تـــاج الـــذكـــر

لَا إِلَهَ إِلَّا اللَّهُ، وَحْدَهُ لَا شَرِيكَ لَهُ، لَهُ المُلْكُ
وَلَهُ الحَمْدُ، وَهُوَ عَلَى كُلِّ شَيْءٍ قَدِيرٌ

DOPAMINE
SCIENTIFIC TEAM

Genetics Test Bank

Genetics-3 (20 MCQs)

Q1. Penetrance refers to:
A) Severity of disease
B) All-or-none expression of a genotype
C) Age of onset
D) Multiple organ involvement
Correct Answer: B

Q2. A condition expressed in less than 100% of individuals carrying the allele shows:
A) Pleiotropy
B) Variable expressivity
C) Reduced penetrance
D) Imprinting
Correct Answer: C

Q3. Retinoblastoma mutant allele is approximately:
A) 50% penetrant
B) 70% penetrant
C) 90% penetrant
D) 100% penetrant
Correct Answer: C

Q4. Variable expressivity refers to:
A) Presence or absence only
B) Different severity among affected individuals
C) Parent-of-origin effect
D) Mitochondrial inheritance
Correct Answer: B

Q5. A classic example of variable expressivity is:
A) Neurofibromatosis
B) Fragile X
C) PWS
D) Angelman syndrome
Correct Answer: A

Q6. Variable age of onset is illustrated by:
A) Huntington disease
B) Waardenburg syndrome
C) PWS
D) Hemochromatosis
Correct Answer: A

Q7. Marfan syndrome is an example of:
A) Imprinting
B) Pleiotropy
C) Sex limitation
D) UPD
Correct Answer: B

Q8. Allelic heterogeneity means:
A) Different genes causing same phenotype

B) Same mutation in all patients
C) Different mutant alleles at same locus
D) Mitochondrial inheritance
Correct Answer: C

Q9. Locus heterogeneity means:
A) Different genes cause similar phenotype
B) One gene causes many phenotypes
C) Different severities
D) Reduced penetrance
Correct Answer: A

Q10. Congenital hearing loss is an example of:
A) Allelic heterogeneity
B) Locus heterogeneity
C) Imprinting
D) Anticipation
Correct Answer: B

Q11. Sex-limited traits are:
A) X-linked only
B) Expressed only in one sex despite autosomal inheritance
C) Mitochondrial
D) Imprinted
Correct Answer: B

Q12. Hemochromatosis is an example of:
A) Sex-linked
B) Sex-limited
C) Sex-influenced
D) Imprinted
Correct Answer: C

Q13. Anticipation is commonly associated with:
A) Trinucleotide repeat expansion
B) Aneuploidy
C) Deletion only
D) Mitochondrial mutations
Correct Answer: A

Q14. Fragile X syndrome is caused by expansion of:
A) CTG
B) CAG
C) CGG
D) GAA
Correct Answer: C

Q15. A full mutation in Fragile X is:
A) 5–54 repeats
B) 55–200 repeats
C) >200 repeats
D) >20 repeats

Correct Answer: C

Q16. Fragile X anticipation typically occurs through:
A) Paternal transmission
B) Maternal transmission
C) Mitochondria
D) Y chromosome
Correct Answer: B

Q17. Huntington disease is characterized by:
A) Congenital onset
B) Late onset neurodegeneration
C) Deafness
D) Obesity
Correct Answer: B

Q18. Genomic imprinting depends on:
A) DNA sequence change
B) Parent of origin
C) Mitochondrial DNA
D) Gene amplification
Correct Answer: B

Q19. Prader-Willi syndrome may result from:
A) Paternal deletion 15q11-13
B) Maternal deletion 15q11-13
C) Trisomy 21
D) BCR-ABL
Correct Answer: A

Q20. Mitochondrial genes are inherited:
A) Paternally
B) Biparentally
C) Maternally
D) Through Y chromosome
Correct Answer: C

Genetics-4 (20 MCQs)

Q1. Neoplasia is characterized by:
A) Infection
B) Uncontrolled cellular proliferation
C) Autoimmunity
D) Fibrosis
Correct Answer: B

Q2. A malignant neoplasm can:
A) Only enlarge
B) Invade and metastasize
C) Never spread
D) Only affect blood
Correct Answer: B

Q3. The most common cancer class is:
A) Sarcoma
B) Leukemia
C) Carcinoma
D) Lymphoma
Correct Answer: C

Q4. Passenger mutations are:
A) Major cancer-causing mutations
B) Random mutations not driving cancer
C) Chromosomal translocations
D) Germline mutations
Correct Answer: B

Q5. Driver genes are identified because they are:
A) Rarely mutated
B) Frequently mutated in cancers
C) Only inherited
D) Mitochondrial
Correct Answer: B

Q6. TP53 is an example of a:
A) Passenger gene
B) Tumor suppressor gene
C) Growth factor
D) Telomere
Correct Answer: B

Q7. Oncogenomics can improve:
A) Diagnosis only
B) Prognosis only
C) Diagnosis, prognosis, and treatment
D) Histology only
Correct Answer: C

Q8. Failure of DNA repair may lead to:
A) Fewer mutations

B) Cellular transformation
C) Apoptosis only
D) Normalization
Correct Answer: B

Q9. Oncogenesis is described as a:
A) Single-step process
B) Multistep process
C) Infectious process
D) Metabolic process
Correct Answer: B

Q10. The Philadelphia chromosome is:
A) t(8;14)
B) t(14;18)
C) t(9;22)
D) t(11;22)
Correct Answer: C

Q11. BCR-ABL causes increased:
A) DNA repair
B) Tyrosine kinase activity
C) Apoptosis
D) Contact inhibition
Correct Answer: B

Q12. Gene amplification can increase:
A) Gene expression
B) DNA repair
C) Chromosome loss
D) Imprinting
Correct Answer: A

Q13. MYC amplification contributes to cancer through:
A) Reduced expression
B) Overexpression
C) Deletion
D) Methylation
Correct Answer: B

Q14. Active telomerase in cancer cells leads to:
A) Telomere shortening
B) Cellular immortality
C) Cell death
D) Differentiation
Correct Answer: B

Q15. Proto-oncogenes normally:
A) Promote growth and survival
B) Cause apoptosis only
C) Silence genes
D) Repair DNA exclusively

Correct Answer: A

Q16. Activation of a proto-oncogene generally requires:
A) Mutation of one allele
B) Mutation of both alleles
C) Mitochondrial inheritance
D) Imprinting
Correct Answer: A

Q17. RAS oncogene becomes:
A) Inactive
B) Constitutively active
C) Deleted
D) Silenced
Correct Answer: B

Q18. Tumor suppressor genes usually require:
A) One mutated allele
B) Two mutated alleles
C) No mutation
D) Gene amplification
Correct Answer: B

Q19. Loss of TP53 function allows:
A) DNA repair before division
B) Continued division despite DNA damage
C) Immediate apoptosis
D) Normal cell cycle arrest
Correct Answer: B

Q20. Tumor heterogeneity results because:
A) All cells acquire mutations simultaneously
B) Different lineages accumulate different mutations
C) Tumors are genetically identical
D) Metastases never evolve
Correct Answer: B

Genetics 5 & 6 Test Bank (25 MCQs)

Q1. Which inheritance pattern is typical of hereditary cancer syndromes?
A. Autosomal recessive
B. Multifactorial only
C. Mendelian with high penetrance mutant gene
D. Mitochondrial
Correct Answer: C

Q2. MEN2A is characterized by a high incidence of:
A. Basal cell carcinoma
B. Medullary thyroid carcinoma
C. Retinoblastoma
D. Colon polyps
Correct Answer: B

Q3. The gene mutated in MEN2 is:
A. RB1
B. APC
C. RET
D. MLH1
Correct Answer: C

Q4. RET encodes a receptor with which intracellular activity?
A. DNA polymerase
B. Tyrosine kinase
C. Ligase
D. Topoisomerase
Correct Answer: B

Q5. MEN2A mutations in RET typically cause:
A. Loss of function
B. Constitutive activation
C. Deletion of gene
D. Imprinting
Correct Answer: B

Q6. Hirschsprung disease is usually associated with RET:
A. Gain-of-function mutations
B. Loss-of-function mutations
C. Amplifications only
D. Duplications only
Correct Answer: B

Q7. According to the two-hit hypothesis, tumor suppressor genes usually require:
A. One mutated allele
B. Three mutations
C. Both alleles inactivated
D. Gene amplification
Correct Answer: C

Q8. RB1 was the first identified:
A. Oncogene

B. Mismatch repair gene
C. Tumor suppressor gene
D. Imprinting gene
Correct Answer: C

Q9. Familial retinoblastoma often presents as:
A. Unilateral single tumor
B. Bilateral or multifocal tumors
C. Adult-onset disease
D. Colon cancer
Correct Answer: B

Q10. The penetrance of inherited retinoblastoma is:
A. 10%
B. 50%
C. >90%
D. 100%
Correct Answer: C

Q11. RB1 normally regulates transition from:
A. S to G2
B. G2 to M
C. G1 to S
D. M to G1
Correct Answer: C

Q12. RB1 inhibits which transcription factor?
A. MYC
B. SMAD
C. E2F
D. RAS
Correct Answer: C

Q13. Loss of heterozygosity is a common mechanism for:
A. Oncogene activation
B. Second hit in TSGs
C. Imprinting
D. Translation
Correct Answer: B

Q14. Lifetime breast cancer risk in BRCA1/2 carriers is approximately:
A. 1–5%
B. 10–20%
C. 46–87%
D. 100%
Correct Answer: C

Q15. BRCA1 and BRCA2 proteins primarily function in:
A. Mismatch repair
B. Double-stranded DNA break repair
C. Translation
D. Splicing

Correct Answer: B

Q16. A common second hit in BRCA-associated tumors is:
A. LOH
B. Mitochondrial mutation
C. Triploidy
D. Imprinting
Correct Answer: A

Q17. FAP is caused by mutations in:
A. RET
B. APC
C. RB1
D. TGFBR2
Correct Answer: B

Q18. Gardner syndrome is best described as:
A. Variant of Lynch syndrome
B. Variant of FAP
C. Variant of MEN2
D. Sporadic cancer
Correct Answer: B

Q19. Lynch syndrome most commonly involves mutations in:
A. BRCA1/2
B. RB1
C. MLH1 and MSH2
D. RET
Correct Answer: C

Q20. A hallmark of Lynch syndrome is:
A. Microsatellite instability
B. Trisomy
C. Anticipation
D. Mosaicism
Correct Answer: A

Q21. Mismatch repair genes normally repair:
A. Double-strand breaks
B. Incorrect DNA base pairing
C. Protein damage
D. RNA errors
Correct Answer: B

Q22. Female heterozygotes with Lynch syndrome have about what risk of endometrial cancer?
A. 5%
B. 20%
C. 40%
D. 90%
Correct Answer: C

Q23. TGFBR2 encodes a:
A. Transcription factor
B. Serine-threonine kinase receptor
C. DNA helicase
D. Ligase
Correct Answer: B

Q24. Loss of TGF-β signaling leads to:
A. MYC suppression
B. Increased cell-cycle inhibition
C. Loss of growth inhibition
D. DNA repair enhancement
Correct Answer: C

Q25. In FAP, colectomy is performed to:
A. Treat thyroid cancer
B. Prevent malignant transformation
C. Repair DNA
D. Reduce MSI
Correct Answer: B

Genetic Variation 1 (20 MCQs)

Q1. According to ACMG 2015, the preferred term replacing mutation and polymorphism is:
A. Variant
B. Allele
C. Genotype
D. Haplotype
Correct Answer: A

Q2. A polymorphism is traditionally defined as a variant frequency:
A. <1%
B. =1%
C. >1%
D. >50%
Correct Answer: C

Q3. Genome mutations primarily affect:
A. Single genes
B. Chromosome number
C. RNA splicing
D. Protein folding
Correct Answer: B

Q4. Trisomy 21 results from:
A. Gene deletion
B. Chromosome missegregation
C. Splicing error
D. Frameshift
Correct Answer: B

Q5. Genome mutations produce:
A. Aneuploidy
B. Missense variants
C. NMD
D. Synonymous variants
Correct Answer: A

Q6. Chromosome mutations alter:
A. Part of a chromosome
B. Only mitochondrial DNA
C. Only RNA
D. Whole genome copy number only
Correct Answer: A

Q7. An example of a chromosome mutation is:
A. Translocation
B. Point mutation
C. Depurination
D. Deamination
Correct Answer: A

Q8. Gene mutations may affect:
A. Nuclear or mitochondrial genomes

B. Only nuclear DNA
C. Only proteins
D. Only RNA
Correct Answer: A

Q9. Gene mutations can arise from:
A. Replication errors
B. Repair failure
C. Mutagens
D. All of the above
Correct Answer: D

Q10. Mutagens are:
A. DNA repair enzymes
B. Agents that enhance mutation frequency
C. Chromosomes
D. snRNPs
Correct Answer: B

Q11. Replication introduces an incorrect nucleotide about once every:
A. 10^{2} bp
B. 10■ bp
C. 10^{1}■ bp
D. 10^{12} bp
Correct Answer: C

Q12. Proofreading corrects more than:
A. 50%
B. 75%
C. 90%
D. 99.9%
Correct Answer: D

Q13. The human diploid genome contains approximately:
A. 6×10■ bp
B. 6×10■ bp
C. 3×10^{3} bp
D. 10^{12} bp
Correct Answer: A

Q14. Replication errors introduce less than how many new base-pair mutations per cell division?
A. 1
B. 10
C. 100
D. 1000
Correct Answer: A

Q15. Per day, a human cell experiences DNA damage in the range of:
A. 10–100
B. 100–1000
C. 10,000–1,000,000
D. 1–10

Correct Answer: C

Q16. A cause of spontaneous DNA damage is:
A. Depurination
B. Demethylation
C. Deamination
D. All of the above
Correct Answer: D

Q17. DNA damage may be caused by exposure to:
A. UV radiation
B. Ionizing radiation
C. Chemical mutagens
D. All of the above
Correct Answer: D

Q18. Permanent mutations often arise during:
A. DNA damage repair
B. Normal transcription
C. Translation
D. Chromosome pairing
Correct Answer: A

Q19. The HGVS prefix for coding DNA reference sequence is:
A. g.
B. c.
C. m.
D. p.
Correct Answer: B

Q20. The HGVS prefix for mitochondrial reference sequence is:
A. n.
B. o.
C. m.
D. r.
Correct Answer: C

Genetic Variation 2 (20 MCQs)

Q1. A synonymous mutation may alter:
A. Pre-mRNA splicing
B. mRNA structure
C. Translation
D. All of the above
Correct Answer: D

Q2. Gain of an exonic splicing enhancer motif can enhance binding of:
A. U5
B. SRSF1
C. E2F
D. RB1
Correct Answer: B

Q3. Deactivation of an exonic splicing silencer motif abolishes binding of:
A. hnRNP regulators
B. SMAD proteins
C. RET
D. APC
Correct Answer: A

Q4. A synonymous variant may alter:
A. Translation elongation rate
B. Ribosomal pause rhythm
C. Protein fate
D. All of the above
Correct Answer: D

Q5. Nonsense-mediated mRNA decay is associated with premature stop codons located approximately:
A. 5 nt upstream
B. 20 nt upstream
C. 50–55 nt upstream
D. 100 nt downstream
Correct Answer: C

Q6. RNA splicing removes:
A. Exons
B. Introns
C. Promoters
D. Enhancers
Correct Answer: B

Q7. The complex responsible for RNA splicing is the:
A. Ribosome
B. Proteasome
C. Spliceosome
D. Centrosome
Correct Answer: C

Q8. Splice donor and splice acceptor sites are located at:
A. Exon-intron boundaries
B. Promoters
C. Telomeres
D. Centromeres
Correct Answer: A

Q9. The branch site is typically found:
A. Within exon 1
B. Less than 20 nt upstream of splice acceptor
C. At promoter
D. At 5' cap
Correct Answer: B

Q10. Most introns contain how many functionally important conserved regions?
A. 1
B. 2
C. 3
D. 4
Correct Answer: C

Q11. The first nucleophilic attack in splicing occurs on the:
A. 5′ GU dinucleotide
B. Poly-A tail
C. Start codon
D. Stop codon
Correct Answer: A

Q12. The conserved branch-site nucleotide involved in splicing is:
A. G
B. C
C. A
D. T
Correct Answer: C

Q13. The initial splicing reaction forms a:
A. Hairpin
B. Lariat
C. Plasmid
D. Chromatid
Correct Answer: B

Q14. U1 snRNP binds the:
A. Branch site
B. Splice donor site
C. Poly-A signal
D. Start codon
Correct Answer: B

Q15. U2 snRNP binds the:
A. Branch site
B. Stop codon
C. Promoter

D. Exon
Correct Answer: A

Q16. Which snRNP binds simultaneously to donor and acceptor sites?
A. U1
B. U2
C. U5
D. U6
Correct Answer: C

Q17. Interaction between donor and acceptor sites is stabilized by:
A. U4/U5/U6-containing particle
B. RB1
C. E2F
D. RET
Correct Answer: A

Q18. After intron release, which sequences are joined together?
A. Introns
B. E1 and E2 exons
C. Promoters
D. Enhancers
Correct Answer: B

Q19. Microsatellite instability is a feature of:
A. Splicing
B. Mismatch repair defects
C. Translation
D. Replication origin activation
Correct Answer: B

Q20. The spliceosome contains proteins and:
A. snRNAs
B. tRNAs
C. rRNAs only
D. miRNAs only
Correct Answer: A

06 Genetic Testing – MCQ Test Bank (20 Questions)

Q1. Which genetic test provides the original whole-genome view at light-microscope resolution?
A. Karyotype
B. FISH
C. CMA
D. NGS
Correct Answer: A

Q2. The approximate resolution of a karyotype is:
A. 1 bp
B. 100 kb
C. 5–10 Mb
D. 10–50 kb
Correct Answer: C

Q3. Colchicine is used during karyotyping to:
A. Label DNA
B. Arrest cells in metaphase
C. Amplify DNA
D. Detect CNVs
Correct Answer: B

Q4. Which abnormality is uniquely detected by routine karyotyping?
A. Point mutations
B. Balanced translocations
C. Small indels
D. Methylation defects
Correct Answer: B

Q5. Mosaicism is usually detectable by karyotype when present in at least:
A. 1–5% of cells
B. 5–10% of cells
C. 10–20% of cells
D. 50% of cells
Correct Answer: C

Q6. A major advantage of FISH is that it can be performed on:
A. Only dividing cells
B. Only bone marrow
C. Interphase nuclei
D. Only cultured lymphocytes
Correct Answer: C

Q7. Typical turnaround time for FISH is:
A. 1–3 weeks
B. 24–48 hours
C. 5–14 days
D. 1 month
Correct Answer: B

Q8. Which type of FISH probe is used for rapid aneuploidy detection?
A. Whole chromosome paint
B. Telomeric probe
C. Centromeric probe
D. LSI probe
Correct Answer: C

Q9. The first-line test for unexplained ID/DD/ASD/MCA is:
A. Karyotype
B. FISH
C. Chromosomal microarray
D. Sanger sequencing
Correct Answer: C

Q10. Diagnostic yield of CMA in unexplained ID/DD/ASD/MCA is approximately:
A. 3%
B. 5%
C. 15–20%
D. 50–60%
Correct Answer: C

Q11. Which CMA platform can detect loss of heterozygosity and UPD?
A. Array-CGH
B. SNP array
C. FISH
D. Karyotype
Correct Answer: B

Q12. A CNV classified as VUS means:
A. Definitely pathogenic
B. Definitely benign
C. Insufficient/conflicting evidence
D. Clinically actionable
Correct Answer: C

Q13. Which abnormality cannot be detected by CMA?
A. Copy-number variants
B. Balanced translocations
C. Deletions
D. Duplications
Correct Answer: B

Q14. Sanger sequencing remains the gold standard for:
A. Whole genome analysis
B. Variant confirmation
C. CNV detection
D. Mosaicism screening
Correct Answer: B

Q15. Average read depth recommended for germline NGS is at least:
A. 10×
B. 20×
C. 30×

D. 200×
Correct Answer: C

Q16. Whole-exome sequencing analyzes approximately:
A. Entire genome
B. 1–2% of the genome
C. Less than 0.1% of genome
D. Only mitochondrial DNA
Correct Answer: B

Q17. Which variant type is considered the 'bread and butter' of clinical NGS?
A. SNVs
B. Triploidy
C. Balanced inversions
D. Methylation defects
Correct Answer: A

Q18. According to ACMG/AMP classification, a VUS should:
A. Guide treatment decisions
B. Be treated as pathogenic
C. Not be used for clinical decisions
D. Always prompt surgery
Correct Answer: C

Q19. ACMG actionable secondary findings include genes associated with:
A. Hereditary cancer
B. Cardiomyopathies
C. Arrhythmias
D. All of the above
Correct Answer: D

Q20. For recurrent miscarriage with suspected balanced translocation, the preferred test is:
A. CMA
B. NGS panel
C. Karyotype ± FISH
D. WES
Correct Answer: C

سيد الاستغفار :

" اللـــهم أنـــت ربـــي لا إله إلا أنـــت
خـلقتني وأنا عبدك وأنا على عـهدك
ووعـــــدك مـــــا استـــــطعت
أعـــــوذ بـــــك مـــــن شـــــر
ما صنعت أبوء لك بنعمتك علي وأبوء
بذنبي فـــــاغفر لي فـــــإنه
لا يغـــــفر الذنـــــوب إلا أنـــــت "