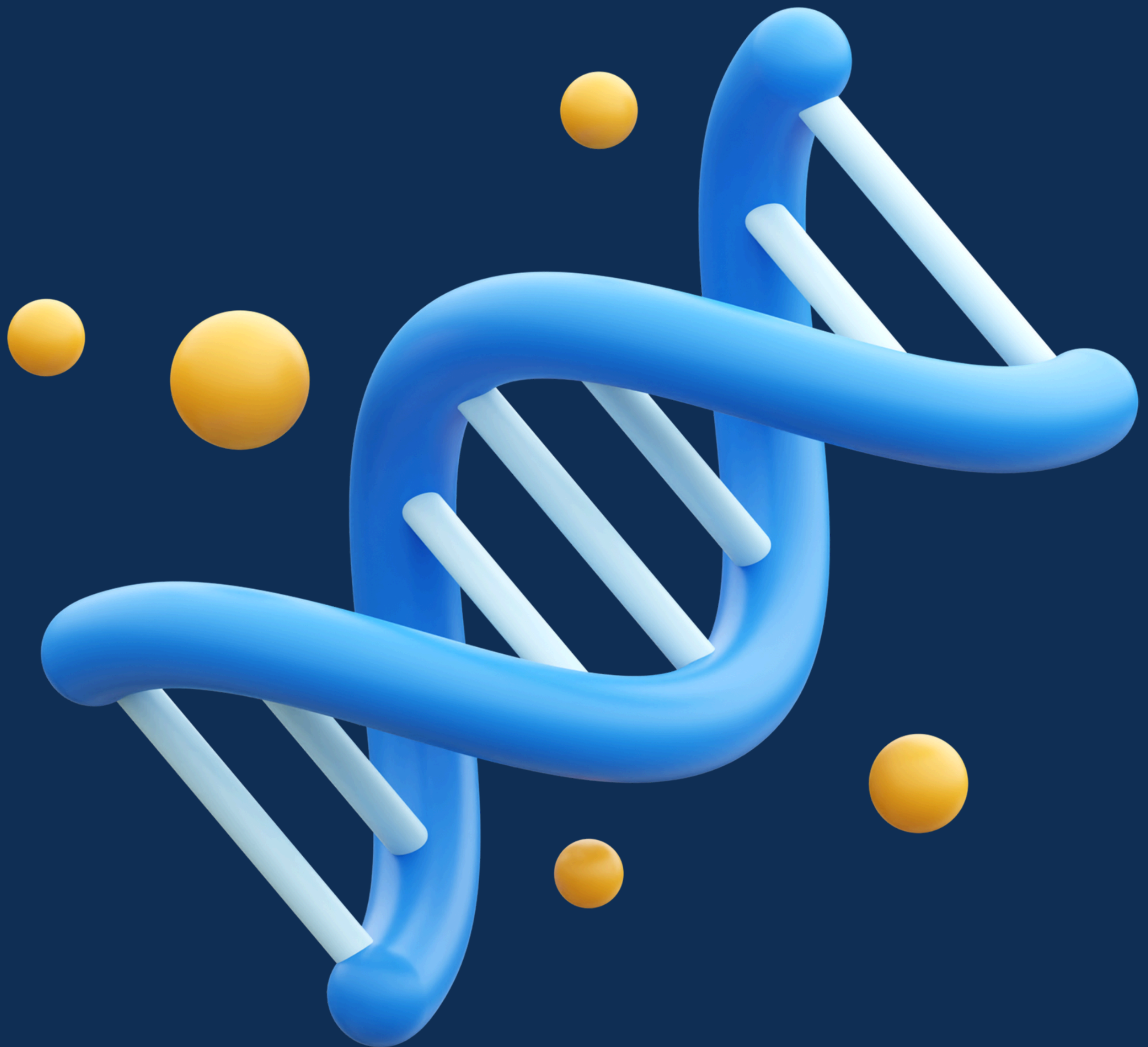


# GENETICS

Your genetics load the gun. Your lifestyle pulls the trigger. - Mehmet Oz



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# introduction

1. What is the basic unit of inheritance that is a segment within a long strand of DNA with specific instructions for the production of one specific protein?
  - a. Chromosome
  - b. Genome
  - c. Allele
  - d. Gene
2. Which branch of biology deals with heredity and variation of organisms?
  - a. Biochemistry
  - b. Genetics
  - c. Microbiology
  - d. Botany
3. Who discovered the basic principles of genetics and inferred laws that allowed predictions about trait inheritance?
  - a. Charles Darwin
  - b. Gregor Mendel
  - c. Louis Pasteur
  - d. Rosalind Franklin
4. How are alleles defined in genetics?
  - a. Individual copies of a particular gene
  - b. Physical manifestation of a trait
  - c. Genetic constitution of an individual
  - d. Singular units of chromosomes
5. What does Mendel's law of segregation state in genetics?
  - a. Genes assort independently of each other
  - b. Alleles for each trait separate during gamete formation
  - c. Dominant alleles always mask recessive alleles
  - d. Traits are transmitted as autosomal or sex-linked
6. Which structure in cells manages the storage, duplication, and expression of DNA?
  - a. Ribosome
  - b. Nucleus
  - c. Mitochondria
  - d. Endoplasmic reticulum
7. What encodes all of the proteins a cell or organism can make?
  - a. Genome
  - b. Ribosome
  - c. Chloroplast
  - d. Lysosome

**8. What is the branch of genetics concerned with human biological variation as it relates to health and disease?**

- a. Molecular genetics**
- b. Cytogenetics**
- c. Population genetics**
- d. Medical genetics**

**9. What is the use of molecular biology techniques to increase scientific knowledge of the natural history of diseases and identify individuals at risk for developing specific diseases?**

- a. Molecular diagnosis**
- b. Genetic counseling**
- c. Gene therapy**
- d. Immunotherapy**

**10. What is the study of the structure and function of chromosomes known as?**

- a. Genomics**
- b. Proteomics**
- c. Cytogenetics**
- d. Transcriptomics**

**11. Which law of genetics states that alleles of different genes assort independently of each other?**

- a. Mendel's law of segregation**
- b. Mendel's law of independent assortment**
- c. Hardy-Weinberg equilibrium**
- d. Gene linkage theory**

**12. What process involves gene transcription to produce mRNA and translation to produce a polypeptide?**

- a. Replication**
- b. Transduction**
- c. Transformation**
- d. Gene expression**

**13. In genetics, what masks or hides the expression of its alternative allele?**

- a. Dominant allele**
- b. Recessive allele**
- c. Homologous chromosome**
- d. Genotype**

**14. How is the genetic composition of an individual with respect to a particular trait defined?**

- a. Phenotype**
- b. Genotype**
- c. Allele**
- d. Homozygous**

**D  
A  
C  
B  
D  
A  
B**

**15. Which law of genetics states that the two alleles for each trait separate during gamete formation?**

- a. Mendel's law of segregation**
- b. Mendel's law of independent assortment**
- c. Law of dominance**
- d. Law of co-dominance**

**16. What is the study of the genetics of populations known as?**

- a. Molecular genetics**
- b. Cytogenetics**
- c. Population genetics**
- d. Medical genetics**

**17. Which type of genes are located on a sex chromosome?**

- a. Autosomal genes**
- b. Recessive genes**
- c. Dominant genes**
- d. Sex-linked genes**

**18. What does molecular diagnosis of human disorders involve?**

- a. Detection of disease-causing variants in DNA**
- b. Structural analysis of proteins**
- c. Cell division rate measurement**
- d. Gene splicing**

**20. What encodes the entire collection of chromosomes in each cell of an organism?**

- a. Genome**
- b. Ribosome**
- c. Nucleus**
- d. Endoplasmic reticulum**



# DNA Packaging, Chromosomes, and Cell division

1. Which proteins are involved in folding and packaging eukaryotic chromosomes?
  - a. Actins
  - b. Tubulins
  - c. Histones
  - d. Ribosomes
2. What is the primary role of histone proteins in chromosome packaging?
  - a. Replication
  - b. Transcription
  - c. DNA compaction
  - d. RNA splicing
3. How many histones are present in each nucleosome core particle?
  - a. 4
  - b. 6
  - c. 8
  - d. 10
4. Which of the following is responsible for condensing chromosomes at mitosis?
  - a. Condensin
  - b. Cohesin
  - c. Polymerase
  - d. Helicase
5. What is the role of cohesins during cell division?
  - a. Promote binding of sister chromatids
  - b. Degrade chromosomes
  - c. Inhibit spindle formation
  - d. Prevent histone acetylation
6. Which protein is important in stabilizing chromatin higher-order structures?
  - a. Polymerase
  - b. Tubulin
  - c. Histone H1
  - d. Actin
7. What is the role of telomeres in chromosomes?
  - a. DNA replication
  - b. Chromosome segregation
  - c. Prevent chromosome shortening
  - d. Transcription regulation

**8. Which type of chromatin is transcriptionally inactive and tightly compacted?**

- a. Euchromatin**
- b. Heterochromatin**
- c. Telomeric chromatin**
- d. Centromeric chromatin**

**9. During which phase of the cell cycle does DNA replication occur?**

- a. G1 phase**
- b. G2 phase**
- c. S phase**
- d. M phase**

**10. What is the primary purpose of mitosis?**

- a. Reduce chromosome number**
- b. Generate genetic diversity**
- c. Distribute replicated chromosomes**
- d. Create haploid cells**

**11. In which stage of mitosis do sister chromatids separate and move to opposite poles?**

- a. Prophase**
- b. Metaphase**
- c. Anaphase**
- d. Telophase**

**12. What type of microtubules attach to the kinetochore at the centromere of each chromosome?**

- a. Aster microtubules**
- b. Polar microtubules**
- c. Kinetochore microtubules**
- d. Centromeric microtubules**

**13. Which stage of mitosis involves the alignment of chromosomes at the cell's equator?**

- a. Prophase**
- b. Prometaphase**
- c. Metaphase**
- d. Anaphase**

**14. What occurs during anaphase of mitosis?**

- a. Chromosomes align at the equator**
- b. Nuclear envelope forms**
- c. Sister chromatids separate**
- d. Chromosomes decondense**

**B  
C  
C  
C  
C  
C  
C**

**15. When do the nuclear membranes reform to form two separate nuclei during cell division?**

- a. Prophase**
- b. Metaphase**
- c. Telophase**
- d. Anaphase**

**16. Which phase of the cell cycle involves the duplication of DNA and chromosomes?**

- a. G1 phase**
- b. S phase**
- c. G2 phase**
- d. M phase**

**17. What is the role of the spindle apparatus in cell division?**

- a. DNA replication**
- b. Energy production**
- c. Chromosome separation**
- d. Protein synthesis**

**18. Which structure assists in positioning the spindle during cell division?**

- a. Aster microtubules**
- b. Polar microtubules**
- c. Kinetochore proteins**
- d. Centromeres**

**19. What balances the forces pushing and pulling chromosomes during metaphase of mitosis?**

- a. Aster microtubules**
- b. Kinetochore proteins**
- c. Polar microtubules**
- d. Centromeres**

**20. At the end of which phase does cytokinesis occur to complete the cell division process?**

- a. Telophase**
- b. G2 phase**
- c. Anaphase**
- d. Prophase**

**C  
B  
C  
A  
C  
A**

# Cell division - Meiosis

1. What is the most common way for eukaryotic organisms to produce offspring?
  - a. Asexual reproduction
  - b. Mutation
  - c. Regeneration
  - d. Sexual reproduction
2. Which type of cell division produces gametes (eggs and sperm)?
  - a. Mitosis
  - b. Interphase
  - c. Meiosis
  - d. Cytokinesis
3. How many rounds of cell division does meiosis involve?
  - a. One
  - b. Two
  - c. Three
  - d. Four
4. What is the result of Meiosis I?
  - a. Two haploid daughter cells with replicated chromosomes
  - b. Four haploid daughter cells with unreplicated chromosomes
  - c. Diploid cells with duplicated chromosomes
  - d. Tetraploid daughter cells
5. The first three substages of prophase I are known as:
  - a. Alpha, Beta, Gamma
  - b. Delta, Epsilon, Zeta
  - c. Leptotene, Zygotene, Pachytene
  - d. Diplotene, Diakinesis, Telophase
6. What happens during pachytene of prophase I?
  - a. Synaptonemal complex dissolves
  - b. Recombination nodules disappear
  - c. Crossing-over occurs between sister chromatids
  - d. Chromosome pairs become tetrads
7. What is visible during diplotene?
  - a. Synaptonemal complex formation
  - b. A tetrad of four chromatids
  - c. Nonsister chromatids pulling apart completely
  - d. Microtubules penetrating the nucleus
8. What occurs during diakinesis?
  - a. Condensation of chromatids
  - b. Nonsister chromatids remain connected at chiasmata
  - c. Homologous chromosomes separate
  - d. Meiotic arrest begins

D  
C  
B  
B  
C  
B  
B  
B



**9. During which phase of meiosis I do homologous chromosomes move to opposite poles?**

- a. Prophase I**
- b. Metaphase I**
- c. Anaphase I**
- d. Telophase I**

**10. What happens during telophase I?**

- a. The nuclear envelope re-forms**
- b. Sister chromatids move to opposite poles**
- c. DNA replication occurs**
- d. Cytokinesis precedes nuclear division**

**11. How many haploid cells are formed at the end of meiosis II?**

- a. Two**
- b. Three**
- c. Four**
- d. Five**

**12. What defines meiosis II as an equational division?**

- a. Reduction of chromosome number**
- b. Random assortment of chromosomes**
- c. Sister chromatids separate**
- d. Association of chromatids at chiasmata**

**13. What increases the possible chromosomal combinations during meiosis?**

- a. Independent assortment of genes**
- b. Sister chromatid attachment**
- c. Centromere division**
- d. Recombination between chromosome pairs**

**14. The law of segregation states that:**

- a. Chromosomes assort independently**
- b. Chromosome number reduces during meiosis**
- c. Alleles segregate during gamete formation**
- d. Chromosomes duplicate once**

**15. What is a consequence of crossing-over in meiosis?**

- a. Random assortment of chromosomes**
- b. Diploid to haploid transition**
- c. Association of chromatids at chiasmata**
- d. Production of new gene combinations**

C  
D  
C  
C  
D  
C  
C

**16. Chromosome combinations during meiosis illustrate:**

- a. Random assortment of maternal and paternal chromosomes**
- b. Diploid to diploid transition**
- c. Homologous chromosomes pairing**
- d. Chromosome duplication**

**17. What does independent assortment mean?**

- a. Fate of a gene on one chromosome is influenced by others**
- b. Genes on different chromosomes segregate independently**
- c. Chromatids separate during meiosis II**
- d. Alleles couple during synapsis**

**18. Which phase involves the sorting events similar to those in mitosis?**

- a. Prophase II**
- b. Metaphase II**
- c. Anaphase II**
- d. Telophase II**

**19. What occurs during telophase II?**

- a. Chromosomes begin to condense**
- b. Nucleoli re-form**
- c. Nuclear envelope breaks down**
- d. Half the number of chromosomes is present**

**20. How many new haploid cells are formed during cytokinesis in meiosis II?**

- a. Two**
- b. Three**
- c. Four**
- d. Five**

# Karyotyping , Chromosome structure

1. Why is the knowledge of chromosomes important in many areas of clinical medicine and research?
  - a. Approximately 10-15% of live born infants have a chromosomal abnormality.
  - b. Chromosomal aberrations are noted in 50% to 67% of spontaneous miscarriages.
  - c. It plays a role in the majority of cells from leukemia samples or solid tumors.
  - d. Only 1% of individuals having sex reversal or pubertal anomalies show chromosomal aberrations.
2. What is the approximate percentage of recognized embryonic and fetal deaths with cytogenetic abnormalities?
  - a. 10%
  - b. 30%
  - c. 45%
  - d. 60%
3. Which of the following is NOT a tissue commonly used for chromosome studies?
  - a. Bone marrow
  - b. Saliva
  - c. Amniotic fluid cells
  - d. Tumor biopsies
4. What is the main referral reason for cytogenetic analyses related to postnatal, childhood growth, and development?
  - a. Cognitive development
  - b. Growth failure
  - c. Behavioral concerns
  - d. Hearing loss
5. Which banding technique involves treating metaphase spreads with trypsin?
  - a. G-banding
  - b. R-banding
  - c. Q-banding
  - d. C-banding
6. Which chromosome banding technique treats metaphase spreads with the chemical quinacrine mustard?
  - a. G-banding
  - b. R-banding
  - c. Q-banding
  - d. C-banding
7. What does R-banding stand for in chromosome analysis?
  - a. Reverse banding
  - b. Red banding
  - c. Rapid banding
  - d. Random banding

B  
C  
B  
B  
A  
C  
A

8. What banding technique stains regions that contain rRNA genes within the NOR?

- a. G-banding
- b. R-banding
- c. C-banding
- d. silver-nitrate

9. What is a band defined as in a chromosome?

- a. A part of the chromosome with no staining properties
- b. A segment without genes
- c. A distinguishable part of the chromosome based on staining properties
- d. A structure only visible in males

10. What divides the chromosomes into the short "p" and long "q" arms?

- a. Nucleolus
- b. Telomeres
- c. Centromere
- d. Chromatids

11. What is depicted in a human chromosome ideogram?

- a. Actual chromosomes
- b. Stained chromosomes
- c. Diagrammatic representation of a karyotype
- d. Electron microscope images

12. High-resolution banding involves staining chromosomes during which phase?

- a. Metaphase
- b. Anaphase
- c. Prophase or prometaphase
- d. Telophase

13. How many bands are observable for all chromosomes in high-resolution banding?

- a. About 50
- b. About 200
- c. About 450
- d. About 700

14. What does G-banding pattern for human chromosome 4 show at increasing levels of resolution?

- a. Increasing darkness
- b. Increasing lightness
- c. Subdivision into sub-bands and sub-subbands
- d. No changes in banding patterns

15. In a high-resolution banding, what increases the number of observable bands per haploid set?

- a. Staining chromosomes during metaphase
- b. Staining chromosomes during prophase
- c. Staining chromosomes during prometaphase
- d. Staining chromosomes during telophase

D  
C  
C  
C  
D  
C  
B

**16. What is essential for cells to undergo chromosome analysis for clinical purposes?**

- a. Cell differentiation**
- b. Cell migration**
- c. Cell proliferation in culture**
- d. Cell apoptosis**

**17. What must be added to blood specimens for culture establishment and harvest?**

- a. Antibiotics**
- b. Immunoglobulins**
- c. Mitogens**
- d. Hemoglobin**

**18. How are chromosomes arranged in a karyogram?**

- a. By size**
- b. By color**
- c. By gene sequence**
- d. By region**

**19. What is the main role of cytogenetics?**

- a. Study of cellular metabolism**
- b. Study of genomic structure only**
- c. Study of chromosomes and genomic structure in relation to human disease and heredity**
- d. Study of protein folding**

C  
C  
C  
C



# Autosomal chromosomes and numerical chromosomal aberrations

1. What is the main purpose of Fluorescent In-Situ Hybridization (FISH)?

- a. Identify different banding patterns
- b. Study genetic aberrations too small for standard DNA sequencing
- c. Analyze metaphase cells for mutation detection
- d. Detect chromosomal polymorphisms

2. How does FISH work in detecting genetic segments?

- a. Patient DNA segment binds to probe, visible fluorescent signal is present
- b. Patient DNA segment doesn't bind to probe, no fluorescent signal seen
- c. Probe binds to any DNA in the cell
- d. Probe remains inactive in the presence of genetic mutations

3. Which of the following is NOT a type of FISH probe?

- a. Centromeric probes (CEP)
- b. Spectral karyotype probes
- c. Locus specific probes
- d. Whole chromosome probes

4. What is the method typically used to observe chromosomes or hybridized regions in FISH?

- a. Electron microscopy
- b. Light microscopy
- c. Fluorescent microscope
- d. Confocal microscope

5. What does Trisomy 21 refer to?

- a. Extra chromosome 21 in males
- b. Monosomy in chromosome 21
- c. Normal female chromosome composition
- d. Down syndrome (extra chromosome 21)

6. Which chromosome abnormality results in Turner syndrome?

- a. 47,XXY
- b. 45,X
- c. 47,XXX
- d. 47,XYY

7. What is the prevalence of Trisomy 18 (Edward syndrome) among live births?

- a. 1 per 6,000 births
- b. 1 per 2,000 births
- c. 1 per 10,000 births
- d. 1 per 4,000 births

8. What is the main feature of Klinefelter syndrome (47,XXY)?

- a. Hypogonadism with large testes
- b. Short stature with lymphedema
- c. high testosterone with gynecomastia
- d. Tall stature with hypogonadism

B  
A  
B  
C  
D  
B  
A  
D

9. How many chromosomes are present in a Trisomy 13 individual?
- 46 chromosomes
  - 47 chromosomes
  - 48 chromosomes
  - 45 chromosomes
10. Which type of aneuploidy involves individuals lacking one chromosome?
- Trisomic
  - Monosomic
  - Tetrasomic
  - Polyploidy
11. What is the characteristic feature of Turner syndrome?
- Tall stature with gynecomastia
  - Hypogonadism with small testes
  - Webbing of the neck
  - High testosterone levels
12. How does FISH contribute to genetic studies?
- Detects large chromosomal changes accurately
  - Determines the exact gene mutations in a patient
  - Allows visualization of small DNA segments
  - Identifies chromosomal translocations
13. What is the main difference between Q banding and R banding?
- Use of different fluorescent dyes
  - Visibility under different microscopes
  - Pattern of light and dark bands
  - Specific identification of centromeres
14. In which scenario would FISH be particularly useful?
- Detecting large chromosomal deletions
  - Analyzing whole chromosomal mutations
  - Studying polymorphisms in interphase cells
  - Identifying small DNA segments
15. Which chromosomal abnormality involves three copies of chromosome 21?
- Monosomy 21
  - Ditrisomy 21
  - Trisomy 21
  - Tetrasomy 21

B  
B  
C  
C  
C  
D  
C

- 16. What contributes to the incidence of autosomal aneuploidy in newborns?**
- a. Maternal age effect**
  - b. Paternal genetic inheritance**
  - c. Gender of the offspring**
  - d. Environmental factors**
- 17. Why are the trisomies 13, 18, and 21 relatively mild compared to other aneuploidies?**
- a. Low gene content on involved chromosomes**
  - b. Presence of multiple chromosomal deletions**
  - c. High occurrence of genetic mutations**
  - d. Structural abnormalities in the chromosomes**
- 18. Which chromosomal condition results in an extra X chromosome in males?**
- a. Trisomy 18**
  - b. Klinefelter syndrome**
  - c. Turner syndrome**
  - d. Trisomy 13**
- 19. What is the estimated survival rate for infants with Trisomy 13?**
- a. 90% survive to 12 months**
  - b. 50% survive within the first weeks of life**
  - c. 95% die in the first year of life**
  - d. 75% survive but with developmental disabilities**
- 20. What is the main negative impact of additional X or Y chromosomes?**
- a. Decrease in physical health**
  - b. Increase in intelligence quotient**
  - c. Higher risk of leukemia**
  - d. Reduction in average IQ**

## Sex chromosomes and structural chromosomal aberration

1. What is the principal cause of human triploidy?
  - a. Dispermy
  - b. Fertilization of a diploid ovum
  - c. Fertilization by a diploid sperm
  - d. Meiosis faults
2. How does tetraploidy typically arise?
  - a. Dispermy
  - b. Endomitosis
  - c. Trisomy
  - d. Aneuploidy
3. Which chromosomal abnormality is typically associated with Klinefelter syndrome?
  - a. 46,XY, del(5p)
  - b. 47,XXY
  - c. 46,XX
  - d. 45,X
4. What type of break leads to a terminal deletion on a chromosome?
  - a. Single break
  - b. Double break
  - c. Inverted break
  - d. Centromere break
5. What is the main phenotypic effect of deletions?
  - a. Increased height
  - b. Intellectual disability
  - c. Enhanced vision
  - d. Improved memory
6. A translocation results from an exchange of genetic material between \_\_ chromosomes.
  - a. Nonhomologous
  - b. Identical
  - c. Tetraploid
  - d. Aneuploid
7. What is the term used for an inversion involving the centromere of a chromosome?
  - a. Pericentric
  - b. Paracentric
  - c. Reciprocal
  - d. Translocation

C  
B  
B  
A  
B  
A  
A

**8. Which type of duplication involves the reorientation of a chromosomal segment?**

- a. Direct**
- b. Inverted**
- c. Reciprocal**
- d. Triplification**

**9. What defines an isochromosome?**

- a. Two identical arms**
- b. Two different arms**
- c. Lost arm**
- d. No arms**

**10. In which syndrome is a 22q11.2 deletion typically observed?**

- a. Down syndrome**
- b. Turner syndrome**
- c. Wolf-Hirschhorn syndrome**
- d. DiGeorge Syndrome**

**11. What can chromosomal microarray testing detect?**

- a. Point mutations**
- b. Balanced translocations**
- c. Tiny deletions**
- d. Gene expression levels**

**12. What is the primary advantage of SNP-based microarrays?**

- a. Can detect deletions only**
- b. Do not Directly compare patient to control**
- c. Require high DNA amounts**
- d. Detect only point mutations**

**13. Mosaicism describes a situation where different cells in the same individual have different \_\_ of chromosomes.**

- a. Sizes**
- b. Shapes**
- c. Numbers**
- d. Colors**

**14. Chimerism involves different cell lines that are derived from \_\_ conceptions.**

- a. Identical**
- b. Different**
- c. Similar**
- d. Nonexistent**

**B  
A  
D  
C  
B  
C  
B**



15. How does an insertion differ from a deletion?

- a. Movement of segments between chromosomes
- b. They are the same
- c. Inversions involved
- d. Loss of DNA material

16. Which syndrome is characterized by a distinctive "greek helmet" facies?

- a. Down syndrome
- b. Wolf-Hirschhorn syndrome
- c. Turner syndrome
- d. Cri-du-chat syndrome

17. What is the main issue associated with duplication 22q11.2?

- a. Intellectual disability
- b. Cardiovascular defects
- c. Skin pigmentation anomalies
- d. Growth deficiency

18. Smith-Magenis syndrome is typically due to a deletion on chromosome \_\_.

- a. 7
- b. 17
- c. 5
- d. 4

19. Neurofibromatosis is usually associated with a deletion on chromosome \_\_.

- a. 7
- b. 17
- c. 5
- d. Y

20. What do deletions in Williams syndrome primarily affect?

- a. 600-800kb segments
- b. 200-400kb segments
- c. 1600kb segments
- d. 1400kb segments

D  
B  
A  
B  
B  
C