Genetic Technologies

Genetic Testing

Tests can be used to diagnose disorders and/or identify those individuals with an increased risk of inheriting a disorder.

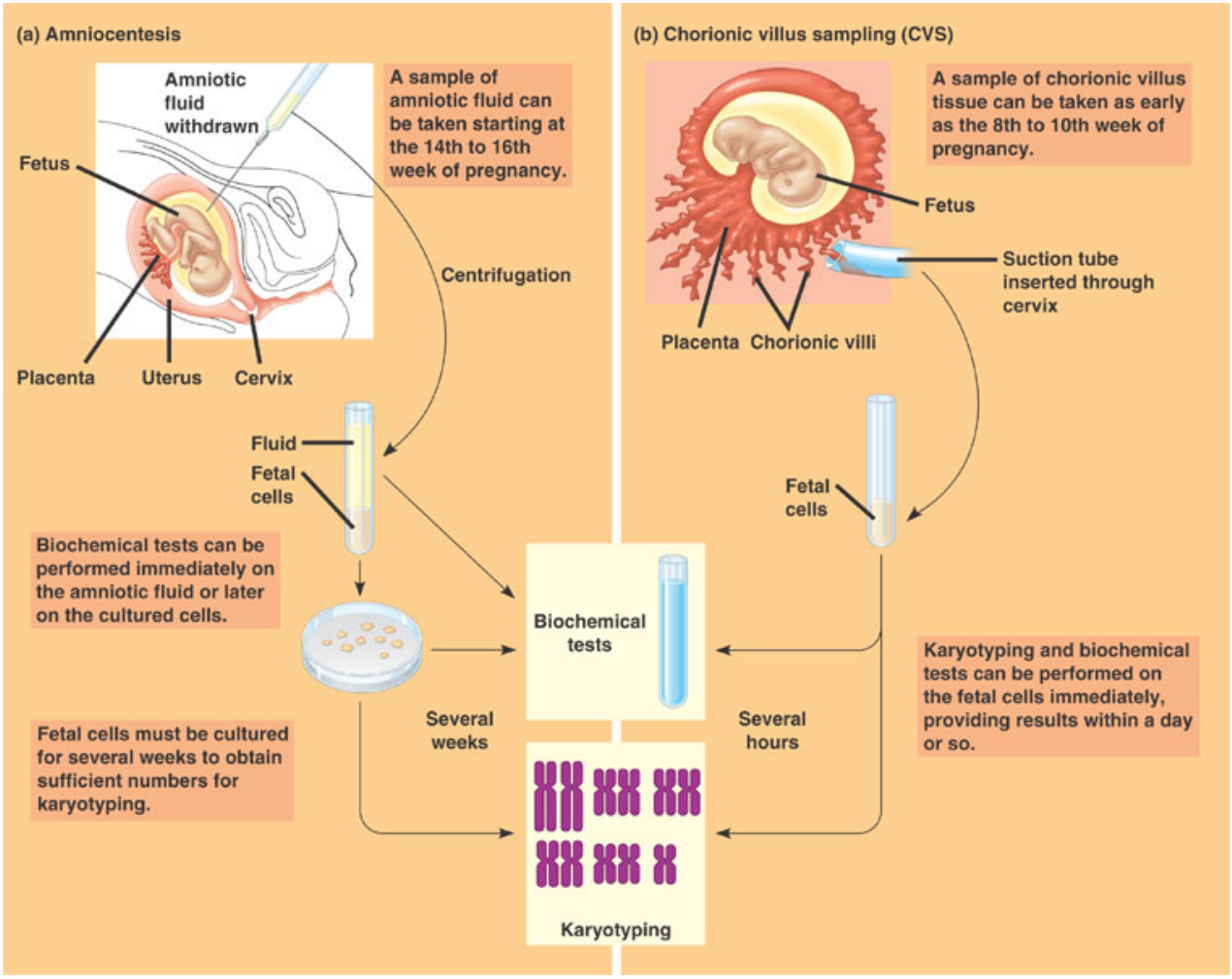
Prenatal Screening

A fetus may be screened for genetic disorders with a karyotype, which detects chromosomal abnormalities. Disorders like Spina Bifida and Down's syndrome are frequently screened for.

Tissue samples may be collected using amniocentisis or chrionic villi sampling (CVS).

Amniocentisis

CVS



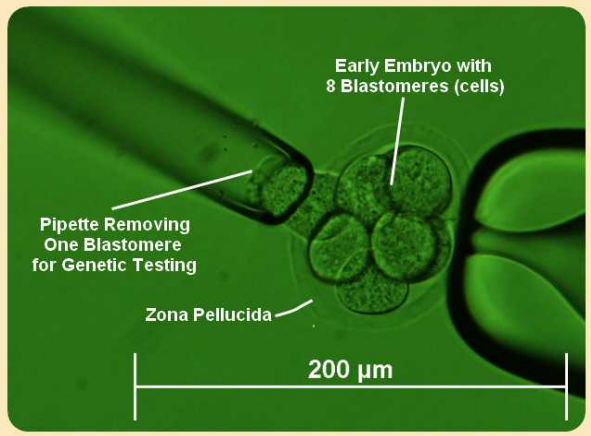
Pros? Cons?

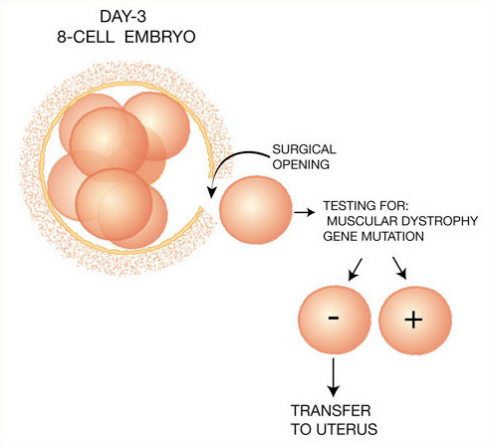
Maternal Serum Screening

Between 16-20 weeks the mother's blood is tested for abnormal levels of certain hormones or proteins which might suggest Trisomy 21, Trisomy 18 or other neural defects such as Spina Bifida.

Pre-Implantation Genetic Diagnosis (PGD)

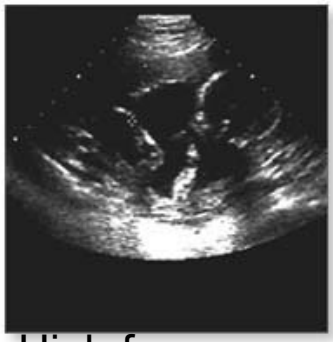
Prior to implantation, embryos can be screened for genetic markers that indicate the presence of a genetic abnormality. Embryos can be pre-selected prior to implantation in the uterus.

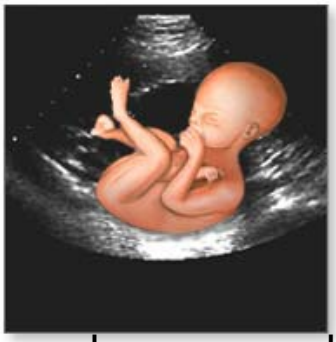


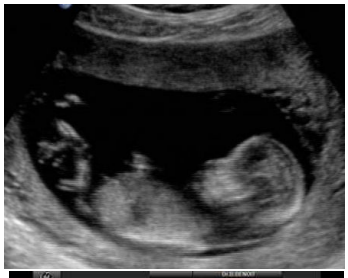


Ultrasound

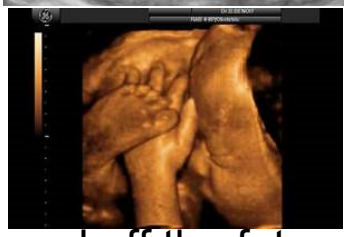
Ultrasound of fetus during week 17 of pregnancy







2D



3D

- High frequency sound waves are bounced off the fetus to create 2D or 3D images
- Useful for detecting physical abnormalities
- Often used in conjunction with other forms of testing

http://www.youtube.com/watch?v=Sh5uSL1UpMg

http://www.youtube.com/watch?v=pVCOMrsP9-E&feature=related

Newborn Screening

Shortly after birth, newborns are screened with a simple heel prick blood test. This test can detect disorders such as PKU, and a special diet from birth can prevent the onset of symptoms of the disorder which include severe developmental delay.

Genetic Screening

Advantages?

- early detection is beneficial for treatment
- to discover predispositions (Cancer, Diabetes, Alzheimers, Heart Disease) that can be minimized later in life by changes in lifestyle
- pre-conception or prenatal screening may inform family planning
- confirmation of diagnosis

Disadvantages?

- emotional and stressful
- potential for labelling population segregation (insurance, employments opportunities, relationships - marital prenumps!)
- conclusiveness of tests - False Positives
- repercussions of decisions based on tests alone

Genetic Counselling

Genetic counsellors have special training in both counselling and educating. They are able to advise and assist families and individuals with both the medical aspects (i.e. interpreting test results) and the emotional ramifications of genetic testing.

Components of the Genetic Counseling Process

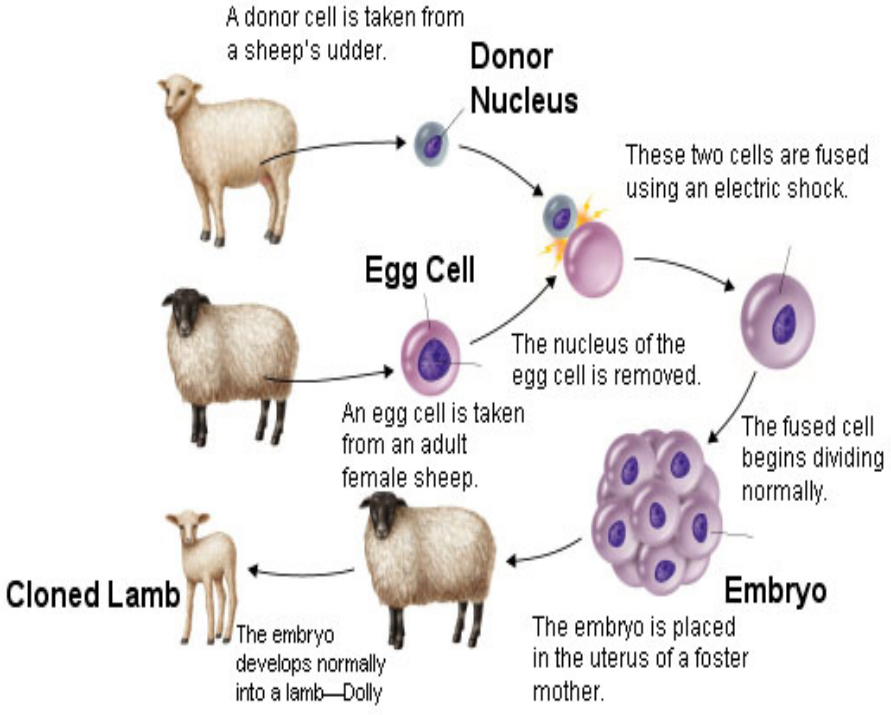
1. Information gathering
2. Diagnosis
3. Risk assessment
4. Information giving
5. Psychological assesment and counseling
6. Help with decision making
7. On-going client support

Canadian Association of Genetic Counsellors
Association Canadienne des Conseillers en Génétique

This service may be offered because of screening results, or screening tests may be performed based on family history. Usually high risk persons such as pregnant women over 35, parents with children or family members with known genetic disorders, certain ethnic or racial groups with a high incidence of a specific disease would seek genetic counselling.
Family history is collected and used to create a pedigree, lab tests and physical exams may be performed to collect data
The disease/abnormailty/syndrome is discussed, parents are educated, risks are weighed and future options and supports are explored.

Cloning

- nuclear cloning has helped to produce whole organisms from a somatic cell (e.g. Dolly the sheep in 1996)
- a somatic nucleus replaces the nucleus of an egg cell from the same species. The egg goes on to divide and become another organism.
- this technique has the potential to reproduce quickly desirable traits, but there have been issue with embryo survival rates and health issues with adults



Human Genome Project

Began in 1990 and published in 2001, the Human Genome Project used **DNA sequencing** (determining the order of nucleotides) revealed new information about our genome:

- only 1.5-2% actually contains genes
- some of our DNA contains sequences inserted from viruses
- rate of mutation in males: females is 2:1
- more than 200 genes are similar to those found in bacteria

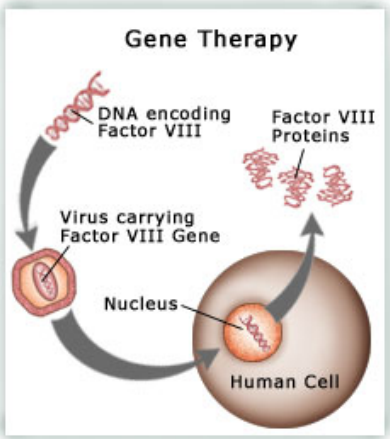
This project lead to the development of new fields of study:

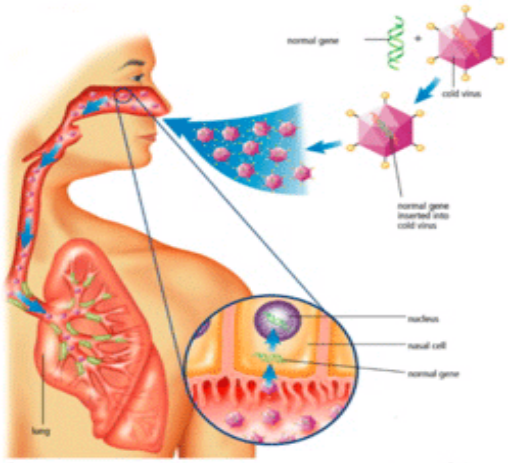
Bioinformatics: the science of handling and analyzing biological data in databases e.g. DNA sequences

Genomics: Although entire genome of other organisms were studied prior to the HGP, the study of the human genome, it genes and their locations on chromosomes had lead to discoveries such as the gene containing the mutation that causes Parkinson's disease. It has also brought about new forms of genetic testing including **DNA Chip Tests**.

Gene Therapy

- Involves inserting a proper working copy of a gene into the cells that lack the ability to produce their own protein.
- Getting the gene into and incorporated into the cells, and having it stay there and be successfully expressed are large challenges. Some possible methods of delivery include tissue transplant, microscopic injections or aerosol inhalers.
- Example - if pancreatic cells are unable to produce insulin, insert functioning gene into the cells so they can. Sometimes the DNA is rejected or it affects multiple organs instead of just the target organs.
- Example - potential treatment for CF, Huntington's disease, skin cancer

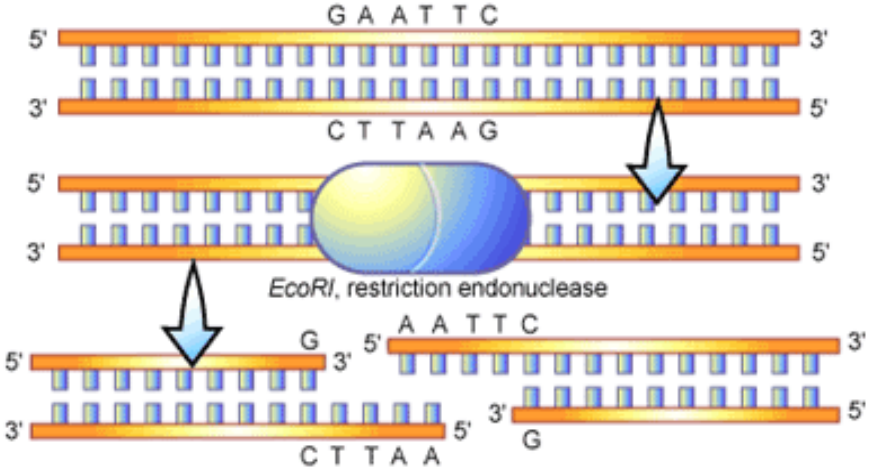




Gene therapy could deliver a working gene into a CF patient's cells.
(courtesy of BC Science 9, © McGraw-Hill Ryerson Limited, 2008.)

Manipulating DNA

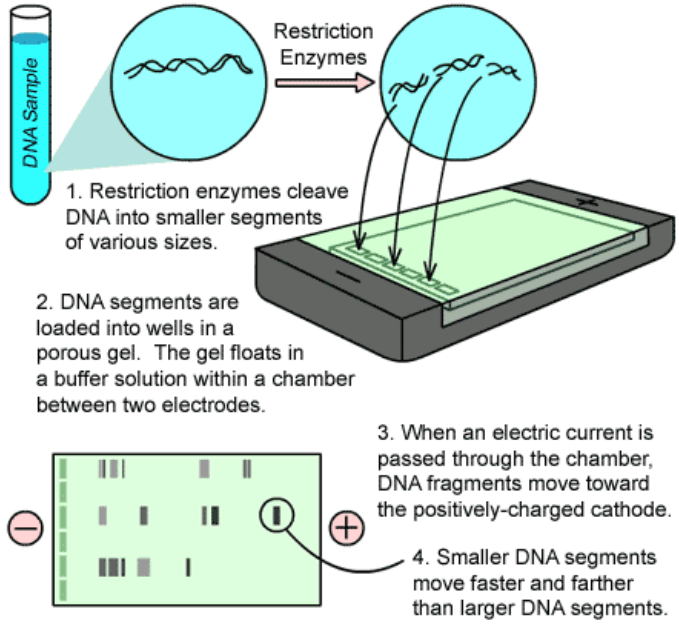
Long and unruly strands of DNA are cut with "molecular scissors" known as **restriction enzymes.** These enzymes are naturally occurring bacterial proteins that defend against viral invaders.



The enzymes cut the DNA at a specific recognition sequence. the pieces that have been snipped up are called **restriction fragments.**

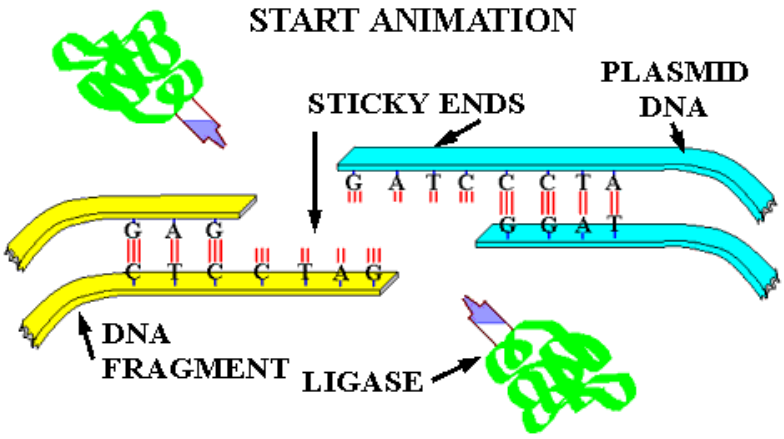
The fragments will vary in length and can be sorted and analyzed using a technique called **Gel Electrophoresis**.

Figure S-2: Gel Electrophoresis



The resulting pattern is often called a **DNA fingerprint**. It can be used to match DNA from an unknown source to a known source, because the banding patterns produced are unique. (e.g. paternity suits, illness outbreaks, crime scenes)

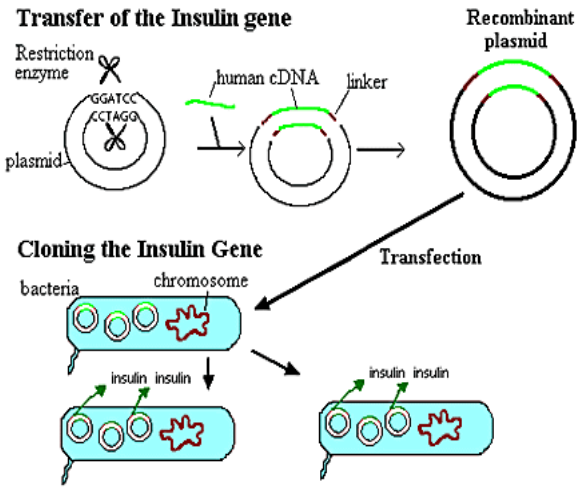
One of the most useful properties about restriction fragments is the creation of **"sticky ends"**. These single stranded sections are able to bind to any complementary sequence - including DNA from other organisms. The DNA fragments are joined by an enzyme called DNA ligase.



The newly formed strands which are a combination of DNA fragments are the basis for **Recombinant DNA Technology.**

Cloning Recombinant DNA

Bacterial plasmids, loops of DNA, can be cut and pasted with recombinant DNA technology techniques. Virtually any gene can be spliced into a plasmid. When the bacteria reproduces, the plasmid and the inserted gene are copied. Colonies of bacteria can be used to express the gene product which may vary from indigo blue dye for jeans to insulin for diabetics.



Transfer and cloning of the Insulin gene

http://www.youtube.com/watch?v=8rXizmLjegI

The importance of recombinant DNA will only increase as research finds ways to use it in:

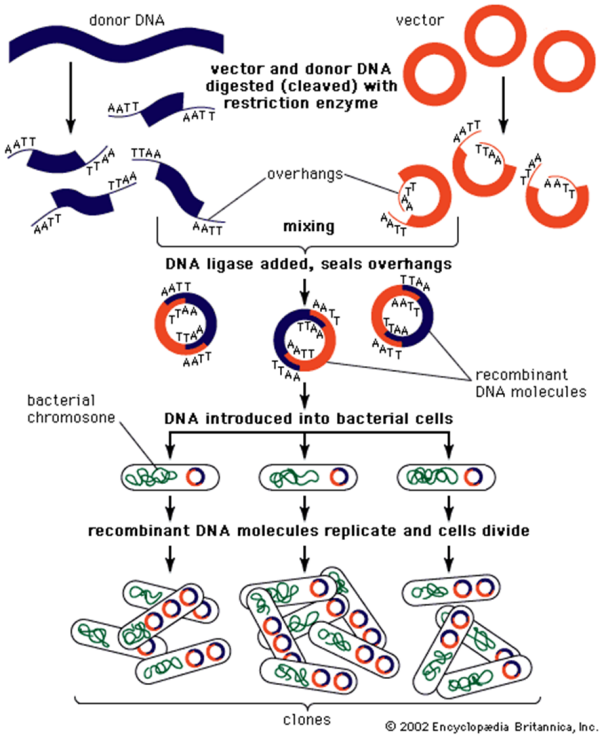
- Engineering new crops
- Developing vaccines
- Preventing and curing diseases
- Gene therapy

DNA Amplification

Generating large samples of a specific DNA sequence from a single gene or DNA fragment.

1. Cloning by means of a Bacterial Vector

- Restriction endonucleases produce a molecule of recombinant DNA
- Recombinant plasmid is inserted into bacterial cell
- Cell multiples and replicates plasmids containing foreign DNA



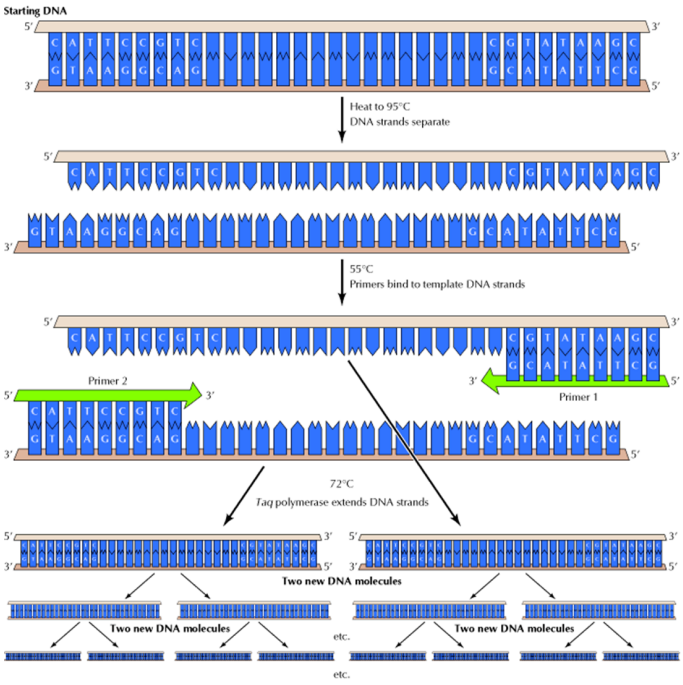
2. Polymerase Chain Reaction (PCR)

This is an automated method in which a DNA fragment sample is placed in a solution along with nucleotides and primers. The DNA is copied in a three step cycle.

I - Solution is heated to break H-bonds between base pairs (DNA helix opens)

II - Solution is cooled, DNA polymerase is added to solution, replication begins

III - Cycle repeats, billions of copies of a DNA sequence are created in very little time.



http://learn.genetics.utah.edu/content/labs/pcr/ Try the PCR Virtual minilab!