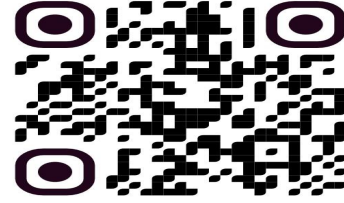


MTJ1



اسم المادة و الكورس

LAB1. INSTRUMENTS & MATERIALS USED IN THE MEDICAL GENETICS FIELD



Sawa University

جامعة ساوة الاهلية

College of health and medical techniques

كلية التقنيات الصحية والطبية

Department of Medical Laboratories

قسم تقنيات المختبرات الطبية

Third Stage

المرحلة الثالثة

محاضرة رقم 1

Lecture No. 1

الجانب العملي: الوراثة الطبية

Medical genetics-Practical

تدريسي المادة : م.د.عباس علي منشد

م.م. مهند نجر اليونسى

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Learning Objectives

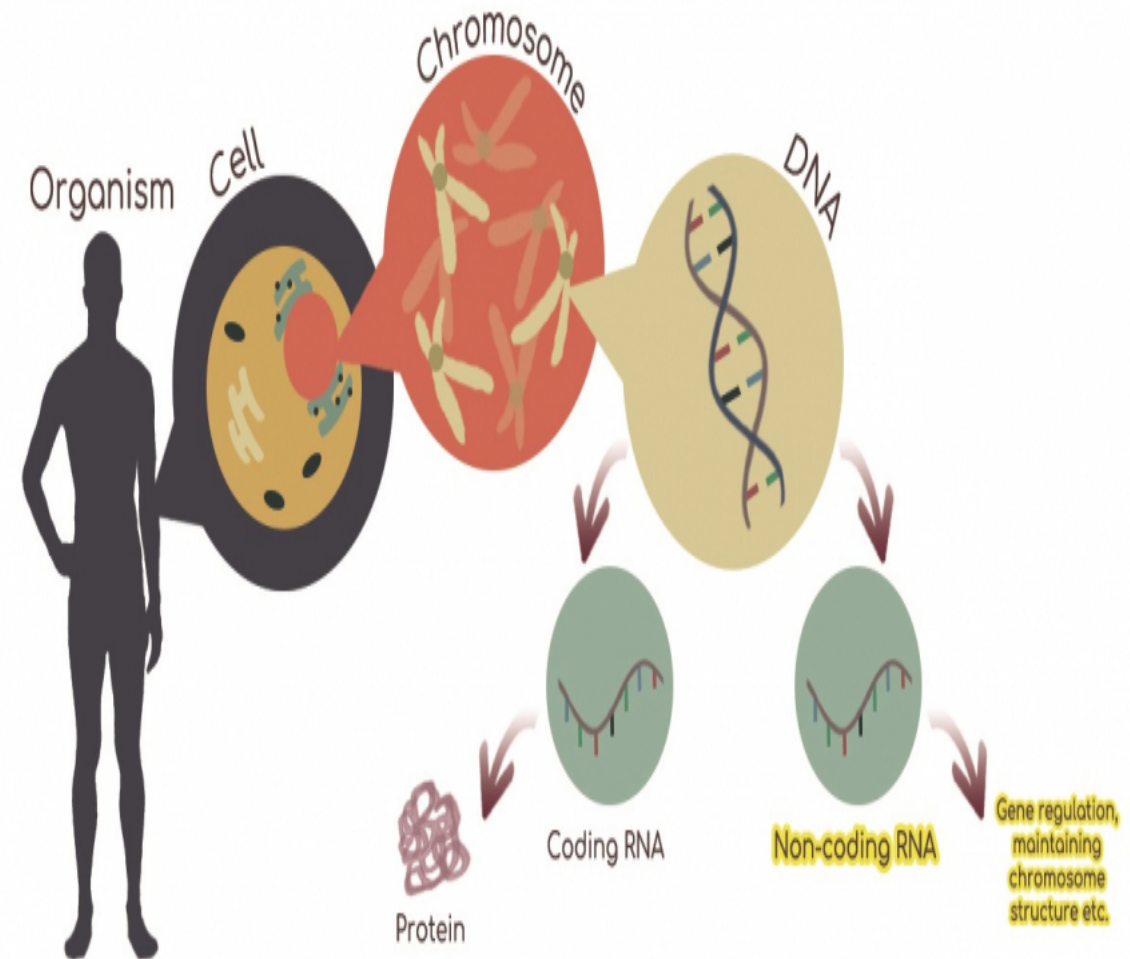
By the end of this practical session, students should be able to:

- Identify the essential instruments used in a medical genetics laboratory.
- Describe the principle of operation of each instrument.
- Explain the applications of these instruments in genetic analysis and diagnostics.
- Recognize the most common consumables used in DNA, RNA, and chromosome analysis.



Introduction

- **Genetics** is the study of genes, genetic variation, and heredity in organisms. Gregor Mendel was the first to study genetics scientifically.
- **Medical genetics** is any application of genetic principles to medical practice. This includes studies of inheritance, mapping disease genes, diagnosis and treatment, and genetic counseling.
- A medical genetics laboratory is a specialized setting designed for the study of genetic material, including DNA, RNA, and chromosomes.
- It combines traditional cytogenetic methods with modern molecular biology techniques. Genetic labs are essential for identifying mutations, diagnosing hereditary diseases, and studying cancer genomics.
- Such labs require precision instruments to ensure accuracy and reproducibility of results.



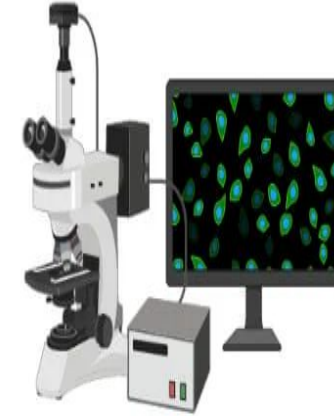
Here are the most common instruments that are used.

Light Microscope & Inverted Microscope

- **The light microscope** uses visible light and glass lenses to magnify specimens up to ~1000x.
- It is the cornerstone of cytogenetics, particularly in karyotyping to identify chromosomal abnormalities.
- **The inverted microscope** has its light source above and lenses below, enabling observation of live cultured cells.
- In genetics, it is used for maintaining fibroblast or amniocyte cultures prior to chromosome analysis.
- Both microscopes are indispensable for detecting structural and numerical chromosome abnormalities.
- Examples include identifying trisomy 21 (Down syndrome) or translocations in leukemia.



Light Microscope



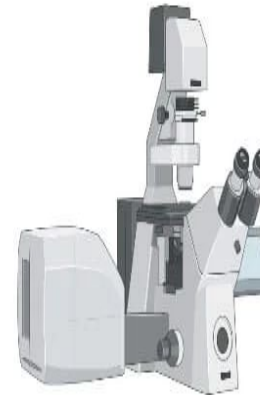
Fluorescence Microscope



Electron Microscope



Stereo Microscope



Confocal Microscope



Atomic Force Microscope



Inverted Microscope

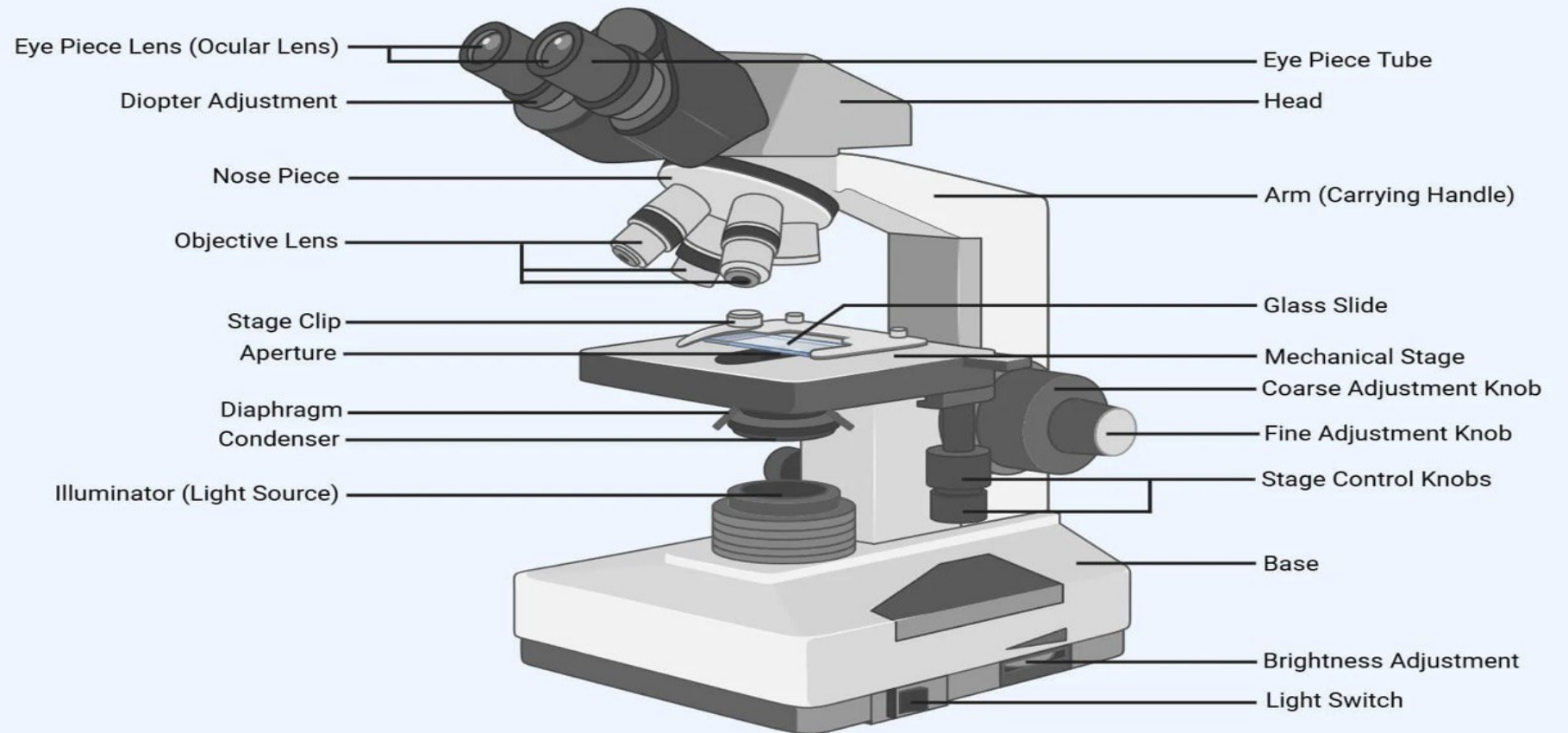


Retinal Imaging Microscope

HERE ARE THE MOST COMMON INSTRUMENTS
THAT ARE USED.

LIGHT MICROSCOPE

Parts of a Microscope



Centrifuge

- A centrifuge spins samples at high speed to separate components according to density.
- In medical genetics, it is mainly used to isolate the buffy coat from whole blood, which is rich in white blood cells.
- White blood cells are the primary source of genomic DNA for downstream analysis.
- Different centrifuges are used: **benchtop centrifuges** for routine separations, and **ultracentrifuges** for viral or plasmid work.
- Speed is expressed in revolutions per minute (RPM) or relative centrifugal force (RCF, g-force).
- Proper balancing of tubes is essential to avoid accidents and ensure reliable separation.



Benchtop centrifuge



High-speed centrifuge



Microcentrifuge



Refrigerated centrifuges



Hematocrit centrifuge



Low-speed centrifuge



Ultracentrifuges



Vacuum centrifuge

Fixed vs. Adjustable Volume Pipettes

- A pipette is a lab device used to measure out or dispense small amounts of liquid in volumes of milliliters (mL) or microliters (μL).
- **Fixed-volume pipettes** deliver only one set volume (e.g., 100 μL).
- They are best suited for dense or viscous liquids, where consistent volume is critical.
- Advantage: Faster operation, no need to adjust the setting.
- Calibration is simpler and quicker compared to adjustable pipettes.
- **Adjustable-volume pipettes** allow changing the volume using a dial/knob.
- Advantage: Versatility – one pipette can handle multiple volumes.
- Limitation: More prone to incorrect settings, requires careful calibration



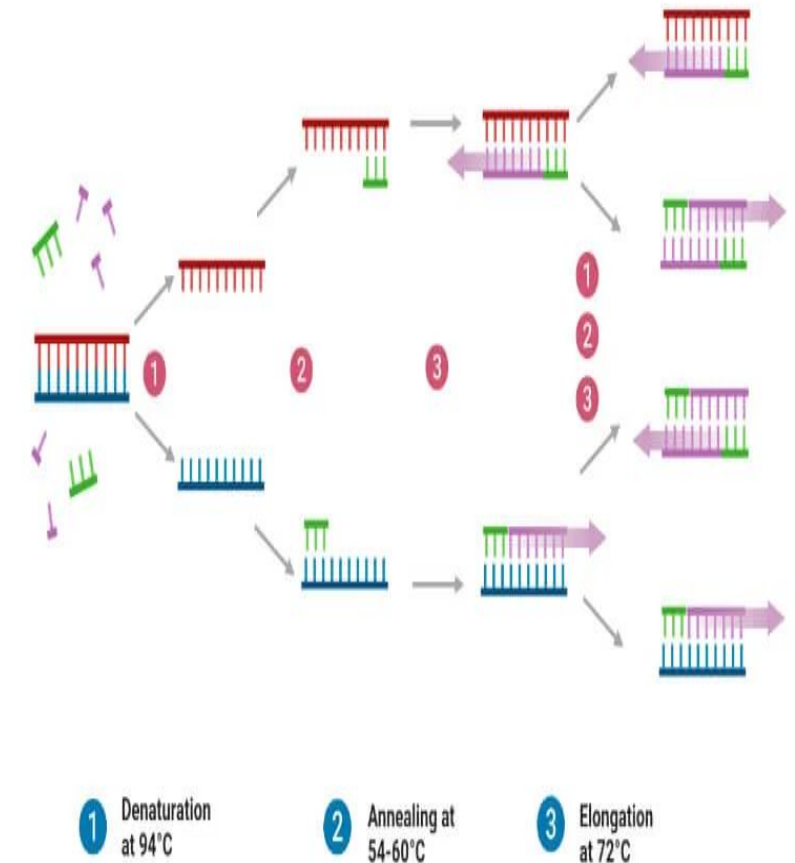
Micropipetting

<https://www.youtube.com/watch?v=NgosWmRjjAo>

PCR Thermal Cycler

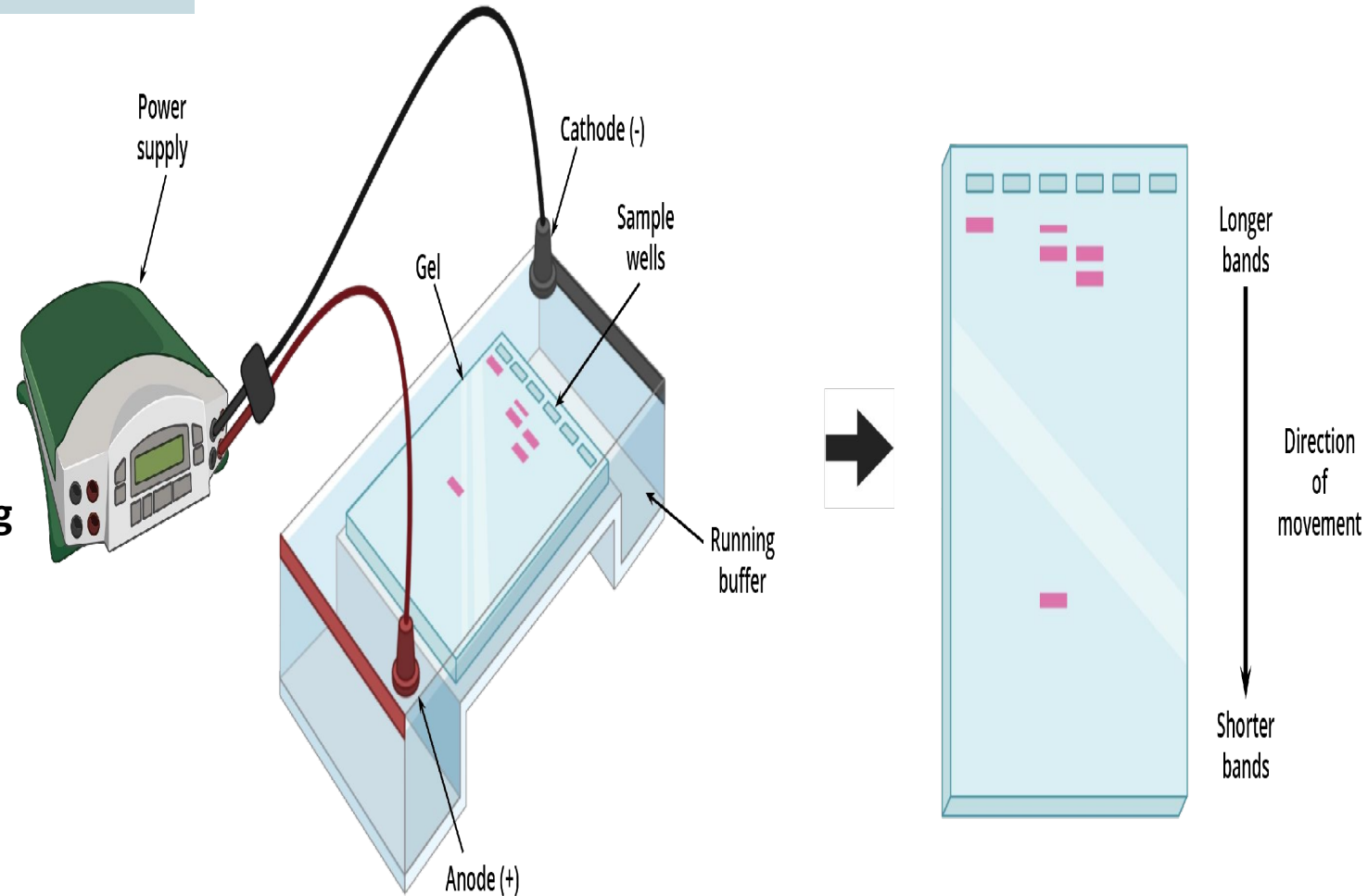
- The **Polymerase Chain Reaction (PCR)** is a revolutionary method for amplifying specific DNA fragments.
- The thermal cycler allows automatic cycling through three key steps:
- **Denaturation (95°C):** DNA strands separate.
- **Annealing (50–65°C):** Primers bind to target sequences.
- **Extension (72°C):** DNA polymerase synthesizes new strands.
- This cycle repeats ~30–40 times, producing millions of DNA copies.
- Applications: mutation detection, infectious disease diagnosis (e.g., COVID-19, HIV), and forensic profiling
- .PCR has become the “gold standard” in molecular diagnostics.

PCR Machine- Principle, Parts, Steps, Types, Uses, Examples



Gel Electrophoresis System

- **DNA, RNA, or proteins** are negatively charged molecules that can be separated by size in a gel matrix.
- **Agarose gel electrophoresis** is used for DNA and RNA; polyacrylamide gels are used for high-resolution or protein studies.
- Molecules migrate toward the positive electrode, with smaller fragments moving faster.
- **Ethidium bromide** or safer dyes (**SYBR Green**) are used for visualization under UV light.
- In genetics, it is used to confirm PCR products, detect restriction fragment length polymorphisms (RFLPs), and **analyze mutations**.



Supporting Instruments

- **Micropipettes & Tips:**

Allow precise liquid handling in microliters, essential for PCR and DNA quantification.

- **Water Bath & Incubator:**

Maintain constant temperatures for enzymatic reactions and cell cultures.

- **Spectrophotometer (Nanodrop):**

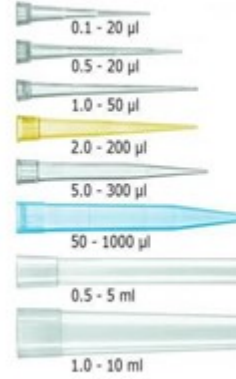
Quantifies DNA, RNA, and protein concentration and assesses purity (A260/A280 ratio).

- **Refrigerators & Freezers (-20°C, -80°C):**

Store DNA samples, enzymes, and reagents.



Supporting Instrum



QUIZ

Why is the buffy coat the preferred source of genomic DNA in humans?

Any questions?



Thank You