Chapter 18
Genetics Ahead

Biology 3201
18.1 - Diagnosis & Treatment of Genetic Disorders

• 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 and can even be predicted before conception.
Genetic Counseling

• A genetic counselor is a medical professional who gathers detailed information from individuals about possible genetic disorders in their family. This information is gathered through interviews, blood tests, and discussions with geneticists.

• The counselor will then construct a family pedigree based on the information to predict the probability of a child inheriting a particular disorder.

• Once this information is communicated to the parents, they then need to make a decision as to whether or not they should conceive a child.
Diagnosis

- Diagnosis can occur at two stages
  1. Pre-implantation diagnosis
  2. Prenatal diagnosis
Pre-implantation Diagnosis

• Pre-implantation diagnosis is performed before pregnancy has occurred.

• Invitro-fertilization of several eggs takes place and are allowed to develop. After two days, eight cells have formed.

• One of these cells is removed and a karyotype is produced, the remaining cells continue to divide.

• The karyotype is analyzed for any genetic disorders. If none are found, the hollow ball of cells is placed in the female’s uterus to continue its development.
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

- Involves 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 the amniotic fluid around a fetus is extracted with a long thin needle.

• This fluid is placed in a special nutrient rich medium and the cells are allowed to multiply for several weeks until there are enough cells to get a karyotype of the fetal cell’s chromosomes.

• Observation of the karyotype will allow scientists to see disorders such as Down Syndrome, etc.

• Due to a potential risk to the fetus, this procedure cannot be done before the fourteenth week of pregnancy.
Chorionic Villus Sampling (CVS)

- Performed around the ninth week of pregnancy.
- Cells are removed from the membrane called the chorion which surrounds the amniotic sac.
- The chorion membrane contains fetal cells which have genetic information inside them.
- These cells are grown in a special medium 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 incision which is made in the woman’s abdomen.

- Procedures such as drainage of excess fluid surrounding the brain and blood transfusions 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.

- Identification of proper blood type and detection of blood disorders are also possible using the process of fetoscopy.
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

• A known sequence of nucleotides which is located close to a gene that causes a disorder.

• If a linked marker is found, then the gene which causes a particular disorder is usually nearby.

"The green section indicates the presence of a desirable gene in an organisms' genetic code that is associated with two genetic markers (red flags)."
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.

• When the probe is mixed with a solution which may contain the suspected gene, the DNA marker and the probe join together, indicating the gene is indeed present
Treatment of Genetic Disorders

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.
  - An individual 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, individual’s 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.
  – Though viruses usually work well, their protein coat can trigger a severe and sometimes fatal immune response in some patients. Thus, scientists are attempting to find an alternative method of inserting genes into defective cells.
So far, all gene therapy techniques that have been used have focused on **somatic gene therapy**.

- Modifying the genes which are located in a patient’s somatic (body) cells. Therapy performed on these cells will benefit the individual being treated, but not his / her offspring.

In the future, most gene therapy will focus on **germ line therapy**. This would involve altering the DNA of an individual’s germ cells or sperm or egg cells.
Limits to Diagnosis & Treatment

• Some genetic disorders are easy to diagnose or predict, using pedigree information, genetic markers, etc.

• However, there are some disorders which are more difficult to diagnose or predict.
  – Example: Alzheimer’s.
Alzheimer’s

• Alzheimer’s is a genetic disorder which is common in people over the age of 65.

• This form of dementia begins with mild forgetfulness and progresses to severe loss of memory, language abilities, and conceptual skills.

• The brains of people who die from Alzheimer’s show abnormalities which include tangles and clumps of nerve fibres.
Types of Alzheimer’s

Familial Alzheimer’s Disease
- (FAD) can strike people as early as the age of 40.

• Sporadic Alzheimer’s Disease
- (SAD) affects people over the age of 60. A gene called EpoA, located on chromosome 19, has been found to be associated with this form of Alzheimer’s.
Ethical Issues

• There is debate concerning the moral and ethical issues involved with the field of gene therapy.

• Through the use of genetic engineering techniques, DNA can be sequenced, analyzed, and altered.

• This manipulation of genetic material can be seen in either a positive or negative light depending on the individuals involved.