Genetics Lecture 1

1. Introduction to Cytogenetics
Cytogenetics is the branch of genetics that studies chromosome structure, number, and their role in disease. The correct human chromosome number (46) was established in 1956, correcting earlier misconceptions. This field is essential in understanding genetic disorders, cancers, and developmental abnormalities. Each human cell contains 23 pairs of chromosomes, where one chromosome of each pair is inherited from the father and the other from the mother.

2. DNA, Genes, and Genome
DNA (Deoxyribonucleic Acid) is the hereditary material composed of two antiparallel strands forming a double helix. It carries genetic instructions necessary for cell function and reproduction. A gene is a functional unit of DNA that encodes proteins or RNA molecules. The human genome represents the entire DNA content of a human being. It was largely sequenced in 2003–2004, but complete telomere-to-telomere sequencing was achieved only recently, highlighting the continuous evolution of genetic knowledge.

3. Chromosome Structure
DNA is packaged efficiently within the nucleus by wrapping around histone proteins to form nucleosomes. These nucleosomes coil further into chromatin fibers, which condense to form chromosomes during cell division. Each chromosome consists of two identical sister chromatids joined at a centromere. Each chromatid contains one continuous double-stranded DNA molecule. This organization allows DNA to be compact yet functional.

4. Chromatin vs Chromosomes
During interphase, DNA exists in a loosely packed form called chromatin, allowing access for transcription and replication. In contrast, during mitosis, DNA condenses into visible chromosomes to ensure accurate distribution to daughter cells. This dynamic structural change is essential for balancing gene expression and genome stability.

5. Cell Cycle
The cell cycle consists of Interphase and the Mitotic (M) phase. Interphase includes: G1 phase (cell growth and protein synthesis), S phase (DNA replication), and G2 phase (preparation for mitosis). The M phase includes mitosis and cytokinesis. The cell must carefully regulate these phases to ensure proper DNA duplication and division.

6. Chromosome Classification
Chromosomes are classified based on the position of the centromere into: Metacentric (centromere in the middle), Submetacentric (slightly off-center), and Acrocentric (near one end). Telocentric chromosomes are not found in humans. The short arm is called the p arm, while the long arm is called the q arm.

7. Human Chromosomes
Human somatic cells are diploid (2n = 46), containing 22 pairs of autosomes and one pair of sex chromosomes (XX or XY). Germ cells (gametes) are haploid (n = 23), containing one set of chromosomes. Fertilization restores the diploid state by combining genetic material from both parents.

8. Mitosis
Mitosis produces two genetically identical daughter cells. It includes four main stages: Prophase (chromosome condensation and spindle formation), Metaphase (alignment of chromosomes at the metaphase plate), Anaphase (separation of sister chromatids), and Telophase (reformation of nuclear membrane). Cytokinesis divides the cytoplasm. Mitosis ensures genetic stability and is

essential for growth and tissue repair.

9. Meiosis
Meiosis is a specialized cell division that produces four haploid gametes. It consists of two divisions: Meiosis I (reduction division) and Meiosis II (similar to mitosis). In Meiosis I, homologous chromosomes separate, reducing chromosome number by half. In Meiosis II, sister chromatids separate.

10. Crossing Over and Recombination
During prophase I of meiosis, homologous chromosomes pair and exchange genetic material through crossing over. This occurs at structures called chiasmata. This process produces new combinations of alleles and increases genetic diversity. Errors in this process can lead to chromosomal abnormalities.

11. Independent Assortment
During metaphase I, homologous chromosome pairs align randomly. This leads to independent assortment of maternal and paternal chromosomes. The number of possible combinations is given by 2^n, where n is the number of chromosome pairs.

12. Genetic Significance
Meiosis contributes to genetic diversity through recombination and independent assortment. Mitosis ensures genetic consistency. Accurate chromosome segregation is crucial; errors can result in diseases such as aneuploidy or cancer.

13. Key Exam Notes
- Mitosis = identical cells, Meiosis = variation
- Meiosis I is reductional division
- Crossing over occurs only in Prophase I
- Independent assortment occurs in Metaphase I
- Chromatin = loose (interphase), Chromosomes = condensed (mitosis)
- Sister chromatids separate in mitosis and meiosis II