SNS COLLEGE OF ALLIED HEALTH SCIENCE

Affiliated to The Tamil Nadu Dr. M.G.R Medical University, Chennai

DEPARTMENT OF RADIOGRAPHY AND IMAGING TECHNOLOGY

COURSE NAME : Quality Control, Radiobiology andRadiation Safety in Radiodiagnosis/Imaging otherthan X-ray related.

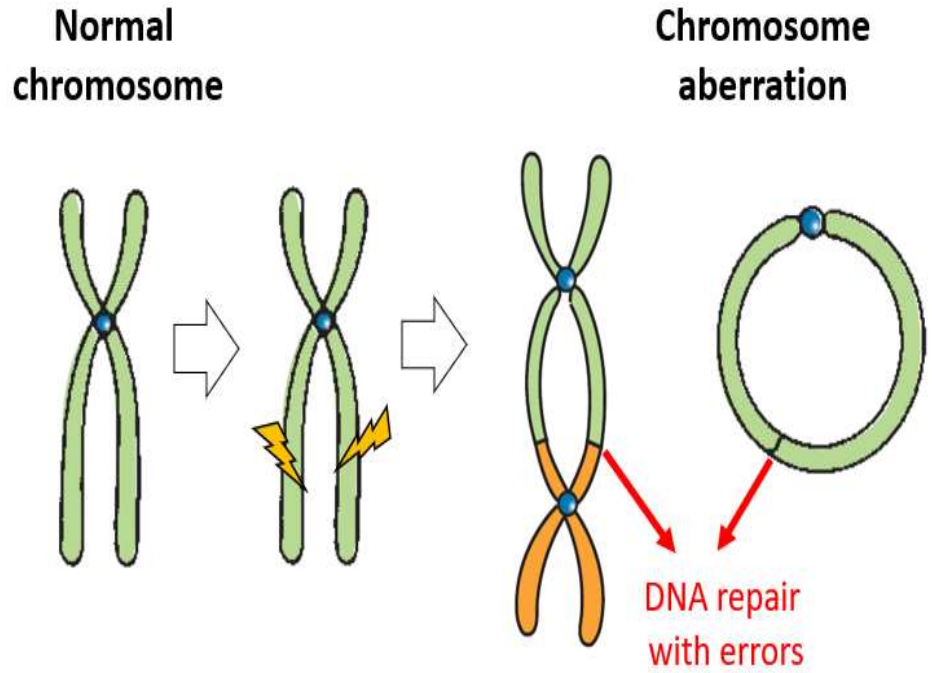
UNIT:1 Radiation Quantities and Units

TOPICS : Chromosomal Aberrations

FACULTY NAME : Ms. DHANA LAKSHMI .M

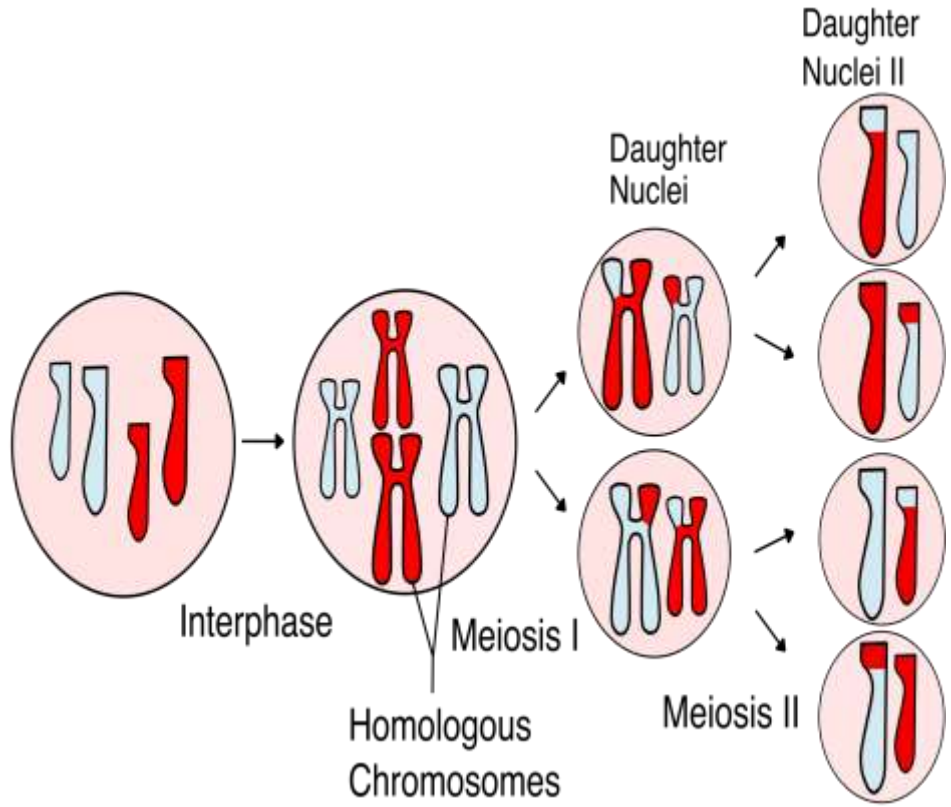
INTRODUCTION TO CHROMOSOMAL ABERRATIONS

- Any change in normal chromosome structure or number
- Also called chromosomal mutations or chromosomal abnormalities



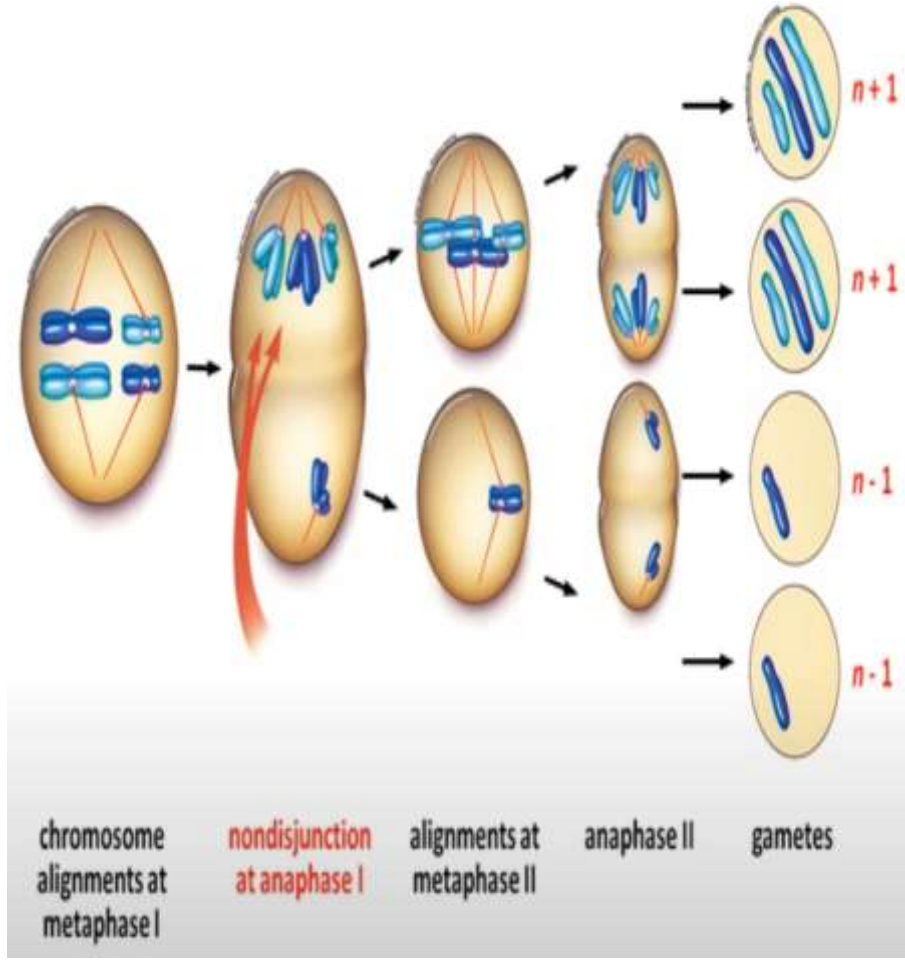
Classification of Chromosomal Aberrations

- Numerical aberrations → Change in chromosome number (Euploidy or Aneuploidy)
- Structural aberrations → Change in chromosome structure (Deletions, Duplications, Inversions, Translocations, Rings, Isochromosomes)

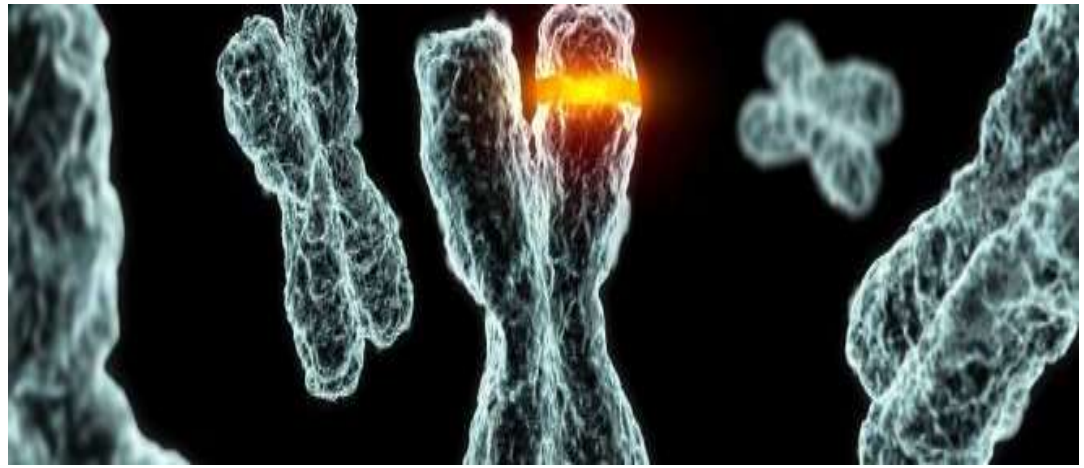


Numerical Aberrations

- Complete extra sets of chromosomes
- Triploidy (3n), Tetraploidy (4n) – common in plants, lethal in humans
- Mostly arise from failure of meiosis or fertilization errors
- Rare survivors usually mosaic (some cells normal, some polyploid)



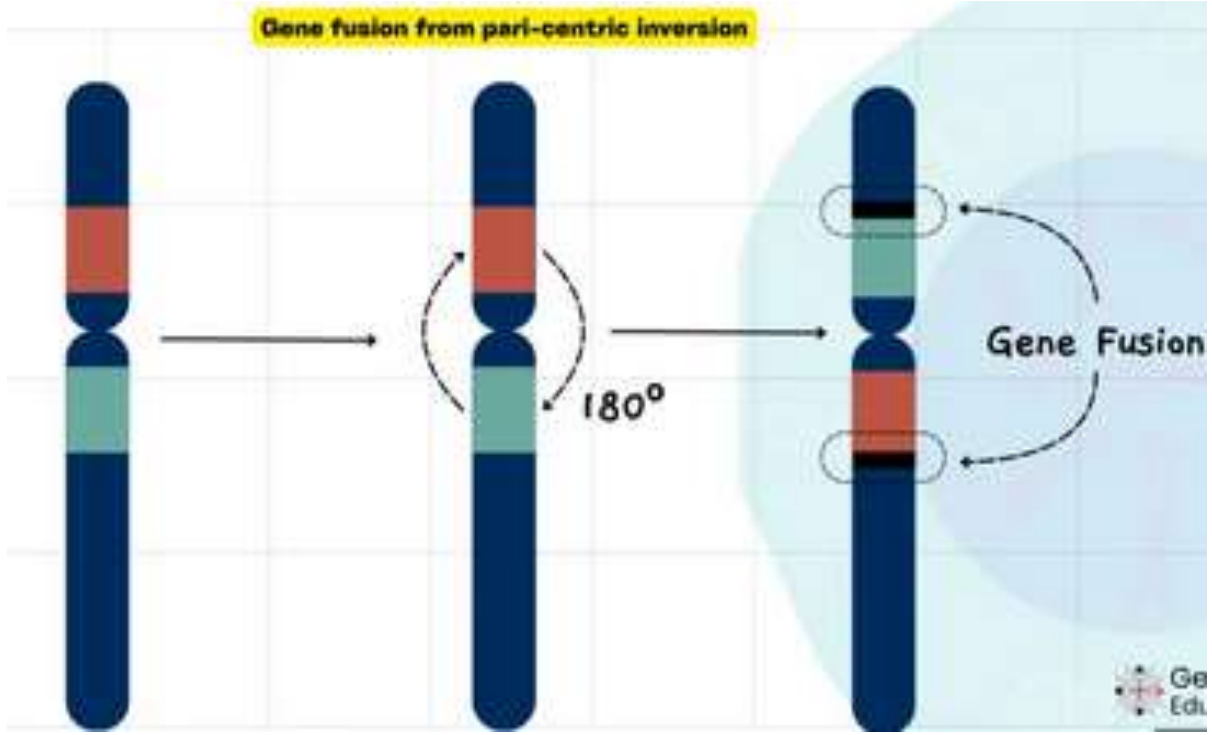
- Gain or loss of individual chromosomes
- Monosomy (2n–1) → e.g., Turner syndrome (45,XO)
- Trisomy (2n+1) → e.g.,
- Trisomy 21 → Down syndrome
- Trisomy 18 → Edwards syndrome
- Trisomy 13 → Patau syndrome
- Cause: Non-disjunction during meiosis I or II



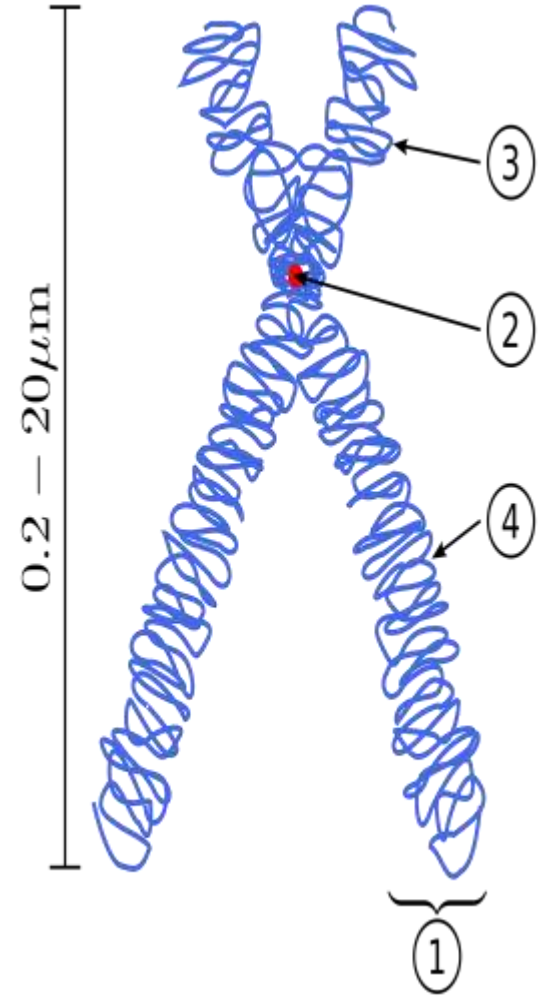
Structural Aberrations

Loss of a chromosome segment

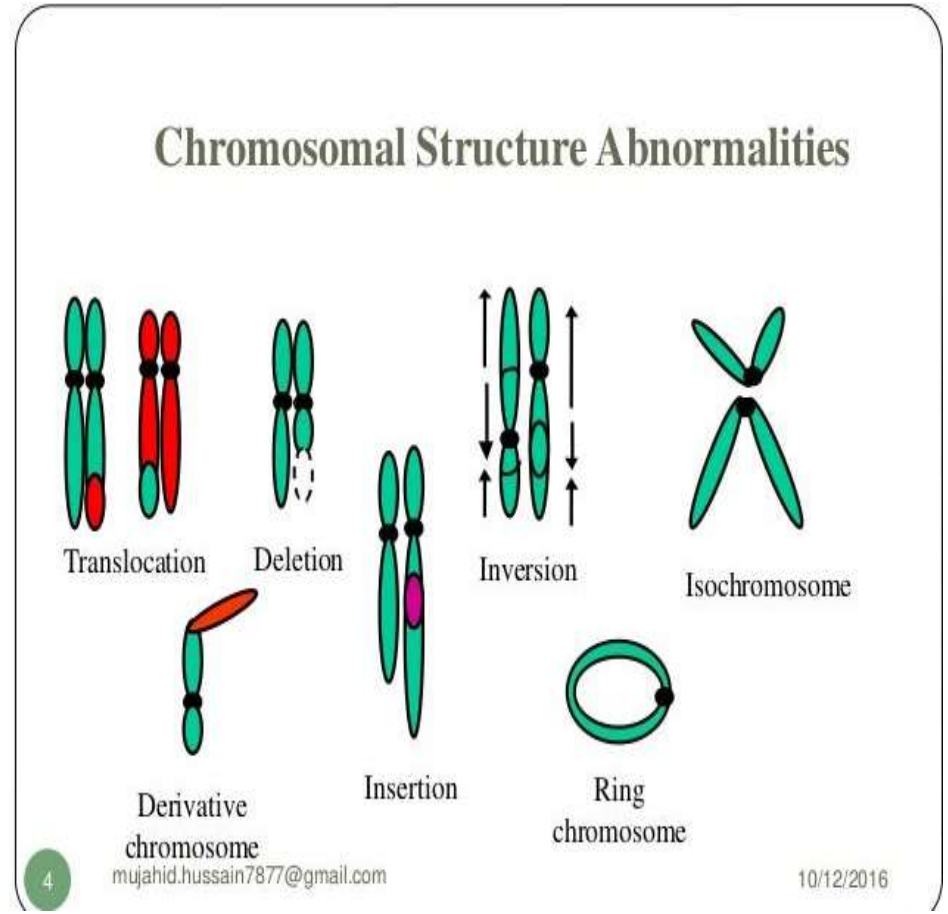
- Types:
- Terminal deletion
- Interstitial deletion



- Extra copy of a chromosome segment
- May be tandem or inverted
- Example:
- Charcot-Marie-Tooth disease type 1A (duplication 17p12)
- Can cause gene dosage imbalance

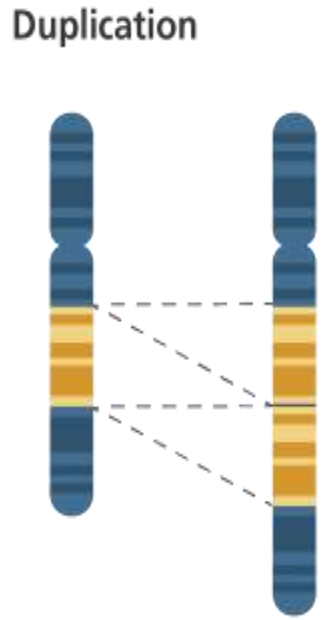


- Segment reversed end-to-end
- Two types:
- Paracentric (does not include centromere)
- Pericentric (includes centromere)
- Usually balanced → carrier normal, but risk of unbalanced gametes in meiosis



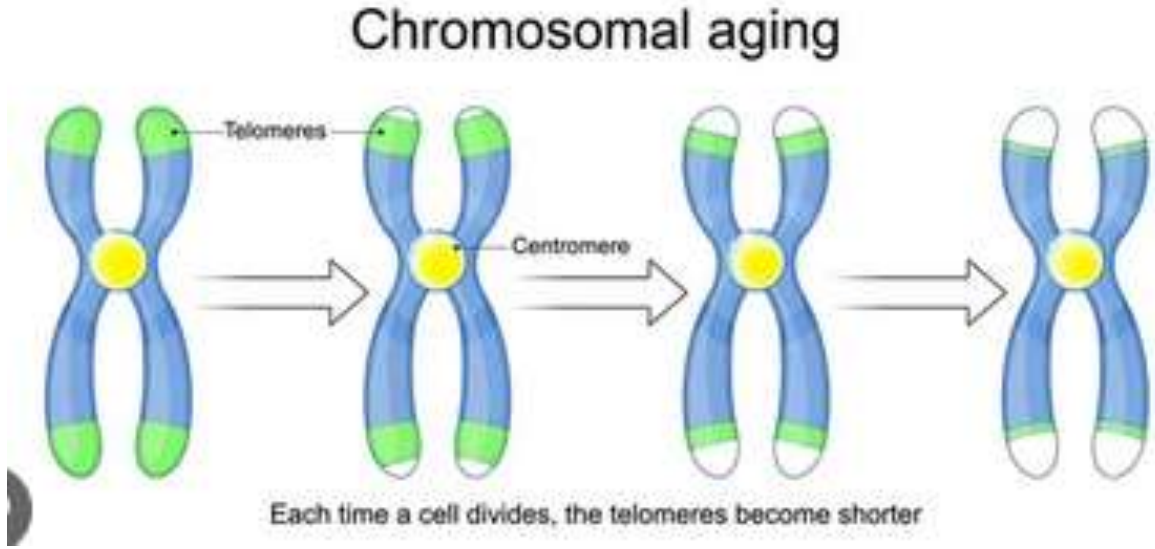
Causes of Chromosomal Aberrations

- Spontaneous errors in cell division
- Ionizing radiation (X-rays, γ-rays)
- Chemicals (alkylating agents, topoisomerase inhibitors)
- Viruses (rare)
- Advanced maternal age (aneuploidy risk ↑)



Syndrome	Abnormality	Major Features
Down syndrome	Trisomy 21	Intellectual disability, flat face, heart defects
Turner syndrome	45,XO	Short stature, infertility, webbed neck
Klinefelter syndrome	47,XXY	Tall, infertility, gynecomastia
Cri-du-chat	5p deletion	Cat-like cry, microcephaly
Chronic Myeloid Leukemia	Philadelphia	Uncontrolled WBC production

- Spontaneous errors in cell division
- Ionizing radiation (X-rays, γ-rays)
- ChemicalS
- Viruses (rare)
- Advanced maternal age



SUMMARY

- Chromosomal aberrations = numerical or structural changes
- Numerical → mostly aneuploidy in humans (trisomy/monosomy)
- Structural → deletion, duplication, inversion, translocation most common
- Many cause syndromes, miscarriages, or infertility
- Detected by karyotype, FISH, chromosomal microarray

References

- https://plantlet.org/chromosomal-aberration-structural-and-numerical/
- https://rsscience.com/chromosome/
- https://www.tutorsglobe.com/homework-help/botany/chromosomal-aberrations-71752.aspx