

Chromosomal Aberrations

* Please revise cell cycle and mitosis before you go on

Objectives:

Upon completion of this lab, the students should be able to:

1. Identify the various aberrations from slides and/or sheets.
2. Evaluate chromosome aberrations and disturbances.
3. Summarize the cause of each aberration.
4. Assess the genotoxic agents, which contribute to its increasing application in environmental monitoring.

Introduction:

Many of toxic chemicals (as pharmaceuticals products, domestic and industrial wastes, pesticides and petroleum products) are present in our environment through both natural and human ways. Once they enter the environment, it's really difficult to eliminate their effect on our biological processes.

Genotoxicity aims to determine the magnitude of genetic risk caused by an environmental agent or chemical under a specified level of exposure.

Higher plants constitute an important material for genetic tests to monitor environmental pollutants due to the possibility of assessing several gene mutations and chromosomal aberrations in cells.

Mitotic index (MI), duration of phases and Chromosomal aberrations (CA) are cellular biomarkers revealing the extent of toxicity of the applied substances.

Chromosomal Aberrations (CAs)

On the chromosomal level, CAs vary by the change in organization, number or structure of chromosomes which occur as a result of the exposure to certain treatments (biological or chemical). Some of these aberrations could be detected in different mitotic phases in comparison with the normal ones.

Disturbances can be recognized through the following categories (see the figures)

- A) Spindle Disturbances
- B) Nucleic Acid Disturbances
- C) Chromosomal Disturbances
- D) Nuclear Disturbances
- E) Cytoplasmic Disturbances
- F) Cellular Disturbances

A) Spindle Disturbances

1- Multipolar anaphase:

- Whereas, chromosomes move in the form of *numerous groups towards poles*.
- The multipolar spindle is formed **due to** the *splitting of its fiber apparatus into many directions*.

2- C-Mitosis (C-Metaphase):

- It is the disorientation and scattering of the chromosomes.
- This is **due to** the *prevention of the normal assembly and function of the spindle fibers*.
- **N.B.**
 - i. This process is followed by the delay in the division of centromere. This results in what is known as *metaphase arrest*, which is detected by an increased metaphase ratio than that of normally dividing cells.
 - ii. C in c-mitosis refers to colchicine; a poisonous alkaloid (extracted from seeds of Colchicum plant) that is well noted to induce such aberrations in addition to polyploidy.

3- Star-shaped figures:

- They appear as star-like (rosette-like) morphology of the segregating chromosomes.
- ***Centromeres are*** drawn to the center of the equatorial plane (at metaphase) or to the poles (at anaphase and telophase).
- This is **due to** the *abnormal organization of spindle fibers*.
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4- Diagonal figures:

- They appear when the poles and equator are not in their normal places.
- This is **due to** the *orientation fault of equatorial plane*.

B) Nucleic Acid Disturbances**Stickiness of chromosomes:**

- The chromosomes appear connected with each other.
- It results from the improper folding of chromosome fibers **due to** the *irreversible depolymerization of DNA*.
- Sticky figures may appear in any of phases.

C) Chromosomal Disturbances**1- Chromosome bridges:**

- They can be seen as the chromosome groups at the 2 poles are connected by long chromatid(s). It is commonly observed during anaphase and telophase.
- Bridges are probably formed by the *incomplete segregation of daughter chromosomes* or the fusion between their ends.
- They can be also attributed to the *fusion of the sticky ends of anaphase or telophase chromosomes* (sticky bridges).

2- Ring chromosome(s):

- It appears as a ring inside the divided phase.
- It is a chromosome whose *arms are fused together to form a ring*.

3- Lagging chromosomes (Laggards):

- Whereas some chromosomes separate from the rest of chromosomes and fail to be included in the grouping.
- This is **due to** the *non-attachment of laggards to spindle fibers so, they cannot move to either of the two poles*.

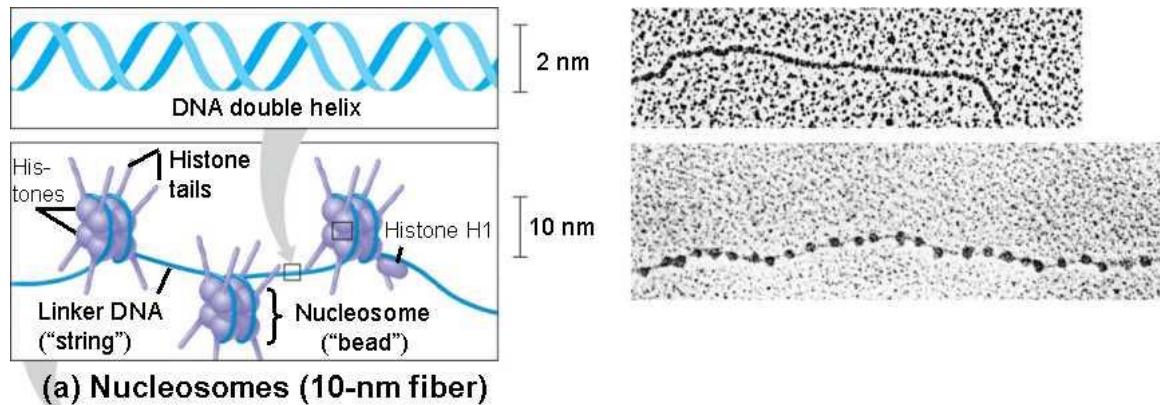
4- Chromosome breakage (fragmentation):

- This represents the most abundant abnormality in chromosome disturbances.
- It appears with one or more chromosomal fragments during mitosis.

- This reflects a probable *activation of a nuclease enzyme* generating small inter-nucleosomal fragments.

N.B.

Nucleosome is the basic unit of DNA package in eukaryotes. It consists of a segment of DNA bound around a protein core as a thread wrapped around a spool.



5- Chromosomal loss:

It appears when chromosome fragments or laggards lost their way in the dividing cell and cannot incorporate in any of the two daughter nuclei.

D) Nuclear Disturbances

1- Micronuclei (MN)

- Cells bearing micronuclei were observed at interphase and prophase stages.
- Micronuclei may originate from *chromosome fragments or laggards*

2- Nuclear budding:

- It appears when a nucleus may originate a bud.
- This is due to the *exceeding DNA material*.

3- Pycnosis

- It appears as extra-condensation and darken stained nucleus.
- This is due to the *irreversible condensation of chromatin* in the nucleus of the cell (at interphase or prophase)
- Pycnotic nuclei undergo **necrosis** (or **apoptosis** i.e. programmed cell death) through *karyorrhexis* (fragmentation) then complete degradation (*karyolysis*) of the nucleus resulting in cell death.



4- Nuclear Lesion(s) (Vacuolated nucleus):

- It appears with numerous vacuoles inside the nucleus.
- It is **due to** the *inhibition of DNA biosynthesis* by the effect of nuclear poisons.

E) Cytoplasmic Disturbances

Bi-nucleated (or multi-nucleated cells):

- Where cells are seen with more than one large nucleus.
- They **occur when** the *nucleus divided normally but cytokinesis process of cell division was inhibited*.

F) Cellular Disturbances

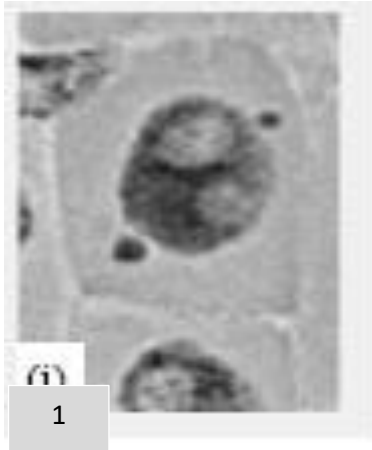
Ghost cell

- It is an **apoptotic** (dead) cell in which the outline is visible but nucleus and cytoplasmic structures are *not stainable*.
- **Apoptosis** is a biological process by which the *unnecessary or damaged cells die*.

Student name:Code number:

STUDENT'S ASSIGNMENT
Give it to your laboratory instructor

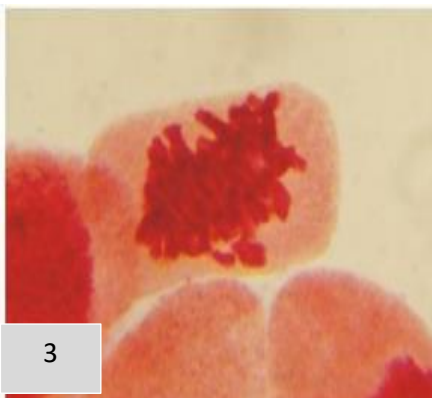
Q. Identify the following Aberrations:



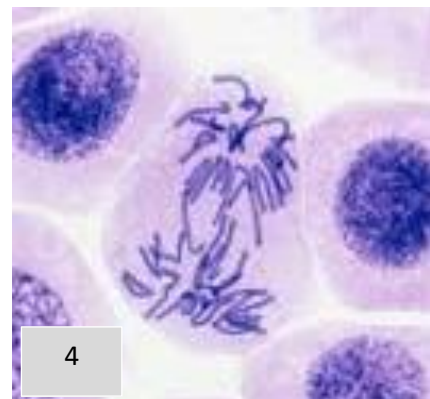
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