

## Designer Genes **KEY**

### SSSS 19 - 20

Written By neuro

1. (A) Must be correctly constructed, (B) , (C) , (D) 12.5%, (E) 25%
2. (A) Down Syndrome, (B) Klinefelter's syndrome, (C) Jacob's Syndrome, (D) Turner Syndrome, (E) Edward's Syndrome, (F) Patau Syndrome
3. (A) Mitochondrial Disorder, (B) 100%
4. (A) Sex linked Recessive, (B) Autosomal Recessive, (C) Sex linked Recessive, (D) Autosomal dominant
5. (A) Autosomal Recessive, (B) X-Linked Dominant, (C) X-Linked Recessive
6. Scientists are able to obtain karyotypes by taking digital pictures of the nucleus of a cell during the "Prophase" stage of mitosis, which is the only part of a cell's life cycle in which all of its DNA condenses into visible chromosomal structures. {2 pt}
7. Frameshift Mutation {2 pts}
8. (A) Nonsense Mutation, (B) The mutation would cause a stop codon to occur earlier within the sequence, making the polypeptide much shorter in length {1 pt}
9. Repeat Expansion {1 pt}
10. Deletion, Frameshift mutation, Nonsense mutation {2 pt}
11. Three reasons why mutations don't generally cause major impacts on organisms is the fact that (1) most mutagens are unable to reach the DNA due to the protection provided by the nucleus and skin pigments, (2) most mutations that impact DNA are corrected by DNA polymerases, and (3) a large portion of the human genome consists of introns, which will not have any impact on DNA function if mutated. {2 pts}
12. (A) 0.30 or 30%, (B) 42% {2 pt}
13. (A) 14.44%, (B) 76 individuals {2 pts}
14.  $\frac{1}{4}$  or 25% {1 pt}
15.  $\frac{3}{16}$  or 18.75% {2 pt}
16.  $\frac{1}{16}$  or 6.25% {3 pt}
17. Topoisomerase {1 pt}
18. (A) DNA Polymerase I, II, and III, (B) DNA Polymerase alpha, beta, and delta {2 pt}
19. Single strand binding proteins function to prevent rejoining of unwound strands and protects the ssDNA from nuclease degradation. {1 pt}
20. (A) DNA polymerase delta [ $\delta$ ], (B) DNA polymerase alpha [ $\alpha$ ], (C) DNA polymerase beta [ $\beta$ ] {1 pt}
21. Alternative Splicing {1 pt}
22. TATA Box {1 pt}
23. 100-200 base pairs {1 pt}
24. AUG; Methionine {1 pt}

25. UAA, UAG, UGA {1 pt}

26. Poly-A Tail

27. (A) Primary protein structure refers to the sequence of amino acids present within the polypeptide chain. (Ex: AUGCGAUUG). (B) Secondary protein structure refers to the folding or coiling of polypeptide chains as a result of hydrogen bonding. (Ex: Alpha helices and beta pleated sheets). (C) Tertiary protein structure refers to the overall 3-D structure of a polypeptide chain. (Ex: Protein shape held together by hydrophobic interactions, hydrogen bonding, and ionic bonding). (D) Quaternary protein structure refers to the structure of a protein macromolecule formed by interactions between multiple polypeptide chains. {2 pts}

28. A nucleosome is the basic structural unit of a eukaryotic chromosome, consisting of a length of DNA coiled around a core of histones. {1 pt}

29. (A) Telomere, (B) A telomere is a region of repetitive nucleotide sequences at the end of each chromosome that functions to protect the chromosome from deterioration or fusion with neighboring chromosomes. {2 pts}

30. A nucleotide consists of a deoxyribose sugar, a nitrogen base, and a phosphate group whereas a nucleoside only consists of a deoxyribose sugar and nitrogen base. {1 pt}

31. Law of Segregation {1 pt}

32. The geneticist is referring to the "short arms" of the 21st chromosome. {1 pt}

33. (A) Pyrimidines are one-carbon nitrogen rings of bases whereas purines are two-carbon nitrogen rings of nucleotide bases. (B) Thymine and cytosine are pyrimidines while adenine and guanine are purines. {2 pts}

34. A kinetochore is a complex protein associated with the centromere during cell division that functions to be the site of attachment for the spindle fibers of centrosomes. {1 pt}

35. A Barr body is a small, densely staining structure in the cell nuclei of female mammals, consisting of condensed, inactivated X chromosomes. They form in female individuals. {2 pts}

36. Systemic Lupus Erythematosus (SLE) {1 pt}

37. Aneuploidy is the state of the presence of abnormal amounts of chromosomes within a cell (e.g. 45, 47, etc.) {1 pt}

38. (A) Heterchromatic bands, (B) Euchromatic bands. {2 pts}

39. Sanger sequencing {1 pt}

40. In RFLP analysis, the DNA of various individuals are exposed to the same restriction enzyme, causing all of the DNA samples to splice at different intervals, creating strands of DNA varying in length. These DNA samples are then put through a gel electrophoresis, in which the individual fragments of the DNA are sorted by size.

Individuals with a similar gel electrophoresis reading are likely related or similar due to the similarity of the action of restriction enzymes upon their DNA. {2 pts}

41. C (Western Blotting) {1 pt}

42. The polymerase from PCR is derived from the *Thermophilus Aquaticus* bacterium, which is found in Yellowstone geysers. {2 pts}

43. Taq polymerase, the polymerase derived from *thermophilus aquaticus* bacteria, is used rather than traditional polymerase because it is able to withstand extremely high temperatures without denaturing, and the PCR technique involves strong heating of the sample. {2 pts}

44. DNA microarray / DNA hybridization {2 pts}

45. Gene therapy is a newly developing form of treatment for genetic diseases in which a pre-programmed viral vector with the healthy form of the gene is inoculated into the diseased patient, replacing the defective gene with a healthy one and therefore curing the disease. {2 pts}

46. crRNA and tracrRNA {1 pt}

47. Site targeted mutagenesis allows scientists and genetic engineers to deactivate certain genes, and therefore analyze their function and importance within cells. {1 pt}

48. Endonucleases {1 pt}

49. PCR is a popular technique because often, to perform certain sequencing tests, a particular amount of the copies of the DNA must be acquired, which scientists can do by using PCR. {1 pt}

50. Restriction enzymes {1 pt}

51. Metaphase plate {1 pt}

52. Prophase {1 pt}

53. 2-4 hrs {1 pt}

54. Nondisjunction is when chromosomes do not separate properly during some stage of meiosis, causing the number of chromosomes per gamete to vary inaccurately. {1 pt}

55. Anaphase {1 pt}

56. Phylogenetics is the study of evolutionary relationships between organisms based upon genetics. {1 pt}

57. Epigenetics is the study of changes brought out in organisms as a result in altered gene expression due to environmental factors rather than the change in the DNA sequence itself. {1 pt}