Medical Genetics

بسم الله الرحمن الرحيم

Test Bank

MID

﴿إِنِّي تَوَكَّلْتُ عَلَى ٱللَّهِ رَبِّي وَرَبِّكُمْ ۚ مَّا مِن دَآبَّةٍ إِلَّا هُوَ ءَاخِذٌۢ بِنَاصِيَتِهَآ ۚ إِنَّ رَبِّي عَلَىٰ صِرَٰطٍ مُّسْتَقِيمٍ﴾

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رَبِّ اشْرَحْ لِي صَدْرِي وَيَسِّرْ لِي أَمْرِي وَاحْلُلْ عُقْدَةً مِّن لِّسَانِي يَفْقَهُوا قَوْلِي

Lec 1

Genetic diversity, and laws of segregation and independent assortment

Q1: Which of the following BEST explains the structural difference between a **nucleosome** and the **30-nm fiber**?

A) Nucleosomes contain sister chromatids, while 30-nm fibers contain homologous pairs.

B) Nucleosomes are formed during the S phase, whereas the 30-nm fiber only appears during the transition to Metaphase.

C) The 30-nm fiber is 2 nm in diameter, whereas the nucleosome is 10 nm in diameter.

D) The nucleosome is the primary level of DNA wrapping around histones, while the 30-nm fiber represents a further coiling of these units.

Ans: D

Q2: A researcher observes a human cell with **23 replicated chromosomes.** Which of the following is the most likely identity of this cell?

A) A somatic cell undergoing Mitosis at Anaphase.

B) A daughter cell immediately following the completion of Meiosis I.

C) A germ cell that has completed Meiosis II.

D) A somatic cell in G_2 phase.

Meiosis I is a reductional division that separates homologous pairs, resulting in haploid cells (n=23) that still consist of sister chromatids.

Ans: B

Q3: Which of the following statements is **NOT** true regarding the S phase of the cell cycle?

A) The amount of DNA in the cell doubles during this period.

B) It occurs during Interphase, between the G1 and G2 stages.

C) Two sister chromatids are formed for every unreplicated chromosome.

D) The chromosome number of a somatic cell increases from 46 to 92.

Ans: D

Q4: If a chromosome's centromere is located nearly at the very end, resulting in one extremely short arm (p) and one long arm (q) in the normal human chromosome , it is classified as:

A) Acrocentric

B) Metacentric

C) Submetacentric

D) Telocentric

Ans: A

أستغفرالله العظيم وأتوب إليه

Q5: Which of the following events is unique to Prophase I of Meiosis and does **NOT** occur in Mitotic Prophase?

A) Condensation of chromatin into visible chromosomes.

B) Synapsis of homologous chromosomes to form tetrads.

C) Formation of a spindle apparatus from centrioles.

D) Breaking apart of the nuclear envelope.

Ans: B

Q6: During which sub-stage of Prophase I does the actual 'crossing over' or recombination at chiasmata take place?

A) Diplotene

B) Zygotene

C) Leptotene

D) Pachytene

Ans: D

Q7: All of the following are genetic consequences of Meiosis **EXCEPT:**

A) The segregation of alleles into different germ cells.

B) Reduction of the chromosome number from diploid (2n) to haploid (n)

C) Random assortment of maternal and paternal chromosomes into gametes.

D) The production of daughter cells that are genetically identical to the parent cell.

Ans: D

Q8: What is the primary mechanism that allows **for more than** 2^{23} (approx. 8.4 million) possible chromosomal combinations in human gametes?

A) The random fusion of sister chromatids during fertilization.

B) Recombination/Crossing-over between homologous pairs.

C) Independent assortment during Meiosis II.

D) The duplication of DNA during the S phase.

Ans: B

Q9: Which of the following BEST describes the role of the centromere in a replicated chromosome?

A) It is the region where two sister chromatids are held together and where spindle fibers attach.

B) It is the protective cap at the end of the chromosome that prevents degradation.

C) It is the site where DNA synthesis is initiated during the S phase.

D) It is the secondary constriction that contains the genes for ribosomal RNA.

Ans: A

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Q10: In a human male, which of the following is true regarding the chromosome composition of a germ cell after Meiosis II?

A) It contains 46 single chromosomes including both X and Y.

B) It contains 22 pairs of autosomes and one XY pair.

C) It contains 22 autosomes and either an X or a Y chromosome.

D) It contains 23 pairs of homologous chromosomes.

Ans: C

Q11: Which phase of Mitosis is characterized by the alignment of chromosomes along the spindle equator?

A) Anaphase

B) Metaphase

C) Prophase

D) Telophase

Ans: B

Q12: A specific human chromosome has a centromere that is slightly off-center, resulting in a short p arm and a longer q arm. This chromosome is:

A) Telocentric

B) Acrocentric

C) Metacentric

D) Submetacentric

Ans: D

Q13: Which of the following describes the equational division of Meiosis?

A) Meiosis II, where sister chromatids separate.

B) Interphase, where DNA content is tripled.

C) Meiosis I, where homologous chromosomes separate.

D) Mitosis, where homologous pairs are duplicated.

Ans: A

Q14: What is the significance of the 'Chiasma'?

A) It is the point where DNA replication begins.

B) It is the gap between two sister chromatids in a telocentric chromosome.

C) It is the structure that attaches the chromosome to the cell membrane.

D) It represents the site of physical exchange between non-sister chromatids of homologous chromosomes.

Ans: D

Q15: Which of the following BEST explains why genetic diversity is essential for gametes?

A) It ensures that every daughter cell has exactly 46 chromosomes.

B) It allows for new combinations of genes to be produced, aiding in the survival of a species.

C) It guarantees that only dominant alleles are passed to the offspring.

D) It prevents the cell from entering the Go resting phase.

Ans: B

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Lec 2&3

Karyotyping, Chromosome structure and nomenclature

سُبْحَانَ اللَّهِ وَبِحَمْدِهِ

Q1: Which of the following statements regarding the incidence of chromosomal abnormalities is **NOT** true?

A) Roughly 20 – 27% of individuals with pubertal anomalies have noted chromosomal aberrations.

B) Between 33% and 67% of spontaneous miscarriages involve chromosomal aberrations.

C) The majority of cells from solid tumours or leukaemia samples exhibit chromosomal abnormalities.

D) Chromosomal aberrations are present in approximately 10% of all liveborn infants.

Ans: D

Q2: Which of the following BEST distinguishes a karyogram from an ideogram?

A) Karyograms are used for research purposes only, while ideograms are used for clinical diagnosis.

B) Karyograms show only metaphase chromosomes, while ideograms only show prophase chromosomes.

C) A karyogram represents the phenotype, whereas the ideogram represents the genotype.

D) A karyogram is a photograph or computer image of ordered chromosomes, while an ideogram is a diagrammatic representation.

Ans: D

Q3: To perform a clinical chromosome analysis on a peripheral blood sample, which cell type is specifically targeted for its ability to proliferate in culture?

A) Red blood cells (Erythrocytes)

B) Platelets (Thrombocytes)

C) T lymphocytes

D) Neutrophils

Ans: C

Q4: In the G-banding (GTG) process, what is the critical role of trypsin treatment prior to staining with Giemsa?

A) It causes the cells to swell and burst for easier chromosome spreading.

B) It melts the DNA helix specifically in AT-rich regions.

C) It partially digests chromosomal proteins to relax the chromatin structure.

D) It inhibits the spindle fibres to arrest cells in metaphase.

Ans: C

Q5: Which of the following correctly describes the staining pattern observed in R-banding?

A) Dark regions are GC-rich and euchromatic.

B) Only the centromeres and telomeres take up the stain.

C) Dark regions are AT-rich and heterochromatic.

D) Light regions represent transcriptionally active gene clusters.

Ans: A

Q6: What is the primary purpose of adding a hypotonic solution during the harvest of cell specimens?

A) To selectively stain the AT-rich regions of the DNA.

B) To cause osmotic swelling of the cells, facilitating chromosome spreading.

C) To fix the chromosomes permanently to the glass slide.

D) To induce mitotic division in resting cells.

Ans: B

Q7: The normal human karyotype consists of 46 chromosomes. How are these typically arranged in a karyogram?

A) By the number of genes they contain, from highest to lowest.

B) Randomly, based on the order they appear on the microscope slide.

C) By increasing length, with chromosome 22 being the longest.

D) As homologous pairs, generally ordered from longest to shortest.

Ans: D

Q8: Why does high-resolution banding allow for the detection of smaller chromosomal abnormalities compared to conventional banding classified as:

A) It requires the use of silver staining to highlight the nucleolar organiser regions.

B) It involves staining chromosomes during prophase or prometaphase when they are less condensed.

C) It uses more powerful dyes that bind to single base pairs.

D) It utilizes electron microscopy to view the DNA sequence directly.

Ans: B

Q9: All of the following are true regarding telomeres EXCEPT:

A) They are associated with degenerate repeats and unique DNA in the sub-telomeric region.

B) They prevent the end-to-end fusion of chromosomes.

C) They consist of the repetitive (TTAGGG)n sequence.

D) They are located at the centromeric region to assist in segregation.

Ans: D

Q10: Which component is essential for the formation of the kinetochore and proper chromosome segregation?

A) The p arm satellites

B) The Telomere

C) The Centromere

D) The G-light bands

Ans: C

Q11: Which of the following is correct regarding the DNA content of a single G-band seen in a standard 400-band karyotype?

A) A G-band represents approximately 100 to 1,000 base pairs of DNA.

B) A G-band represents several million to 10 million base pairs of DNA.

C) A G-band contains only a single gene.

D) A G-band is composed entirely of non-coding satellite DNA.

Ans: B

Q12: In the standard nomenclature for a normal human male chromosomal complement, the notation used is:

A) 46, XX

B) 23, XY

C) 44, XY

D) 46, XY

Ans: D

Q13: Which banding technique is specifically used to identify gene-rich regions near the telomeres that might be missed by G-banding?

A) R-banding

B) C-banding

C) Q-banding

D) Silver staining

Ans: A

Q14: Based on the ideogram of Chromosome 7, how many regions are identified on the p and q arms respectively?

A) p: 1 region; q: 2 regions

B) p: 2 regions; q: 2 regions

C) p: 2 regions; q: 3 regions

D) p: 3 regions; q: 4 regions

Ans: C

Q15: What is the specific function of Phytohemaglutenin (PHA) in the preparation of a blood specimen for cytogenetic analysis?

A) It inhibits the spindle apparatus during metaphase.

B) It degrades proteins to allow for Giemsa staining.

C) It acts as a fixative to preserve cell structure.

D) It acts as a mitogen to stimulate T lymphocytes to divide.

Ans: D

Q16: Which of the following BEST explains the staining difference in G-banding between light and dark regions?

A) Light regions are areas composed strictly of repetitive satellite DNA.

B) Light regions represent euchromatin that is GC-rich and more transcriptionally active.

C) Dark regions are GC-rich and contain the majority of active genes.

D) Dark regions represent areas where the DNA helix has been melted by heat.

Ans: B

Q17: A researcher is looking to identify a specific DNA sequence on a chromosome. Which of the following is a research use for cytogenetic evaluation?

A) Determining the metabolic rate of a cell.

B) Counting the total number of mitochondria in a lymphocyte.

C) Localization of DNA onto a chromosome.

D) Measuring the concentration of glucose in the blood.

Ans: C

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Lec 4

Autosomal Chromosomes and Numerical Chromosomal Aberrations

Q1: If a nondisjunction event occurs during Meiosis I, which of the following outcomes is most likely for the resulting gametes?

A) Two gametes will be n + 1 and two gametes will be n - 1.

B) The gametes will exhibit polyploidy, resulting in 2n or 3n sets.

C) All four gametes will be aneuploid with the n + 1 genotype.

D) Two gametes will be normal (n), one will be n + 1, and one will be n - 1.

Ans: A

Q2: Which of the following BEST distinguishes a triploid organism from a trisomic organism?

A) Triploidy results from structural alterations, while trisomy results from nondisjunction.

B) Triploidy is a form of aneuploidy, while trisomy is a form of euploidy.

C) Triploid organisms are common in animals, while trisomic organisms are only found in plants.

D) Triploidy involves three complete sets of chromosomes (3n), whereas trisomy involves three copies of a single chromosome (2n + 1).

Ans: D

Q3: All of the following are correctly paired with their characteristic clinical features **EXCEPT**:

A) Trisomy 13: Scalp defects and polydactyly.

B) Trisomy 21: Prominent occiput and rocker bottom feet.

C) Trisomy 18: Unusual hand position and short sternum.

D) Trisomy 21: Epicanthic folds and simian line.

Ans: B

Q4: Which of the following is the most frequent cause of Trisomy 21?

A) Nondisjunction during maternal Meiosis I.

B) Robertsonian translocations of chromosome 21.

C) Nondisjunction during paternal Meiosis II.

D) Post-zygotic mitotic errors (PZM).

Ans:A

Q5: Which chromosomal structural alteration involves the movement of a segment from one chromosome to a non-homologous chromosome?

A) Duplication

B) Inversion

C) Translocation

D) Deletion

Ans: C

Q6: Which of the following statements regarding polyploidy is TRUE?

A) Triploidy (3n) in humans results in a total of 47 chromosomes.

B) Polyploids generally appear more normal in phenotype than aneuploids.

C) A tetraploid organism has only one complete set of chromosomes.

D) Polyploidy is a common condition in most mammalian species.

Ans: B

Q7: What does a “monosomic” zygote possess following fertilisation?

A) Two chromosomes of the same type that failed to separate.

B) Only one copy of a particular chromosome.

C) A complete lack of sex chromosomes.

D) One extra chromosome in addition to the normal diploid set.

Ans: B

Q8: Which of the following is **NOT** true regarding the incidence of Down syndrome?

A) It is associated with a 15-fold increase in the risk for leukemia.

B) The male to female ratio for the syndrome is roughly 3: 2.

C) The condition affects approximately 1 out of every 700 to 800 children.

D) The risk of Down syndrome is unrelated to the age of the mother.

Ans:D

Q9: In the context of chromosomal alterations, what is the definition of an 'Euploid' state?

A) A chromosomal segment that has been reversed in its orientation.

B) A condition where an organism has an abnormal number of a specific chromosome.

C) Any chromosome number that is an exact multiple of the haploid number (n).

D) The specific state of having exactly 46 chromosomes in every cell.

Ans: C

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Q10: Which of the following is a characteristic feature of Patau syndrome (Trisomy 13) but **NOT** Edwards syndrome (Trisomy 18)?

A) Low-set, malformed ears

B) Congenital heart disease (CHD)

C) Mental retardation

D) Polydactyly

Ans: D

Q11: A student views a karyotype showing 69 chromosomes with the sex chromosomes XXY. What is the correct clinical designation for this individual?

A) Trisomy 21

B) Klinefelter syndrome

C) Tetraploidy

D) Triploidy

Ans: D

Q12: Which statement regarding maternal errors in nondisjunction is most accurate for chromosome 21?

A) Errors in maternal Meiosis I are more than three times as frequent as errors in maternal Meiosis II.

B) Paternal errors are more likely to occur in Meiosis I than in Meiosis II.

C) Maternal and paternal errors contribute equally to the prevalence of Trisomy 21.

D) Post-zygotic mitotic errors are the leading cause of chromosomal abnormalities in acrocentric chromosomes.

Ans: A

Q13: Which of the following chromosomal structures is formed when a segment of DNA is repeated?

A) Aneuploidy

B) Deletion

C) Duplication

D) Inversion

Ans: C

Q14: If an individual is diagnosed with 'Partial Trisomy 21 (21q)', what does this mean in terms of their chromosome structure?

A) They have three full copies of chromosome 21 in only some of their cells.

B) They have a deletion on the short arm of chromosome 21.

C) They have a translocation between chromosome 21 and a sex chromosome.

D) They have an extra copy of only the long arm of chromosome 21.

Ans: D

Q15: Which of the following is the BEST explanation for why aneuploidy typically results in more severe symptoms than polyploidy in humans?

A) Polyploidy is a more common condition, so the human body is better adapted to it.

B) Aneuploidy causes an imbalance in the relative concentrations of gene products from different chromosomes.

C) Polyploidy only affects plants, so it cannot cause symptoms in humans.

D) Aneuploidy involves more chromosomes than polyploidy.

Ans: B

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Lec 5

Cytogenetics

Q1: Which of the following BEST explains why a balanced Robertsonian translocation carrier typically exhibits a normal phenotype despite having only 45 chromosomes?

A) Robertsonian translocations only occur between non-homologous chromosomes, preventing any loss of genetic dosage.
B) The cell compensates for the missing chromosome by increasing the transcription rate of the remaining homologous genes.
C) The lost short arms contain redundant ribosomal RNA genes also found on other acrocentric chromosomes.
D) The translocation involves only heterochromatic regions that do not contain any functional genetic material.

Ans: C

Q2: In the context of Chronic Myelogenous Leukaemia (CML), how does the formation of the 'Philadelphia chromosome' lead to unregulated cell division?

A) The BCR -ABL fusion gene encodes a tyrosine kinase that remains constitutively active.

B) The translocation results in the deletion of a tumour suppressor gene on chromosome 9.

C) The derivative chromosome 9 inhibits the normal function of white blood cells through a dominant-negative effect.

D) The translocation moves the ABL gene to a heterochromatic region of chromosome 22, causing gene silencing.

Ans: A

Q3: All of the following are true regarding triploidy **EXCEPT**:

A) A partial hydatidiform mole is a common association when the extra set of chromosomes is paternal in origin.

B) A digynic triploid fetus is typically well-grown with an enlarged, cystic placenta.

C) The condition is responsible for approximately 20% of all spontaneous abortions.

D) Dispermy is the most frequent cause, accounting for approximately 66% of cases.

Ans: B

Q4: Which of the following karyotypes represents the most common **sex** chromosome disorder found in live-born males:

A) Trisomy 21

B) Trisomy 13

C) 47, XXY

D) 45, X

Ans: C

Q5: A conceptus is found to have a 46,XX karyotype, but all genetic material is paternal in origin. Which of the following conditions does this BEST describe?

A) Partial hydatidiform mole

B) Ovarian teratoma

C) Diandric triploidy

D) Complete hydatidiform mole

Ans: D

Q6: What is the critical difference between the origin of mosaicism and chimerism?

A) Mosaicism arises from a genetic change in a single zygote, while chimerism results from the fusion or exchange of cells between two zygotes.

B) Chimerism occurs only in females due to X-inactivation, whereas mosaicism can occur in both sexes.

C) Mosaicism involves cells from two different zygotes, whereas chimerism involves a mutation within a single zygote.

D) Mosaicism is always pathological, whereas chimerism is a normal developmental process.

Ans: A

Q7: Which statement regarding the pseudoautosomal regions (PAR) of the X and Y chromosomes is correct?

A) Recombination in these regions is suppressed to maintain male-specific traits.

B) They are the only regions where the X and Y chromosomes are homologous and can pair during meiosis.

C) They account for the significant difference in gene count between the X (900 - 1600 genes) and Y (70 - 200 genes).

D) They contain the SRY gene which determines male anatomical features.

Ans: B

Q8: During a reciprocal translocation, what determines whether the resulting derivative chromosomes will be stable during mitosis?

A) Whether the exchange occurs between homologous or non-homologous chromosomes.

B) The proximity of the break points to the SRY gene.

C) The total length of the DNA segments being swapped.

D) Whether the fragments exchanged are both acentric.

Ans: D

Q9: A child is born with a catlike cry, severe mental retardation, and usually faces early mortality. This syndrome is caused by which chromosomal event?

A) Trisomy of chromosome 18 resulting from nondisjunction.

B) A specific deletion in the short arm of chromosome 5

C) A Robertsonian translocation involving chromosome 21.

D) A reciprocal translocation between chromosomes 9 and 22.

Ans: B

Q10: Which of the following describes the phenotype of a 45,X individual?

A) Short stature, webbed neck, and rudimentary ovaries

B) Tall stature, gynecomastia, and small testes.

C) Normal female appearance but with a high risk of producing triploid offspring.

D) Macrocephaly and severe intrauterine growth retardation.

Ans: A

Q11: A carrier of a balanced Robertsonian translocation between chromosomes 14 and 21 is at risk for having a child with which condition?

A) Chronic Myelogenous Leukaemia

B) Down syndrome via trisomy 21

C) Turner syndrome

D) Cri du chat syndrome

Ans: B

Q12: The presence of two or more distinct cell lines in the same person derived from a single zygote is known as:

A) Chimerism

B) Uniparental Disomy

C) Mosaicism

D) Tetraploidy

Ans: C

Q13: Which of the following Y-chromosome regions is primarily responsible for determining male **anatomical** and **reproductive** development?

A) Pseudoautosomal region

B) AZF region

C) Sex-determining region Y (SRY)

D) Heterochromatic region of Y chromosome

Ans: C

Q14: Tetraploidy in humans typically arises through which of the following mechanisms?

A) Meiotic nondisjunction of all chromosome pairs in both parents.

B) The fusion of two separate diploid zygotes in the early blastocyst stage.

C) Double fertilization of a single ovum by two sperm cells.

D) Endomitosis, where DNA replicates without subsequent cell division in a normal zygote.

Ans: D

Q15: Which **autosomal** aneuploidy has the highest prevalence at birth?

A) Trisomy 13

B) Trisomy 21

C) Balanced Robertsonian Translocations

D) Trisomy 18

Ans: B

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Lec 6

Contrasting Mendelian and non-Mendelian Inheritance

Q1: At the biochemical level, the phenotype of enzyme activity in a Tay-Sachs disease carrier is an example of which inheritance pattern?

A) Codominance

B) Pleiotropy

C) Complete dominance

D) Incomplete dominance

Ans: D

At the biochemical level

Q2: Which of the following statements regarding dominant alleles is CORRECT?

A) Dominance relationships depend entirely on the level at which the phenotype is examined.

B) Dominant alleles are always more common in a population than recessive alleles.

C) A dominant allele physically subdues or inhibits the expression of a recessive allele.

D) Dominant alleles are only expressed when the organism is homozygous for that trait.

Ans: A

Q3: In Labrador retrievers, the gene for pigment colour (B for black, b for brown) is influenced by a second gene for colour deposition (C for colour, c for no colour). Which term BEST describes this relationship?

A) Pleiotropy

B) Incomplete dominance

C) Epistasis

D) Polygenic inheritance

Ans: C

Q4: Which of the following is **NOT** true regarding human ABO blood groups?

A) The I^A and I^B alleles show codominance when inherited together.

B) The blood group phenotypes are determined by three distinct alleles.

C) The I^A allele is incompletely dominant over the i allele.

D) An individual with blood group O must be homozygous recessive (ii)

Ans: C

Q5: What is the primary characteristic of quantitative characters′ in a population?

A) They vary along a continuum rather than being ′either-or′ traits.

B) They result from the interaction of exactly two genes with complete dominance.

C) They are always determined by a single gene with multiple alleles.

D) They are unaffected by environmental factors.

Ans: A

Q6: All of the following are true regarding Tay-Sachs disease **EXCEPT:**

A) The condition is most commonly found in populations of Ashkenazi Jewish descent.

B) It is an autosomal recessive disorder at the organismal level.

C) The disease is caused by a mutation on chromosome 21 in the HEX A gene.

D) It is caused by a buildup of non-functional lysosomes in neurons.

Ans: C

Q7: What does the norm of reaction represent in genetics?

A) The likelihood that a mutation will occur on a specific gene locus.

B) The specific ratio of phenotypes in a Mendelian cross.

C) The frequency of a dominant allele within a specific population.

D) The phenotypic range of a genotype as influenced by the environment.

Ans: D

Q8: Why are humans generally considered poor subjects for genetic research compared to organisms like pea plants?

A) The generation time is too long and the number of offspring is too few.

B) Human traits do not follow Mendelian patterns of inheritance.

C) Breeding experiments are commonly performed but are highly expensive.

D) Human phenotypes are entirely determined by the environment.

Ans: A

Q9: Which of the following BEST explains pleiotropy?

A) Multiple genes working together to produce a single trait.

B) The interaction where one gene masks the effect of another gene

C) A single gene having multiple phenotypic effects.

D) A situation where two alleles are both fully expressed in the phenotype.

Ans: C

Q10: What is the primary utility of pedigree analysis in human genetics?

A) To trace inheritance patterns and predict the probability of traits in future generations.

B) To manipulate the genetic code of future offspring.

C) To prove that Mendelian principles do not apply to complex human traits.

D) To calculate the exact soil acidity needed for specific phenotypes.

Ans: A

Q11: The phenotype of the F1 hybrid is somewhere between the phenotypes of the two parental varieties. This is known as:

A) Complete dominance

B) Epistasis

C) Multifactorial inheritance

D) Incomplete dominance

Ans: D

Q12: Which of the following is a classic example of polygenic inheritance in humans?

A) ABO Blood groups

B) Skin color

C) Cystic fibrosis

D) Tay-Sachs disease

Ans: B

Q13: At the molecular level, the alleles for Tay-Sachs disease are considered:

A) Recessive

B) Epistatic

C) Codominant

D) Incompletely dominant

Ans: C

Q14: If a trait like polydactyly is dominant, why is it not the most common phenotype in the population?

A) Dominant alleles are naturally selected against in all environments

B) Dominant alleles eventually become recessive over time.

C) The recessive allele is much more prevalent in the population's gene pool.

D) Extra digits are a polygenic trait that rarely aligns correctly.

The prevalence of an allele (how common it is) is a separate concept from whether it is dominant or recessive.

Ans: C

Q15: Which term describes characters that are influenced by both many genes and environmental factors?

A) Epistatic

B) Multifactorial

C) Pleiotropic

D) Mendelian

Ans: B

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Lec 7

Features of Autosomal Dominant and Autosomal Recessive Pedigrees & Diseases

Q1: Which of the following is **NOT true** regarding the standard construction and numbering of a pedigree?

A) Siblings are typically listed from left to right in order of birth, from oldest to youngest.

B) Generations are identified using Arabic numerals and individuals within a generation use Roman numerals.

C) The male partner in a relationship is usually positioned to the left of the female partner.

D) Each horizontal line in the diagram represents a distinct generation within the family tree.

Ans: B

Q2: All of the following are typical features of autosomal recessive inheritance **EXCEPT:**

A) Affected individuals often have at least one affected parent.

B) The gene product is typically an enzymatic protein.

C) The trait may appear suddenly in a generation after a history of consanguinity.

D) Both males and females are affected in a roughly 1:1 ratio.

Ans: A

Q3: Which feature is **MOST** characteristic of autosomal dominant inheritance patterns in a large pedigree?

A) The trait is exclusively transmitted from fathers to sons.

B) Vertical transmission, where the trait appears in every generation without skipping.

C) Horizontal transmission, where affected individuals are usually found only within a single sibship.

D) The gene product involved is almost always an enzymatic protein.

Ans: B

Q4: Which of the following **BEST** explains the genetic status of a 'compound heterozygote'?

A) The individual possesses one normal allele and one mutant allele at a specific locus.

B) The individual possesses two different mutant alleles at a specific locus.

C) The individual possesses two identical mutant alleles at a specific locus.

D) The individual possesses multiple alleles across different loci that contribute to a single phenotype.

Ans: B

Q5: Which of the following statements regarding Cystic Fibrosis (CF) is **correct**?

A) The disorder is caused by the substitution of a single amino acid in the haemoglobin protein.

B) It is a dominant disorder that affects 1 in 2, 500 people of African descent.

C) The primary defect involves abnormal chloride transport channels in plasma membranes.

D) Heterozygotes for CF typically show severe pulmonary symptoms in middle age.

Ans: C

Q6: Which of the following **BEST** describes the evolutionary implication of the sickle-cell allele in human populations?

A) The allele is rapidly being eliminated from the gene pool due to its lethal nature in homozygotes.

B) The frequency of the allele is high in all global populations due to genetic drift.

C) Heterozygotes have a survival advantage in regions where malaria is prevalent.

D) The mutation has been naturally selected to increase the oxygen-carrying capacity of red blood cells.

Ans: C

Q7: In a pedigree for an autosomal recessive condition, two heterozygous parents (Aa×Aa) have an **unaffected** child. What is the probability that this child is a carrier?

A) 3/4

B) 1/4

C) 1/2

D) 2/3

Since the child is unaffected, the aa genotype is excluded; of the remaining outcomes (AA,Aa,aA), two out of three are carriers.

Ans: D

Q8: Which of the following **BEST** distinguishes 'variable expressivity' from 'penetrance'?

A) Variable expressivity refers to the range of symptom severity, while penetrance is the proportion of individuals who show any symptoms at all.

B) Variable expressivity describes dominant traits, whereas penetrance describes recessive traits.

C) Variable expressivity is determined by the environment, while penetrance is determined solely by the genotype.

D) Penetrance refers to the age of onset, while variable expressivity refers to the specific organs affected.

Ans: A

Q9: Achondroplasia is a form of dwarfism caused by a rare dominant allele (D). Which of the following is correct regarding its inheritance?

A) If two individuals with Achondroplasia (Dd) have a child, there is a 100% chance the child will have the condition.

B) The majority of cases (80%) result from new mutations rather than inheritance from an affected parent.

C) The condition is exclusively passed through the maternal line due to mitochondrial inheritance.

D) It follows a recessive pattern where two copies of the mutant allele are required for the dwarfism phenotype.

Ans: B

Q10: Which of the following provides the **BEST** example of 'pleiotropy' in human genetics?

A) Phenylketonuria, because it is caused by the absence of a single hepatic enzyme.

B) Neurofibromatosis Type 1, because symptoms can vary significantly between different family members.

C) Marfan syndrome, where a single gene defect affects the skeleton, eyes, and cardiovascular system.

D) Familial hypercholesterolemia, because homozygotes are more severely affected than heterozygotes.

also cystic fibrosis and sickle-cell disease

Ans: C

Q11: Which statement regarding Huntington Disease is **NOT true**?

A) Affected individuals experience a loss of motor control and dementia.

B) It is associated with unstable trinucleotide repeats in the DNA.

C) It is an autosomal dominant disorder characterized by the progressive loss of brain neurons.

D) The symptoms typically manifest in early childhood, leading to a failure to reach developmental milestones.

Ans: D

Q12: Which of the following is **correct** regarding the 'Law of Independent Assortment'?

A) Alleles at different loci are transmitted to gametes independently of each other.

B) Parental characteristics always blend in the offspring to create an intermediate phenotype.

C) The two alleles at a single locus segregate into different gametes during meiosis.

D) Genes located close together on the same chromosome will always follow this law.

Ans: A

Q13: In pedigree nomenclature, how is a 'proband' typically designated?

A) By placing the symbol in brackets ([]).

B) By shading only the left half of the square or circle.

C) By using a diamond symbol instead of a square or circle.

D) With an arrow pointing toward the symbol representing the individual.

Ans: D

Q14: Which of the following is a clinical feature of Neurofibromatosis Type 1 (NF1)?

A) Tall stature with exceptionally long limbs and dislocated lenses.

B) Progressive muscular weakness and the development of early-onset cataracts.

C) Severe arteriosclerosis and the appearance of xanthomas in childhood.

D) Café-au-lait spots and Lisch nodules on the iris.

Ans: D

Q15: Which of the following **BEST** describes a 'consanguineous mating' in a pedigree?

A) A mating that results in the birth of monozygotic twins.

B) A mating between individuals of different ethnic or racial origins.

C) A relationship between two individuals who are close biological relatives.

D) A relationship that has been terminated by divorce or separation.

Ans: C

Q16: Which symbol represents a 'spontaneous abortion' (SAB) in standard pedigree nomenclature?

A) A diamond with the letter 'P' inside.

B) A small square with 'SB' written underneath.

C) A square or circle with a diagonal line through it.

D) A small triangle.

Ans: D

Q17: Which of the following is **NOT** an example of an autosomal dominant disorder?

A) Huntington Disease.

B) Marfan Syndrome.

C) Myotonic Dystrophy.

D) Tay-Sachs Disease.

Ans: D

Q18: The primary biochemical defect in Phenylketonuria (PKU) is the inability to:

A) Break down long-chain fatty acids in the brain.

B) Metabolise the amino acid phenylalanine into tyrosine

C) Transport chloride ions across the cell membrane.

D) Produce functional haemoglobin for red blood cells.

Ans: B

Q19: Which statement regarding 'dominant lethal alleles' is **correct**?

A) They are rare in the population and often arise through new mutations.

B) They are the most common cause of all known genetic disorders.

C) They are easily maintained in the population by heterozygous carriers.

D) They only cause death if the individual is homozygous for the allele.

Ans: A

Q20: In an autosomal dominant pedigree, what is the phenotypic ratio of offspring if one parent is affected (heterozygous Aa) and the other is unaffected (aa)?

A) All offspring will be unaffected but will be carriers.

B) 1 : 1 ratio of affected to unaffected.

C) 3: 1 ratio of affected to unaffected.

D) All offspring will be affected.

A Punnett square of Aa×aa yields 50% Aa (affected) and 50% aa(unaffected).

Ans: B

Q21: All of the following are consequences of the chloride transport defect in Cystic Fibrosis **EXCEPT**:

A) Reduced insulin production due to excess glucose in the blood.

B) Abnormal absorption of nutrients in the small intestine.

C) Mucus buildup in the lungs leading to bacterial infections.

D) Production of abnormally salty sweat by the skin glands.

Ans: A

Q22: In the context of autosomal dominant inheritance, how do homozygotes (AA) typically differ from heterozygotes (Aa)?

A) There is no phenotypic difference between AA and Aa in any human disorder.

B) Homozygotes are phenotypically normal, while heterozygotes express the disease.

C) Homozygotes are much more common in the population than heterozygotes.

D) Homozygotes are usually more seriously affected than heterozygotes.

Ans: D

Lec 8

X-Inactivation and Features

Sex Linked Pedigrees and Diseases

Q1: Which of the following BEST explains the necessity of dosage compensation in mammalian genetics?

A) It prevents the Y chromosome from over-expressing sex-determining genes during early embryonic development.

B) It allows autosomal traits to function normally with only a single dose of a gene in both males and females.

C) It facilitates the random activation of both X chromosomes in specific tissues to increase genetic diversity.

D) It ensures that females, who possess two X chromosomes, do not produce double the amount of X-linked gene products compared to males.

Ans: D

Q2: According to the Lyon hypothesis, which statement regarding X-inactivation is **NOT** true?

A) The inactivation occurs very early in embryonic life, specifically between 3 to 7 days after fertilisation.

B) The choice of which X chromosome (maternal or paternal) to inactivate is entirely random in each cell.

C) Once an X chromosome is inactivated in a cell, its descendants may reactivate it depending on environmental factors.

D) The inactive X chromosome physically condenses into a structure known as a Barr body.

Ans: C

Q3: A female is heterozygous for an X-linked recessive mutation causing an absence of sweat glands. Which outcome BEST describes her phenotype?

A) She will have a completely normal phenotype because the dominant 'normal' allele is expressed in every cell.

B) She will lack sweat glands entirely because the mutant X chromosome is preferentially activated in skin tissue.

C) She will have reduced sweat gland function uniformly across her entire body due to partial expression of both alleles.

D) She will be a mosaic, having some patches of skin with sweat glands and other patches without them.

Ans: D

Q4: In a pedigree for an X-linked recessive disorder, what is the defining reason for the 'absence of male-to-male transmission'?

A) A father provides his Y chromosome to his sons and his only X chromosome to his daughters.

B) Affected males are typically infertile and cannot produce male offspring.

C) X-linked recessive alleles are lethal in the hemizygous state during male foetal development.

D) The X chromosome in males is hemizygous and therefore cannot be replicated during spermatogenesis.

Ans: A

Q5: Which of the following genes would be most likely to 'escape' X-inactivation in a human female?

A) Any gene located outside the pseudoautosomal region that lacks a related copy on the Y chromosome.

B) The SRY gene, which determines male sex characteristics.

C) The gene responsible for Duchenne Muscular Dystrophy located on the short arm of the X chromosome.

D) A gene located in the pseudoautosomal region with a matching counterpart on the Y chromosome.

Ans: D

Q6: Which statement BEST describes the clinical difference between Duchenne and Becker Muscular Dystrophies?

A) Becker Muscular Dystrophy only affects females, while Duchenne only affects males.

B) They represent allelic heterogeneity, where different mutations in the same gene lead to varying severities of the disease.

C) Duchenne is caused by a chromosomal deletion, while Becker is caused by X-inactivation failure.

D) Duchenne is an X-linked recessive trait, whereas Becker is an X-linked dominant trait.

Ans: B

Q7: All of the following are features of X-linked dominant inheritance **EXCEPT:**

A) There is a complete absence of male-to-male transmission.

B) Affected females often show more mild and variable expression than affected males.

C) The disorder is typically found twice as frequently in females as in males.

D) Affected males transmit the disorder to approximately 50% of their daughters.

Ans: D

Q8: A woman who is a carrier for Haemophilia A (X^NX^n) marries a normal male (X^NY). What is the probability that their first child will be an affected son?

A) 100%

B) 0%

C) 25%

D) 50%

Ans: C

Q9: Which of the following BEST distinguishes an X-linked dominant trait from an autosomal dominant trait in a pedigree?

A) The presence of affected individuals in every generation.

B) The higher incidence of the trait in males than in females.

C) The offspring of an affected male; in X-linked dominant, all daughters are affected but no sons are affected.

D) The fact that affected females can have affected sons.

Ans: C

اللهم إنك عفو كريم تُحب العفو فاعف عنا.

Q10: What is the clinical significance of Vitamin D Resistant Rickets as an example of X-linked inheritance?

A) It is an X-linked recessive disorder where carrier females are completely asymptomatic.

B) It is an X-linked dominant disorder that manifests with short stature and low serum phosphate.

C) It is an autosomal trait that mimics X-linkage due to gender-specific hormone levels.

D) It is a Y-linked disorder, meaning it is only ever passed from fathers to sons.

Ans: B

Q11: A male with an X-linked recessive condition is described as 'hemizygous'. What does this term imply?

A) He has two X chromosomes, but one has been completely deleted during embryonic development.

B) He has only one copy of the X chromosome, so a single recessive allele will determine his phenotype.

C) The recessive allele on his X chromosome has mutated to become dominant.

D) His Y chromosome contains a 'backup' copy of the gene that is currently inactive.

Ans: B

Q12: Regarding X-linked recessive inheritance, what does the term 'diagonal inheritance' refer to?

A) The observation that symptoms of the disease worsen as it is passed down through generations.

B) The process where an X-linked dominant allele is passed to all children regardless of sex.

C) The transmission of the trait from an affected male to his grandsons through his carrier daughters.

D) The physical movement of the X chromosome during meiosis I.

Ans: C

Q13: If a male with an X-linked recessive disorder has children with a woman who is not a carrier, which statement is true regarding their offspring?

A) All of their daughters will be obligate carriers of the disorder.

B) 50% of their daughters will be affected by the disorder.

C) All of their sons will be affected by the disorder.

D) The disorder will 'skip' this generation and no offspring will carry the gene.

Ans: A

Q14: Which of the following is a primary role of the Y chromosome?

A) It undergoes random inactivation in males to balance the gene dosage with females.

B) It contains the primary genes for cognitive development and red-green vision.

C) It primarily encodes genes related to sex determination, such as the SRY factor.

D) It serves as a backup for most metabolic genes found on the X chromosome.

Ans: C

Q15: Why might a female carrier of an X-linked recessive disorder, like Haemophilia, occasionally show mild symptoms?

A) Females naturally have lower levels of clotting factors regardless of their genotype.

B) Due to X-inactivation, she may have a higher proportion of cells where the normal X is inactivated.

C) The mutant allele 'escapes' inactivation and suppresses the normal allele on the other X chromosome.

D) The recessive allele becomes dominant when the individual is under high physiological stress.

Ans: B

الحمد لله حمداً دائمًا لا يحيط به عدد، ولا يقطعه أمد

Lec 9

Pedigrees Practice

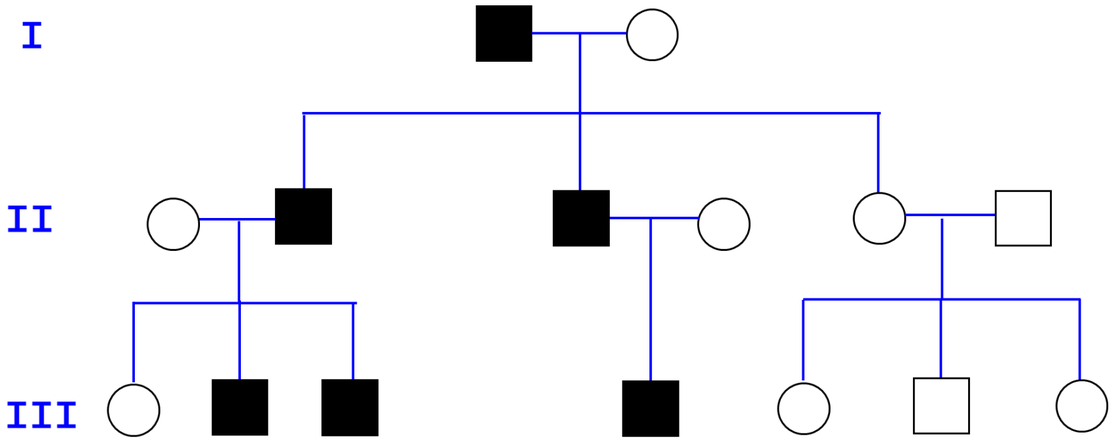
Q1: In the following pedigree, identify the mode of inheritance.

A) Autosomal recessive

B) X-linked dominant

C) X-linked recessive

D) Y-linked



Ans: D

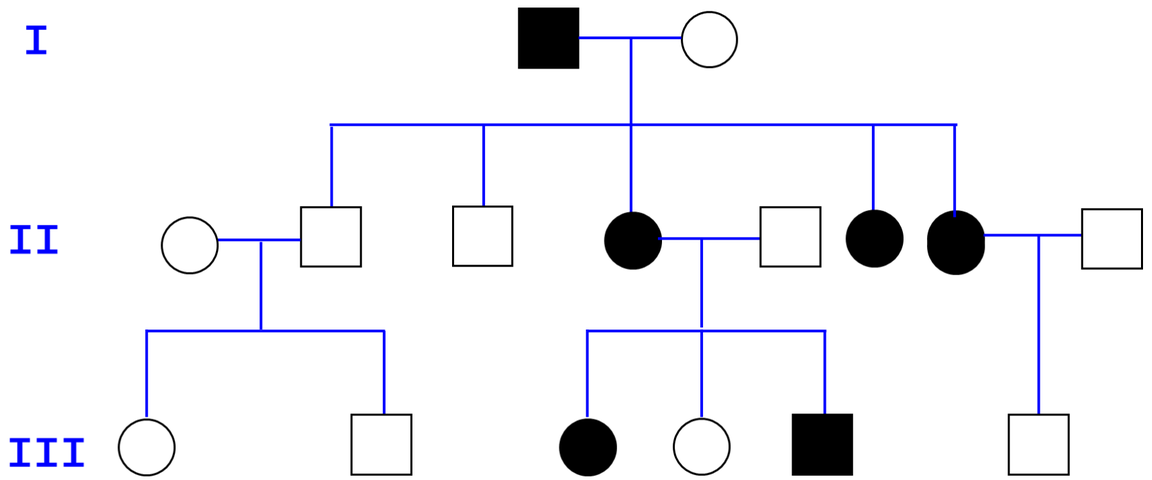
Q2: In the following pedigree, identify the mode of inheritance.

A) Autosomal recessive

B) X-linked dominant

C) X-linked recessive

D) Autosomal dominant



Ans: B

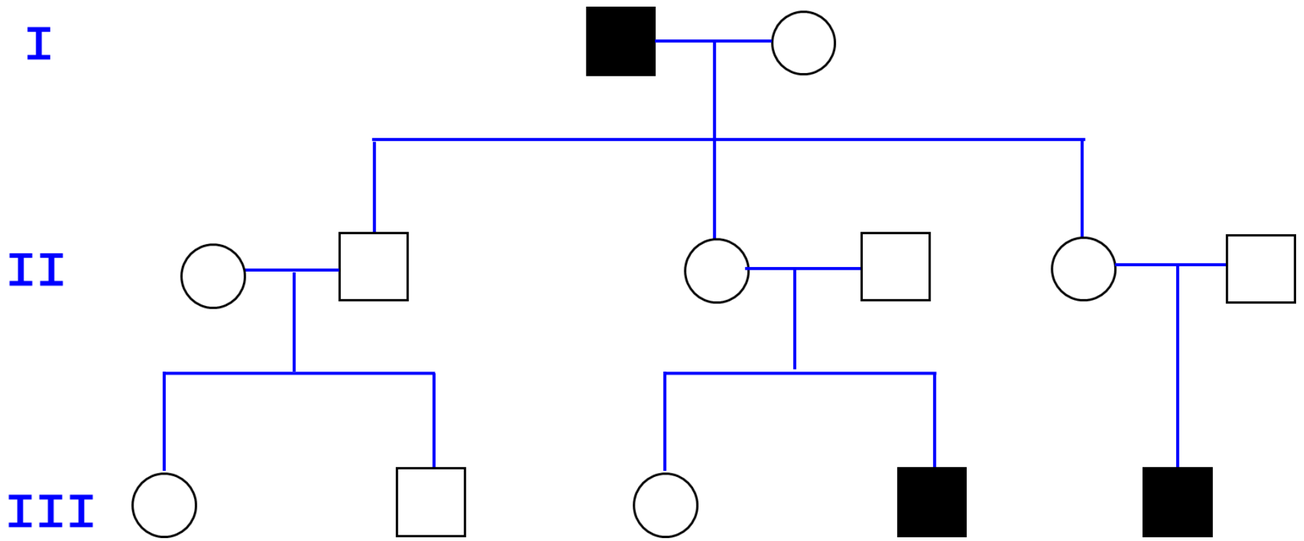
Q3: In the following pedigree, identify the mode of inheritance.

A) Autosomal recessive

B) X-linked dominant

C) X-linked recessive

D) Autosomal dominant



Ans: C

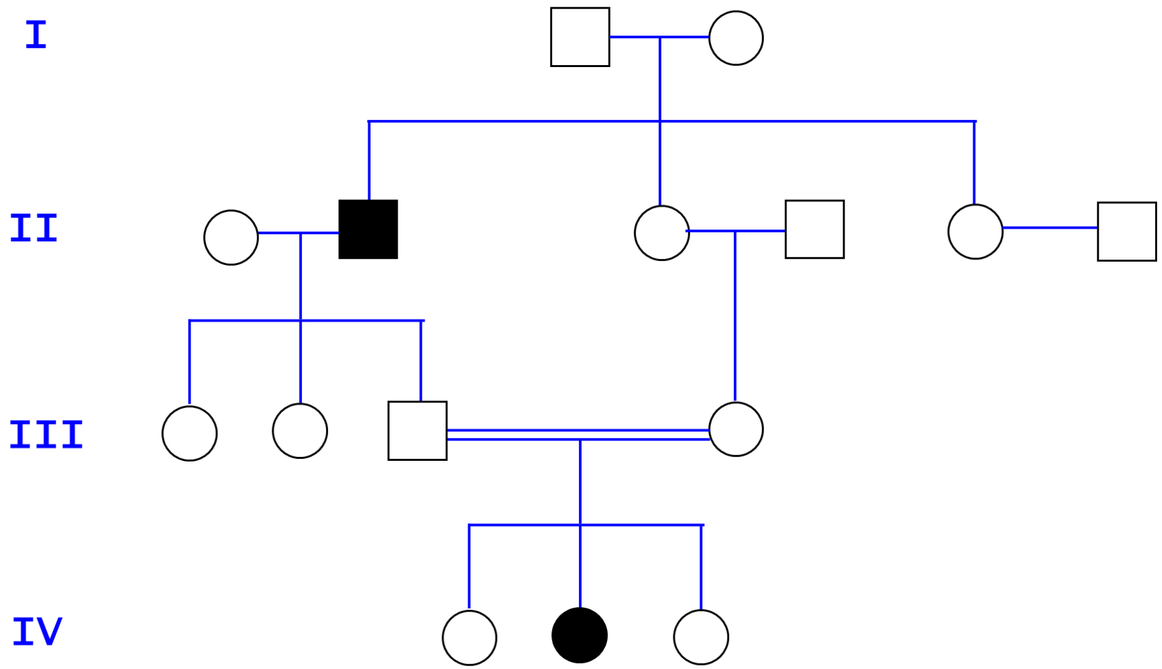
Q4: In the following pedigree, identify the mode of inheritance.

A) Autosomal recessive

B) X-linked dominant

C) X-linked recessive

D) Autosomal dominant



Ans: A

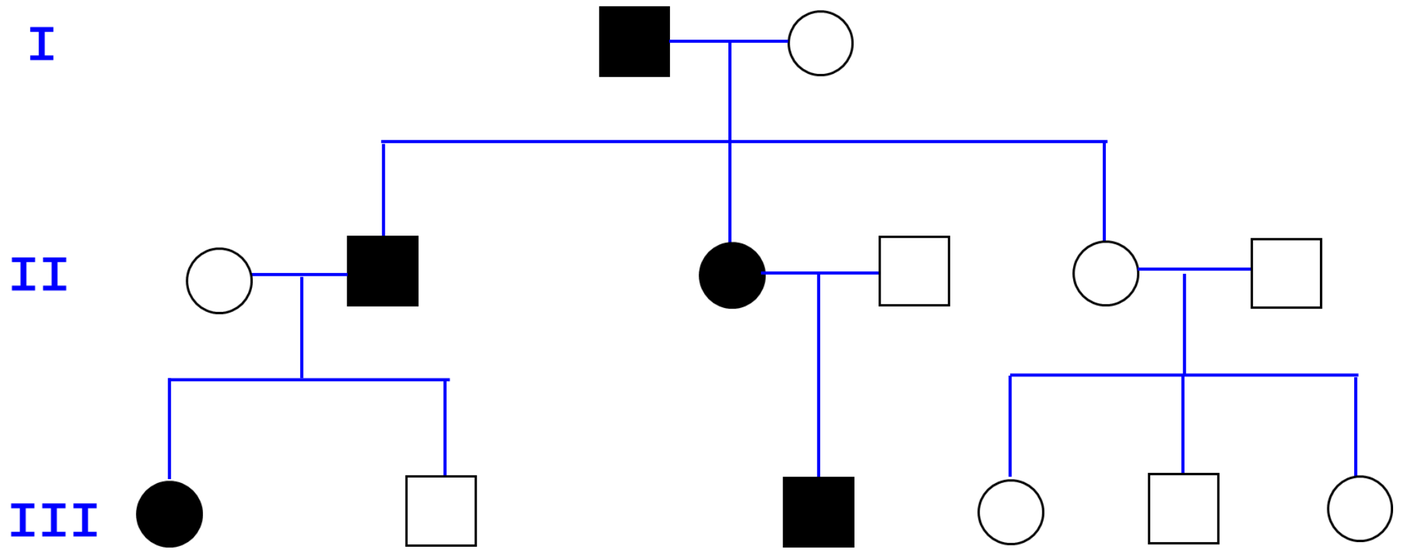
Q5: In the following pedigree, identify the mode of inheritance.

A) Autosomal recessive

B) X-linked dominant

C) X-linked recessive

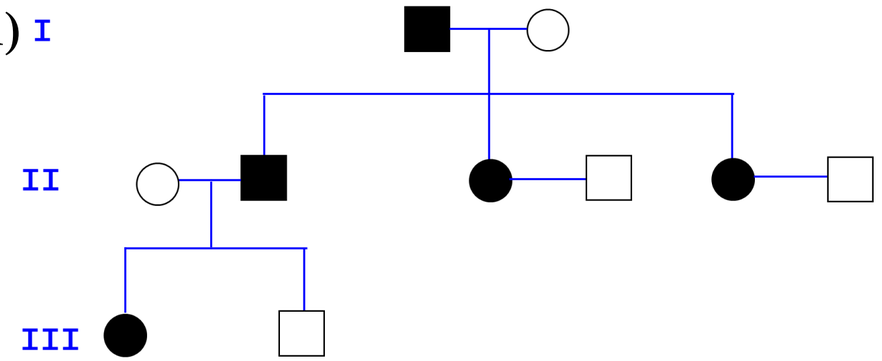
D) Autosomal dominant



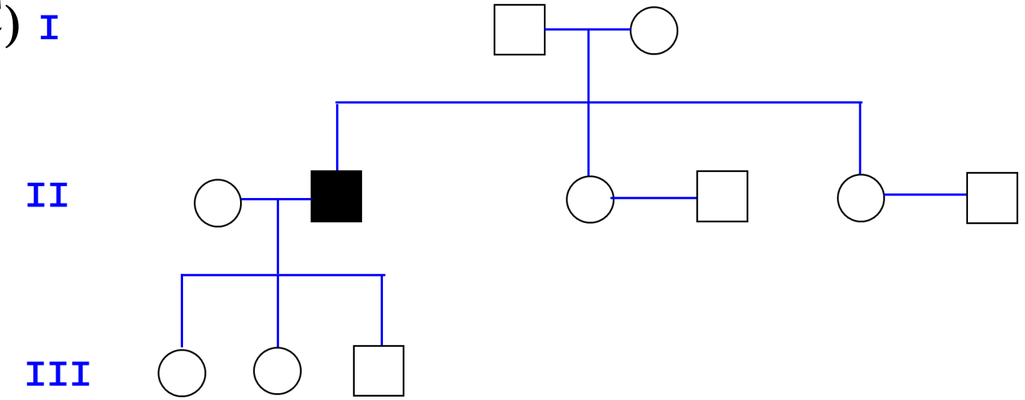
Ans: D

Q6: Determine which of the following pedigrees exhibits autosomal recessive inheritance for a particular trait.

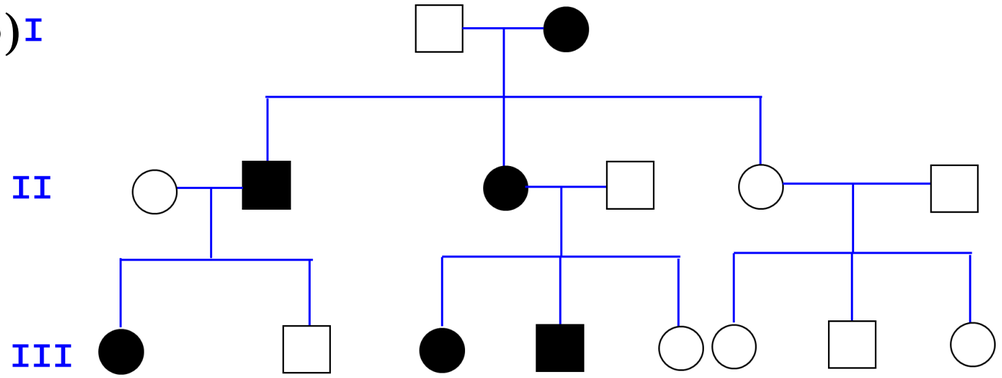
A)



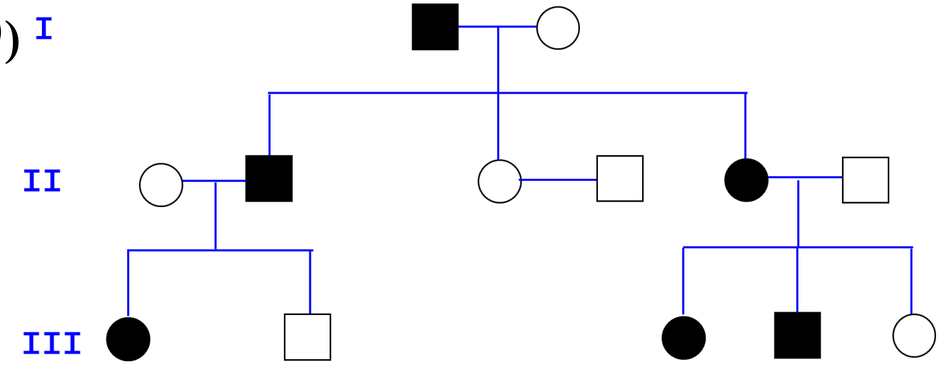
C)



B)



D)



Ans: C

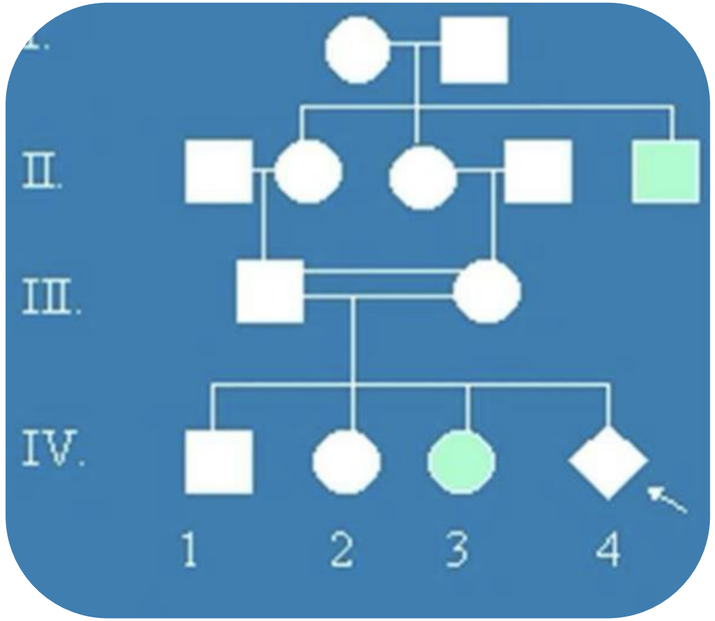
Q7: Which of the following BEST explains why the mode of inheritance is classified as autosomal recessive rather than X-linked recessive?

A) The trait appears to skip generations, appearing in generation IV but not in Ill.

B) The affected individual IV-3 is the result of a consanguineous marriage.

C) There are more unaffected individuals than affected individuals in the pedigree.

D) The presence of an affected female (IV-3) with an unaffected father (III-1).



Ans: D

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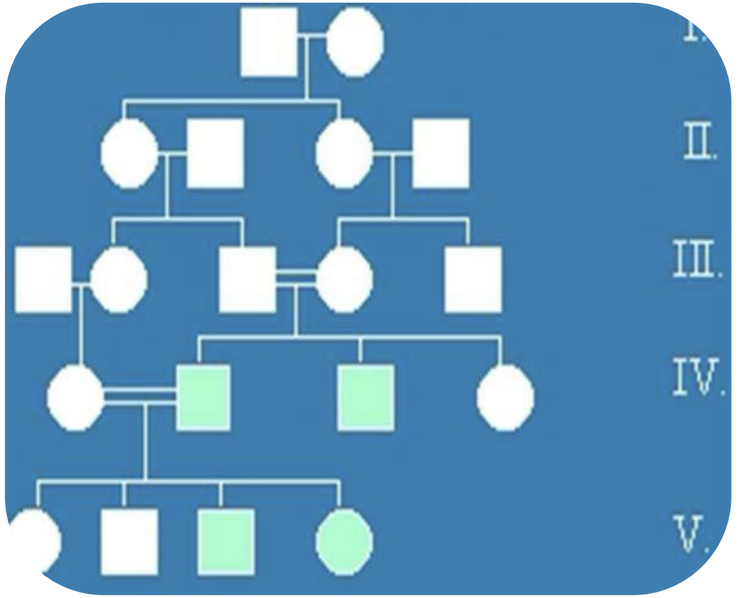
Q8: This pedigree describes 'apparent male to male transmission' in an X-linked recessive trait. Which of the following BEST explains how this occurs?

A) The trait is actually autosomal but mimics X-linked inheritance.

B) The father (IV-2) passed his affected X chromosome directly to his son.

C) The mother (IV-3) is a carrier who passed the affected X to her son (V-3).

D) The trait underwent a spontaneous mutation in generation V.



Ans: C

رسالة من الفريق العلمي

اللهم إن عمر عطية في ذمتك وحبل جوارك، فقِه من فتنة القبر وعذاب النار،
أنت أهل الوفاء والحق، فاغفر له وارحمه إنك أنت الغفور الرحيم.

★ينصح به بشدّة★

نسأل الله أن يبارك في أوقاتنا وأعمالنا، وأن يزيدنا علمًا ونورًا، وأن يجعل ما نقدّمه في ميزان حسناتنا و أن يرزقنا جميعًا الإخلاص والتوفيق، وأن يكرمنا بحفظ كتابه الكريم وتدبّره والعمل به آناء الليل وأطراف النهار.

لا تنسونا من صالح دعائكم

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