Genetic Counseling

- Until recently, it was very difficult to determine the health of an unborn baby.

- Today, with new research and technology, information can be gathered during:
  > fetal development
  > before conception (sperm and eggs)

Genetic Counseling

- Genetic counselor: A medical professional who gathers information from people who have a history of genetic disorders in their family.

- The counselor can make a family tree and predict the probability of a child inheriting a particular disorder.

- Once the parents find out, they then need to make a decision if they should conceive a child.
Genetic Counseling

- **Diagnosis can occur at two stages**
  - Pre-implantation diagnosis
    
    (Before conception - Sort through zygotes)
  
  - **Prenatal diagnosis**

- Performed after a woman has conceived a child.

- There are several methods which can be performed here:
  1. Ultrasound
  2. Amniocentesis
  3. Chorionic villus sampling
  4. Fetoscopy

Ultrasound

- Sending sound waves through the amniotic fluid which the fetus is suspended in.

- The sound waves bounce off the fetus and are used to create a black and white image of the fetus.

- The image is studied to determine any physical abnormalities such as missing limbs, a malformed heart, etc.
Amniocentesis

- A small amount of amniotic fluid around a fetus is taken.

- This fluid is grown in a petri dish for several weeks until there are enough cells to get a karyotype of the fetal cell’s chromosomes.

- The karyotype will allow scientists to see disorders such as Down Syndrome, etc.

Chorionic Villus Sampling (CVS)

- Cells are removed from the membrane called the chorion which surrounds the amniotic sac.

- These cells are grown until a karyotype can be made.

- The karyotype is then used to diagnose a genetic disorder.
Fetoscopy

- an endoscope, a long tube with a camera on one end, is inserted through a small cut that is made in the woman’s abdomen.

- Some surgeries can be performed on the fetus while still in the womb.

- Allows for the safe collection of blood samples from the fetus.

- Genetic material from the blood sample can be used to create a karyotype or to test for a number of different genetic disorders.

Genetic Markers

- Any characteristic that provides information about an organism’s genome.

- Are identified at the molecular level within DNA
  > Provides clues about the genes associated with particular disorders

- There are two types of DNA genetic markers:
  1. Linked markers
  2. Gene - specific markers
Linked Genetic Markers

- This is when there is a known sequence of nucleotides close to a gene that causes a disorder. (that sequence is 'linked' to the disorder)
- If a linked marker is found, then the gene which causes a disorder is usually nearby.

Gene - Specific Marker

- Sequence of DNA which is actually a part of the gene itself.
- This type of marker always indicates the present of a disorder causing gene.
- These DNA markers are found using a probe which consists of a nucleic acid sequence which is complementary to the marker sequence.
- The DNA marker (if present) and the probe join together, indicating the gene is indeed present
Treatment of Genetic Disorders

Four methods of treatment:

- screening and prevention
- surgery
- environmental control
- gene therapy

* Not all disorders can be treated*

Genetic Screening and Prevention

- Genetic disorders can be detected at birth.
- Blood tests can be used to detect a number of disorders early and thus allow doctors to carry out preventive measures.
- Phenylketonuria (PKU) is an example of such a disorder. If detected early, a child with PKU can be given a special diet to promote healthy growth and allow them to lead normal lives.

Surgery

- Some genetic conditions can be treated through surgery.
- Babies born with certain disorders can have them corrected through surgical procedures.

Cleft palate or a vertical groove in the roof of a child’s mouth can be corrected through reconstructive surgery.
### Treatment of Genetic Disorders

**Environmental Control**
- Sometimes, treatment of a disorder involves manipulation or control of the affected individual’s environment.

- An example of such a disorder is albinism.
  - A person with albinism lacks the pigment melanin. This pigment, in normal individuals, offers protection from the Sun’s harmful radiation.
  - Since there is no treatment for albinism, individuals with the disorder must limit their exposure to direct sunlight.

**Gene therapy**
- Medical procedure in which a normal or modified gene is transferred into the defective cells of an individual.

- The normal gene will, in theory, reverse the symptoms of the genetic disorder by allowing the recipient’s cells to function normally and synthesize any missing polypeptides (proteins).

- Viruses are usually used to transfer the normal gene to a defective cell.

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### Genetic Engineering

**Genetic Engineering**: The manipulation of an organism’s genetic material to modify the proteins it produces.

Tools and techniques used in genetic engineering:

- Restriction enzymes

- DNA amplification
  - Bacterial vectors
  - Polymerase Chain Reaction (PCR)

- Gel electrophoresis
DNA amplification

- **DNA amplification**: The process of creating a large sample of a DNA from a single gene or DNA fragment.

- **There are two different methods of doing this**
  1. **Cloning Using A Bacterial Vector**
     - A sample of DNA is treated with a restriction enzyme.
     - The DNA is then broken into fragments.
     - These fragments are then placed into bacterial plasmids.
     - The plasmid is then returned to a bacterial cell. As the cell multiplies, it synthesizes the plasmid containing the foreign DNA. This allows for millions of copies of the DNA fragments to be produced.

  2. **Polymerase Chain Reaction (PCR)**
     - PCR method allows researchers to amplify a very specific sequence within a DNA sample doing the following:
       - The DNA sample is placed in a solution with nucleotides and primers.
       - The hydrogen bonds between nitrogen base pairs are broken with heat, allowing the DNA double helix to open.
       - Next, the solution is cooled, DNA polymerase is added and replication begins.
       - Both DNA strands replicate which results in two copies of the original DNA. The cycle then repeats itself.

- Each cycle doubles the amount of DNA.

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Gel Electrophoresis

- **Gel electrophoresis**: Is used to separate molecules according to their size and electrical charge. This can be used to separate DNA fragments so that they can be analyzed.

  - A solution containing DNA fragments is applied to one end of a gel.

  - An electric current is then applied to the two ends of the gel making it polarized.

  - Since DNA has a negative charge, the fragments tend to move towards the positive end of the current.

  - The smaller fragments move more quickly than the larger fragments and this causes a separation of fragments into a pattern of bands called a DNA fingerprint.
Analyzing DNA

- The processes of using restriction enzymes, DNA amplification, and gel electrophoresis can be used by researchers to analyze and compare DNA samples.
- Determining a DNA pattern is very useful in crime scene investigation.
- It is also useful in solving disputes over parentage. (As in the thinking lab)

The Human Genome Project p.618-619

- The goal of this project is to map all the genes in humans.
- The significant findings were that 99.9% of DNA from humans is identical and that humans have about 30,000 genes.
- This process generated some controversy.
- Why controversy?
New Knowledge, New Problems

• Advances in knowledge such as the completion of the Human Genome Project raises significant legal and ethical issues.
  > Who should have access to genetic information and for what purposes?
  > Another issue is; who owns the genetic information which is gathered from individuals or groups?

• From these questions we can see that there are a number of issues which people need to be concerned with when it comes to genetic information.

Genetically Modified Organisms (GMO) and Food (GMF) p.621-632

• These are organisms or food that have changes made to their DNA.
  - This may include transferring genes from one species to another.

Some examples are:
1. Herbicide resistant plants
2. BST producing bacteria - Bacteria produce bovine somatotrophin (cow growth hormone), which is then given to cows so that they will produce more milk.
3. Golden rice - Rice that can make vitamin A so it is more nutritious.
4. Transgenic salmon - Salmon that are more resistant to cold.
5. Insulin producing bacteria.
6. PCB - Eating bacteria - Special bacteria that can help clean up pollutants.
7. Oil eating bacteria - Special bacteria that can help clean up oil spills.

Concerns about using these organisms include:
  - environmental threats;
  - health effects;
  - social and economic issues.
Cloning involves making an exact genetic copy of a living organism.

Plants and animals, including some large mammals, have been cloned.

The current technique involves:
1. Taking the nucleus from the individual that is to be cloned.
2. That nucleus is placed into an egg cell which has had its nucleus removed.
3. The modified egg is implanted into a surrogate mother and a new clone baby is born.

What are some potential benefits and concerns surrounding cloning?

Careers in Biotechnology

- Genetics is an expanding and increasingly important field of study.

There are many careers that rely heavily on genetic research, these include:

1. **Cytogeneticist** - Studying the genetics of cells.
2. **Medical geneticist** - Helping diagnose and treat genetic disorders.
3. **Genetic engineer** - Producing new GMOs or gene therapies.
4. **Genetic counselor** - Scientists who can diagnose a genetic disorder and present options for treatment to patients. The options can include genetic engineering.