1. **Function Of Pedigree?**

A pedigree may be used to establish the probability of a child having a particular disorder or condition. It may be used to discover where the genes in question are located (x, y, or autosome chromosome), and to determine whether a trait is **dominant** or recessive.

2. **Define Epigenetics?**

Epigenetics is the study of heritable phenotype changes that do not involve alterations in the DNA sequence

3. **RAPD And ALFP Difference?**

RAPD is Random Amplification of Polymorphic DNA'. It is a type of PCR, but the segments of DNA that are amplified are random.

AFLP is a PCR-based tool used **in genetics** research, DNA fingerprinting, and **in** the practice of **genetic** engineering. Developed **in** the early 1990s by Keygene, **AFLP** uses restriction enzymes to digest genomic DNA, followed by ligation of adaptors to the sticky ends of the restriction fragments.

4. **Point Mutation?**

A **point mutation** or substitution is a genetic **mutation** where a single nucleotide base is changed, inserted or deleted from a sequence of DNA or RNA. **Point mutations** have a variety of effects on the downstream protein product—consequences that are moderately predictable based upon the specifics of the **mutation**.

5. **Complications Of Inheritance?**

Complications to inheritance patterns: Multiple genes - epistasis, polygenic traits Genes & the environment - sex-influenced traits, incomplete penetrance.
Complications : A common recessive inheritnace can give dominant pattern, Autosomal dominant inheritance with non-penetrance, Autosomal dominant inheritance with variable expression.

6. **Mutation Effects In Functioning Of Genes?**

Some mutations don't have any noticeable effect on the phenotype of an organism. This can happen in many situations: perhaps the mutation occurs in a stretch of DNA with no function, or perhaps the mutation occurs in a **protein**-coding region, but ends up not affecting the amino acid sequence of the **protein**.

7. **Causes Of Epigenetics?**

Stress Hormone **Causes Epigenetic** Changes. Researchers found that chronic exposure to a stress hormone **causes** modifications to DNA in the brains of mice, prompting changes in gene expression

8. **Genetic Mapping In Humans?**

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9. **Forward, Reverse, And Neutral Mutations: 3marks**

Forward mutation: Mutation that alters the wild type phenotype into mutant phenotype.
Reverse mutation: Mutation that changes a mutant phenotype back into the wild type.
Neutral mutation: Mutation that alters the amino acid sequence of the protein but does not change its function as replaced amino acid is chemically similar or the affected AA has little influence on protein function

10. **Point Mutations And Enlist 3 Structural Changes In Chromosomes?**

A **point mutation** or substitution is a genetic **mutation** where a single nucleotide base is changed, inserted or deleted from a sequence of DNA or RNA. **Point mutations** have a variety of effects on the downstream protein product—consequences that are moderately predictable based upon the specifics of the **mutation**.
Deletion, duplication, translocation, transcription.

Structural Change in the Structure of Chromosomes. ... Chromosomes are the vehicle of hereditary material or genes. Any alteration, addition or deletion of chromosomal part leads to alteration of number, position or sequence of genes in the chromosome.

11.5 Complications In Inheritance?

Epistasis, polygenic traits Genes & the environment - sex-influenced traits, incomplete penetrance.

12. Modification Of Epigenetica

Epigenetic modification - non coding RNA is a new mechanism for gene regulation. RNA which is not used for making proteins can be cleaved and used to inhibit protein-coding RNAs (siRNAs, microRNAs). Three common types of modification - Cytosine methylation¬ Histones modifications¬ siRNAs¬

13. Mutation Alter Genes Expression?

Non-coding mutations: Promoter/enhancer element trinucleotide repeats in 5 and 3'UTRs Splice sites. Effect of mutation on gene function. Null allele (loss of function) - no gene product.
Hypomorph – decreased activity. Gain of function - increased activity. Dominant negative-antagonizes normal product. Neomorph - novel activity. Predicting that a gene product won't do the job. Deletion, nonsense, frameshift of sequence is deleterious. Mutation in splice site usually has sever effects. Missense mutations depend on location in protein. Is it non-conservative. Is AA conserved in evolution.

14. RFLP And RAPD ?

RFLP is a technique that exploits variations in homologous DNA sequences. **RFLP** analysis was an important tool in genome mapping, localization of genes for genetic disorders, determination of risk for disease, and paternity testing.

RAPD is Random Amplification of Polymorphic DNA'. It is a type of PCR, but the segments of DNA that are amplified are random.

15. Genetic Mapping?

Gene mapping describes the methods used to identify the locus of a gene and the distances between genes. The essence of all genome mapping is to place a collection of molecular markers onto their respective positions on the genome.

16. Sickle cell anemia?

Sickle cell anemia (**sickle cell disease**) is a disorder of the blood caused by an inherited abnormal hemoglobin (the oxygen-carrying protein within the red blood **cells**). The abnormal hemoglobin causes distorted (sickled) red blood **cells**. The sickled red blood **cells** are fragile and prone to rupture.

17. Point mutation and cause?

A **point mutation** or substitution is a genetic **mutation** where a single nucleotide base is changed, inserted or deleted from a sequence of DNA or RNA. **Point mutations** have a variety of effects on the downstream protein product—consequences that are moderately predictable based upon the specifics of the **mutation**.

Cause:

The protein may lose its function, which can **result in**a disease in the organism. For example, sickle-cell disease is **caused** by a single **point mutation** (a missense **mutation**) in the beta-hemoglobin gene that converts a GAG codon into GUG, which encodes the amino acid valine rather than glutamic acid.

18. Mutation nomenclature?

Standard **nomenclature** based on genomic DNA reference sequences and RNA reference sequences is not shown. "Genomic DNA reference sequence" simply indicates any human DNA sequence in the database that is not based on a cDNA sequence. ... Example of standard **mutation nomenclature** based on a coding DNA sequence.

19. VNTR and application?

VNTR are short nucleotide sequences organized as a tandem repeats on genomes. Found on many chromosomes, Show variations in length between individuals.

Applications of VNTR Microbiology DNA fingerprinting Genetic diversity Forensics Mapping of genomes Breeding patterns of wild or domesticated animals .

20. Single And Multifactorial Mutation?

Single gene disorders are caused by DNA changes in one particular **gene**, and often have predictable inheritance patterns. Over 10,000 human disorders are caused by a change, known as a **mutation**[?], in a **single gene**[?]. These are known as **single gene** disorders

Multifactorial Disorders. Researchers are learning that nearly all conditions and **diseases** have a genetic component. Some disorders, such as sickle cell disease and cystic fibrosis, are caused by **mutations** in a single gene. ... Conditions caused by many contributing factors are called complex or **multi factorial** disorders.

21. 5 Complication Inheritance Pattern?

Complications to inheritance patterns Multiple genes - epistasis, polygenic traits Genes & the environment - sex-influenced traits, incomplete penetrance

22. Autosomal Domenet Disordered?

A single abnormal gene on one of the first 22 nonsex (**autosomal**) chromosomes from either parent can cause an **autosomal** disorder. **Dominant** inheritance means an abnormal gene from one parent can cause disease. ... Examples of **autosomal dominant disorders** include Marfan syndrome and neurofibromatosis type 1.

23. Function Of STR?

Short Tandem Repeat (**STR**) analysis is one of the most useful methods in molecular **biology** which is used to compare specific loci on DNA from two or more samples. A short tandem repeat is a microsatellite, consisting of a unit of two to thirteen nucleotides repeated hundreds of times in a row on the DNA strand.

24. During DNA Extraction How Proteins Are Denatured?

Adding proteinase

25. Define Locus?

A **locus** (plural **loci**) in **genetics** is a fixed position on a chromosome, like the position of a gene or a marker (**genetic** marker). Each chromosome carries many genes; human's estimated 'haploid' protein coding genes are 19,000–20,000, on the 23 different chromosomes.

26. Sex Linked Traits And Disorder?

Genes on the X or Y chromosome are called **sex-linked**. ... For example, an X-**linked** recessive allele in humans causes haemophilia, which is much more common in males than females because males are hemizygous and therefore express the**trait** when they inherit one mutant allele.

27. Define Multiple Alleles With Example?

Multiple alleles Three or more alternative forms of a gene (**alleles**) that can occupy the same locus. However, only two of the **alleles** can be present in a single organism. For example, the ABO system of blood groups is controlled by three **alleles**, only two of which are present in an individual.

28. Genetic Disorder?

A **genetic disorder** is a **genetic** problem caused by one or more abnormalities in the genome. Most **genetic disorders**are quite rare and affect one person in every several thousands or millions. **Genetic disorders** may be hereditary, meaning that they are passed down from the parents' **genes**.

29. Genetic Mapping?

Gene mapping describes the methods used to identify the locus of a gene and the distances between genes. The essence of all genome mapping is to place a collection of molecular markers onto their respective positions on the genome.

30. What Is Location Of Intronic Sequence With Respect To Exons?

Sequences that are joined together in the final mature RNA after RNA splicing are**exons**. **Introns** are **found** in the genes of most organisms and many viruses, and can be **located** in a wide range of genes, including those that generate proteins, ribosomal RNA (rRNA), and transfer RNA (tRNA).

31. Is There Any Rules For Mutation Nomenclature?

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32. Write Down At Least Three Mitochondrial Diseases?

MELAS

Mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes is MELAS.

Kearns-Sayre Syndrome

NARP

NARP stands for neurogenic weakness, ataxia and retinitis pigmentosa.

33. Structul chamge in chromosome?

Structural Change in the **Structure** of **Chromosomes**. ... **Chromosomes** are the vehicle of hereditary material or genes. Any alteration, addition or deletion of **chromosomal** part leads to alteration of number, position or sequence of genes in the **chromosome**.

34.

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