live to have children



Autosomal Dominant Inheritance

- Genetic disorders which are caused by autosomal dominant alleles, recessive condition is normal
- Very rare in humans, but they do exist.
- Caused by chance mutations or after individuals have passed their child bearing age.
- Two examples:
  - Progeria
  - Huntington's disease

Progeria (Pp)

- Rare disorder causing affected person to age rapidly
- Usually dies by age 10 - 15
- Affects 1 in 8 million newborns
- Results from a spontaneous point mutation in a gene
- Mutated gene is dominant over the normal condition (pp)

15 yr old male

16 yr old female

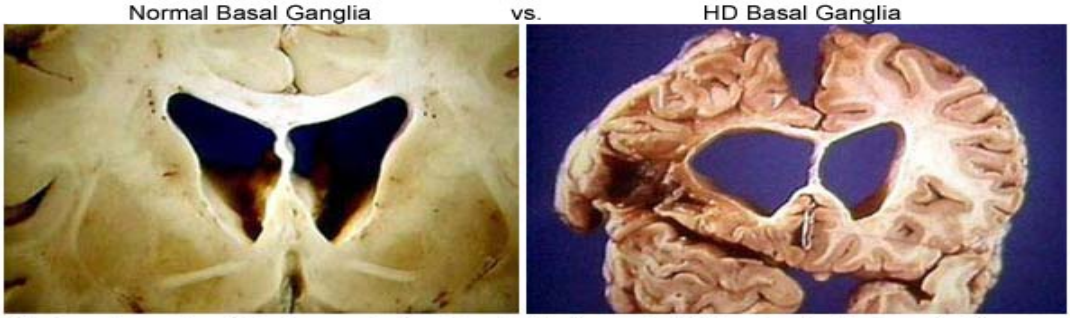
Huntington Disease

- Lethal disorder in which the brain progressively deteriorates over a period of about 15 years
- Symptoms arise after the age of 35
  - After the person has had a chance to pass the allele to their children
- Symptoms include:
  - Irritability and memory loss
  - Involuntary leg / arm movements
  - Symptoms worsen s brain deteriorates
    - Loss of speech and further loss of memory
  - Person dies by 40 – 60 yrs old before they know if their children have the mutant allele

Huntington Diseased Brain

Figure D-4: Effect of HD on the Basal Ganglia

Normal Basal Ganglia vs. HD Basal Ganglia



The basal ganglia of the human brain, showing the impact of HD on brain structure in this region. Note especially that the brain of a person with HD has bigger openings due to the death of nerve cells in that region.

Source: Singer, Jonathan. Huntington's Disease. Online. Available at: http://ist-socrates.berkeley.edu/~jmp/HD.html

Incomplete Dominance

- Disorder exhibits a phenotype which is midway between the dominant and recessive traits
- Familial Hypercholesterolemia (FH)
  - Normal cells have surface receptors which absorb low-density lipoproteins (LDLs) from the blood.
  - Individuals who have the FH disorder have cells which only have half the normal number of LDL receptors on their surface
  - Person then suffers from high cholesterol because LDLs are not efficiently absorbed from the blood
  - Normal cells have surface receptors which absorb low-density lipoproteins (LDLs) from the blood.
  - Individuals who have the FH disorder have cells which only have half the normal number of LDL receptors on their surface

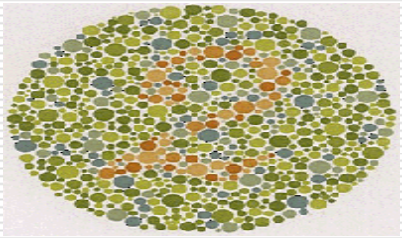
X-Linked Recessive Inheritance

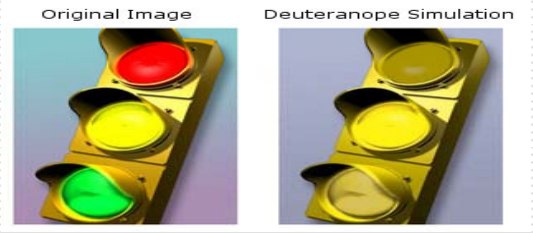
- Disorders linked to genes on the X chromosome
- Are due to the recessive form of the gene, and only occurs if there is no dominant form of the gene present
- Example: Colour blindness

Colour Blindness

- Genotypes: X^cX^c X^cY
- Heterozygous females will have normal vision but they will be carriers → X^CX^c
- Person is unable to distinguish between colours red and green
- Affects about 8% of males and 0.04% of females
- Do sample problems

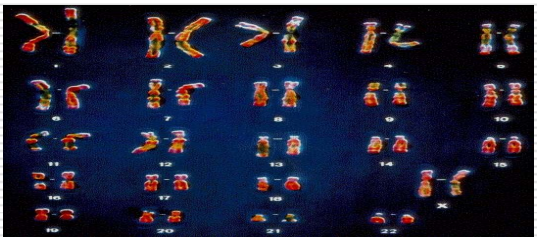
Can you see the numbers?

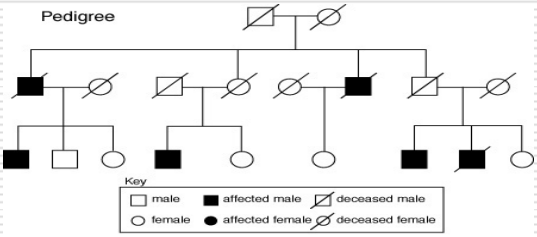




Human Genetic Analysis

- Geneticists are able to analyze the patterns of human inheritance using two methods
  - Examination of karyotypes
  - Construction of pedigrees





Human Karyotype

- Within our body cells, humans normally possess 46 chromosomes.
  - 44 of these are autosomes (body chromosomes)
  - 2 are sex chromosomes.
- A karyotype is a photograph of the chromosomes which are located in the nucleus of a somatic cell
- Once a photograph has been taken of the chromosomes in a cell's nucleus, they are cut out and arranged in pairs according to their size, shape, and appearance.
- By observing the karyotype, disorders may become apparent.

YOU WILL BE DOING A KARYOTYPE LAB FOR HOMEWORK ☺

Constructing Pedigrees

- A pedigree is a chart which shows the genetic relationships between individuals in a family.
- Using a pedigree chart and Mendelian genetics, scientists can determine whether an allele (gene) which is responsible for a given condition is dominant, recessive, autosomal, sex-linked, etc.
- A pedigree can also be used to predict whether an individual will inherit a particular genetic disorder.
- An example of such a disorder is hemophilia. This is a disorder in which a person's blood lacks certain clotting factors, thus the blood will not clot. Because of this, a small cut or bruise may kill an individual.

Chapter 16 Test

- Date: TBA
- All information and terminology from chapter 16
  - The only crosses on this test will be X-Linked problems
    - Ex. Colour blindness or hemophelia
  - Multiple Choice and short answer
- NO GENETICS PROBLEMS!