



Medical Analysis Department

Faculty of Science



Lab.1

Genetic Testing Methodology

Human Genetics

Lab.1

2nd/ Grade – Spring Semester 2021-2022

Dr. Shataha S. Jumaah/Lecturer

✉ shatha.saadi@tiu.edu.iq

13/0/2/2022

Genetic Testing Methodologies

Over the last decade, as the number of genetic tests has increased, so has the variety of genetic testing procedures.

Vary based on the type of abnormality being evaluated, the type of test used will vary.

There are three types of genetic testing accessible in general.

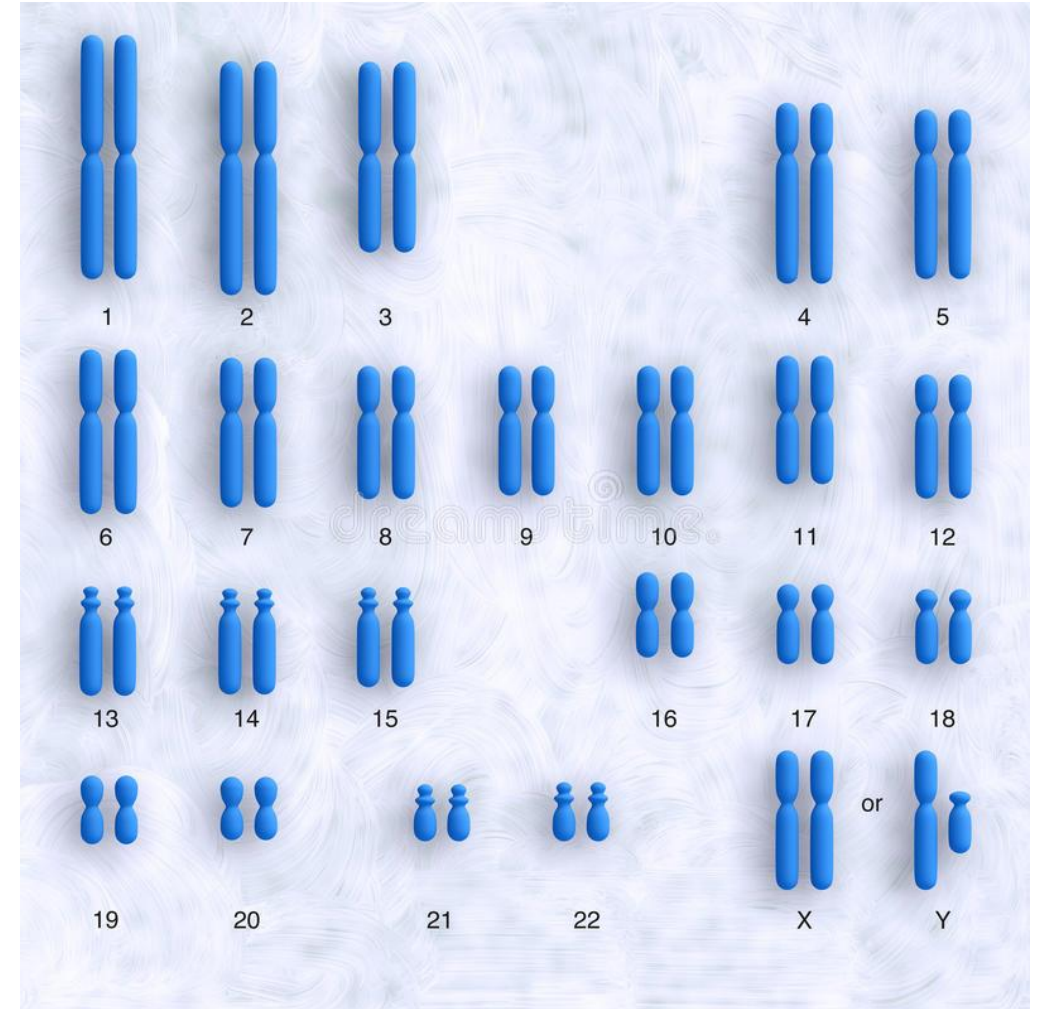
1. Cytogenetic Testing.

2. Biochemical Testing.

3. Molecular Testing.

1. Cytogenetic Testing.

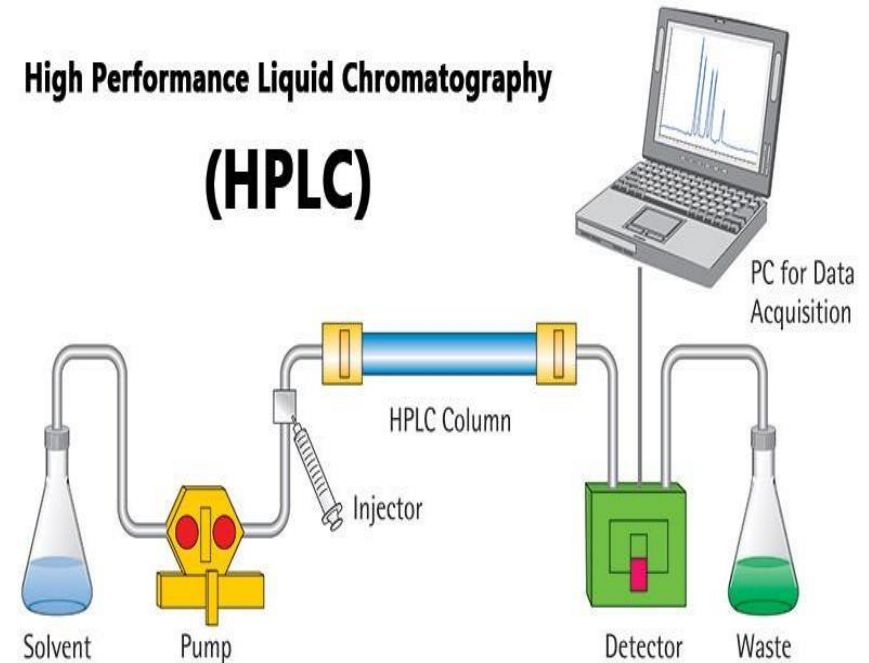
Involves testing samples of tissue, blood, or bone marrow in a laboratory to look for changes in chromosomes, including broken, missing, rearranged, or extra chromosomes. Changes in certain chromosomes may be a sign of a genetic disease or condition or some types of cancer



2. Biochemical Testing.

Techniques that examine the protein instead of the gene.

These tests require a tissue sample in which the protein is present, typically blood, urine, amniotic fluid, or cerebrospinal fluid. Like high performance liquid chromatography (HPLC)



3. Molecular Testing.

Checks for particular genes, proteins, or other substances

Direct DNA analysis is applicable when the gene sequence of interest is known. A DNA test can be performed on any tissue sample and requires very small amounts of sample. Like PCR



A genetic material to identify genetic changes &

including: -

- ▶ **Chromosomes**
- ▶ **Deoxyribonucleic acid (DNA)**
- ▶ **Ribonucleic acid (RNA) to detect genetic material and/or identify genetic changes.**

Types of Samples

Blood

EDTA tube
containing 3-5ml
whole sterile
blood

Cultured cells

Buccal

Samples preferred to
perform DNA testing
for patients having
had a previous
heterologous
B.M. transplant.

Saliva

samples can
be used for
all tests

Other Tissue

Types

FFPE, Fresh,
Frozen Tissue
Samples, BM

Ethical issues of Genetic Testing

- 1. Informed consent:-** is a process for getting permission before conducting a healthcare intervention on a person, for conducting some form of research on a person, or for disclosing a person's information.
- 2. Protection of privacy :-** Genetic privacy laws take three alternative approaches to protecting patient privacy:
 - A.** Requiring informed consent on the part of the individual
 - B.** Restricting unfair usage of genetic data by employers, health care providers or insurance companies
 - C.** Limiting retelling without the consent of the individual or defining genetic data as the 'property' of the individual.

- 3. Genetic counseling :- checking family medical history and medical records.**
- 4. The issue of minors :- emancipated minors القاصرين have practically no legal rights to obtain genetic or genomic testing without parental permission.**
- 5. Prenatal diagnosis :- These tests can tell you whether your fetus actually has certain disorders. These tests are done on cells from the fetus or placenta obtained through amniocentesis or chorionic villis sampling (CVS).**

| THANK YOU