Types of mutation: Followings are the types of mutations.

➢ Points Mutations

➢ Variations in chromosomes number

➢ Variations in chromosomes structure

➢ Point mutation

Base Pair Substitutions Silent,

Missense, Nonsense ,

Base pair insertions/deletions ,Triplet Repeats

,Frameshift

➢ Variation in Chromosome Number

Aneuploidy,

Hyperploidy, Hypoploidy ,

Euploidy ,

Monoploidy, Diploidy, Polyploidy

➢ Variation in chromosome structure Deletions,Translocations ,Duplications ,Inversions

,Mutations and effects

DNA polymorphism

 DNA polymorphism is a sequence difference compared to a reference standard present in at least 1–2% of a population.

DNA Polymorphism may be single nucleotide or thousands of BASES

Polymorphisms can be due to a single base or thousands of bases.

DNA Polymorphism may or may not have phenotypic effects

Polymorphisms may or may not have phenotypic effects.

DNA POLYMORPHISM - SNP

DNA Polymorphism are found throughout the genomes

Polymorphisms are found throughout the genome.

Polymorphic sequence can serve as marker

 If the location of a polymorphic sequence is known, it can serve as a marker for locating other genes or genetic regions.

Polymorphic markers have different versions

 Each polymorphic marker has different versions or alleles.

Polymorphisms can be silent, or be exhibited at levels of Proteins

The principle of Southern blotting involves the transfer of DNA fragments from a gel onto a nitrocellulose or nylon membrane, which is then hybridized with a labeled DNA probe that is complementary to the target sequence of interest. The labeled probe hybridizes specifically to the target sequence, allowing it to be visualized and detected.

Diploid cells and haploid cells

Diploid cells are body cells. They are called as (2n) cells. Examples: skin cells, brain cells, bone cells, blood cells. In humans, ~ 220 types of somatic cells.

Haploid cells Sex cells produce gametes. Gametes are haploid (n) cells. Sperms and ova are gametes which are produced in males and females respectively.

Organic method of DNA extraction

There are several organic methods for DNA extraction, but a commonly used protocol is the phenol-chloroform method. Here's a brief overview of the steps involved:

Cell lysis: Cells are first lysed to release the DNA. This can be done using a variety of methods, such as mechanical disruption (e.g. grinding or homogenization), enzymatic digestion, or detergent-based lysis.

Removal of proteins: The lysate is treated with a protease (such as proteinase K) to digest the proteins that are present in the sample. This step helps to remove any proteins that could interfere with DNA isolation.

Phenol-chloroform extraction: Phenol and chloroform are organic solvents that can be used to separate DNA from other cellular components. In this step, an equal volume of phenol-chloroform is added to the sample and mixed thoroughly. The mixture is then centrifuged, which causes the phases to separate. The aqueous (top) layer contains the DNA, while the organic (bottom) layer contains other cellular components.

How protein can be denatured from DNA extraction ?

Proteins can be denatured by SDS or by heat.

Applications of VNTR

 Microbiology

 DNA fingerprinting

 Genetic diversity

 Forensics

 Mapping of genomes

 Breeding patterns of wild or domesticated animals

Meiosis I – Reductional division

Meiosis I results in two haploid daughter cells. It is called the reductional division. Homologous chromosomes separate.

Meiosis II: Second cell division where sister chromatids separate. As a result of meiosis II, two haploid cells become four haploid cells. Meiosis II behaves like mitosis, where no reduction in chromosomes number.

Mono-allelic, di-allelic and poly-allelic

 30% genes in humans are di-allelic - they exist in two forms.

 70% are mono-allelic; they exist only in one form.

 Few are poly-allelic.

Environmental effects on allelic expression

 Allele expression may be affected by environmental

conditions

 Examples:

- Coat color in arctic foxes
- Himalayan rabbits
- Siamese cats

Explanation

 ch allele affected by temp >33 C tyrosinase enzyme inactivates and reduces

melanin pigment.

HGNC rules

 ">" indicates a substitution at DNA level (like c.76A>T)

 "_" (underscore) indicates a range of affected residues, separating the first and last residue

affected (like c.76_78delACT).

 "del" indicates a deletion (like c.76delA).

 "dup" indicates a duplication (like c.76dupA); duplicating insertions are described as duplications, not as insertions.

 "ins" indicates a insertion (like c.76_77insG)

 "inv" indicates an inversion (like c

Linkage map of drosophila chromosome

Linkage map

 A linkage map is a genetic map of a species that shows the position of its known genes or genetic markers relative to each other in terms of recombination frequency.

Frequency of recombinant offspring reflect the distance

Frequency of recombinant offspring reflected distances between genes on chromosomes.

Greater the distance, more chances of cross over

 The greater the distance between two genes, the more points between them where crossing over can occur.

Linkage map

 Recombination frequency between cn and b is 9%

 Between cn and vg is 9.5%

 Recombination frequency between b and vg is 17%

Mutation nomenclatur

 All variants should be described at the most basic level, i.e. DNA level.

 Descriptions should always be in relation to a reference sequence, either a genomic or a coding DNA sequence.

 Although theoretically a genomic reference sequence seems best, in practice a coding DNA reference sequence is preferred.

 Good for promoters, alternative splicing etc.

 When the entire genomic sequence is not known, a cDNA reference sequence should be used.

 To avoid confusion in the description of a variant it should be preceded by a letter indicating the

type of reference sequence used.

 Several different reference sequences can be used.

 "c." for a coding DNA sequence (like c.76A>T)

 "g." for a genomic sequence (like g.476A>T)

 "m." for a mitochondrial sequence (like m.8993T>C,)

 "r." for an RNA sequence (like r.76a>u)

 "p." for a protein sequence (like p.Lys76Asn)

 Describing genes / proteins, only official HGNC (HUGO

What is active reagent to trizol ? Write steps of rna extraction from tissue by trizol reagent

Trizol (also known as TRI Reagent) is a commonly used reagent for RNA extraction from biological tissues. The active reagent in Trizol is a mixture of phenol and guanidine isothiocyanate, which help to denature proteins and disrupt cellular membranes, allowing for RNA isolation. Here are the general steps for RNA extraction from tissue using Trizol:

Homogenization: Tissue samples are first homogenized in Trizol reagent using a homogenizer or other mechanical disruption method to release the RNA.

Phase separation: Chloroform is added to the sample, and the mixture is vigorously shaken and then centrifuged. This causes the sample to separate into three phases: an upper aqueous phase (containing RNA), an interphase (containing DNA and protein), and a lower organic phase (containing lipids and other contaminants).

RNA precipitation: The upper aqueous phase is carefully removed and mixed with an equal volume of isopropanol to precipitate the RNA. The mixture is then incubated at room temperature for several minutes, allowing the RNA to form a visible pellet.

RNA purification: The RNA pellet is washed with 75% ethanol to remove any remaining contaminants, and then resuspended in an appropriate buffer for downstream applications. Optionally, the RNA can be treated with DNase to remove any contaminating DNA.

Nonsense mutation: Nonsense mutations convert an amino acid into a stop codon. The effect is to shorten the resulting protein. Sometimes this has only a little effect, as the ends of proteins are often

relatively un-important to function. However, often nonsense mutations result in completely nonfunctional proteins.

Loss of function, Gain of function

Loss of function: Complete or partial loss of the normal function. Structure of protein is so altered that it is no longer works correctly. Mutation can occur in regulatory regions that affect transcription, translation or splicing of the protein. Frequently recessive

Gain of function: Produces an entirely new trait. Such a mutation causes a trait to appear in inappropriate tissues or at inappropriate times in development. It is frequently dominant.

Spontaneous and Induced mutation

Spontaneous mutation: Mutations that result from natural changes in DNA

Origins of Spontaneous mutation

Induced mutation results from changes caused by environmental, chemicals and radiations.

Mutagen: Any environmental agent that increases the rate of mutation above the spontaneous is called a mutagen such as chemicals & radiations, X-rays, Mustard gas etc

Chromosome Types: Centromere Location.

Types of chromosomes according to the Centromere location.

Chromosomes can be classified into different types based on the location of their centromere, which is the specialized region of the chromosome that plays a critical role in chromosome segregation during cell division. The three main types of chromosomes based on their centromere location are:

Metacentric chromosomes: These chromosomes have a centromere located at or near the middle, resulting in arms of roughly equal length.

Submetacentric chromosomes: These chromosomes have a centromere that is slightly off-center, resulting in one arm that is shorter than the other.

Acrocentric chromosomes: These chromosomes have a centromere that is located very close to one end, resulting in one very short arm and one long arm. In humans, chromosomes 13, 14, 15, 21, and 22 are acrocentric.

There is also a fourth type of chromosome, telocentric chromosomes, in which the centromere is located at the very end of the chromosome. However, telocentric chromosomes are relatively rare in most organisms, and humans do not have any telocentric chromosomes.

Role of mgcl2 in PCR and EDTA in gel electrophoresis.

Magnesium chloride (MgCl2) is a divalent cation that is commonly added to polymerase chain reaction (PCR) reactions to facilitate the activity of Taq polymerase, which is the enzyme responsible for amplifying the DNA template. MgCl2 is a cofactor that helps stabilize the enzyme and optimize its activity, leading to improved efficiency and specificity of the PCR reaction. The optimal concentration of MgCl2 varies depending on the reaction conditions and the specific Taq polymerase used, but is typically in the range of 1.5 to 4 mM.

On the other hand, Ethylenediaminetetraacetic acid (EDTA) is a chelating agent that is commonly used in gel electrophoresis to prevent the degradation of DNA by nucleases. EDTA binds to divalent cations such as Mg2+ and Ca2+, which are cofactors for many nucleases, rendering them inactive. By chelating these cations, EDTA can protect the DNA from degradation during electrophoresis and subsequent staining or visualization. EDTA is typically added to the loading buffer or electrophoresis buffer at a concentration of 0.5 to 5 mM.

Erythrocyte lysis In RNA/Erythrocyte extraction from RNA purification

Erythrocyte lysis is a process used to break open red blood cells (erythrocytes) in order to release their contents, including RNA. This process is commonly used in RNA extraction protocols to obtain RNA samples from whole blood, which contains a mixture of various cell types, including erythrocytes, leukocytes, and platelets.

One commonly used method for erythrocyte lysis in RNA extraction is the use of a hypotonic buffer, such as ammonium chloride, which causes the erythrocytes to swell and burst, releasing their contents into the solution. The leukocytes and platelets are relatively resistant to lysis under these conditions, allowing for the isolation of RNA from these cell types.

After erythrocyte lysis, the RNA can be extracted from the solution using various methods, such as column-based or bead-based purification. The extracted RNA can then be further purified and quantified for downstream applications, such as gene expression analysis or sequencing.

Hot star PCR and suicide PCR.

Hot start PCR and suicide PCR are two types of modified PCR techniques that are used to improve the specificity and sensitivity of the amplification.

Hot start PCR is a modification of standard PCR in which the reaction mix is heated to an intermediate temperature (usually 95-98°C) before the addition of the Taq polymerase enzyme. This allows the reaction to undergo an initial denaturation step without the presence of the polymerase, which can reduce nonspecific amplification caused by the presence of primer-dimer or misprimed products. After the initial denaturation, the reaction is cooled to the optimal temperature for Taq polymerase activity and the polymerase is added. This technique can improve specificity and sensitivity of PCR, especially for difficult targets or complex templates.

Suicide PCR, on the other hand, is a technique that uses a modified primer in which the 3' end of the primer contains a degenerate base that can only be extended by the correct target DNA template. If the target DNA is not present, the primer will not be extended and will act as an inhibitor for subsequent PCR reactions. This helps to reduce the formation of nonspecific amplification products and can improve the specificity of PCR. The modified primer is designed to be used in a limited-cycle PCR reaction, in which a small number of cycles (e.g., 1-3 cycles) are performed before the reaction is stopped. The products of the limited-cycle PCR are then used as a template for a standard PCR reaction, which is carried out with regular primers to amplify the target DNA specifically. This technique is particularly useful for amplifying low-abundance targets or targets that are difficult to amplify with standard PCR.

Down Syndrome Clinical Features:

Down syndrome is a genetic disorder caused by the presence of an extra copy of chromosome 21. The clinical features of Down syndrome can vary widely, but some common features include:

Intellectual disability

Developmental delays, such as delayed speech and motor skills

Hypotonia (low muscle tone)

Upward slanting eyes, small ears, and a small nose

Short stature

Single palmar crease (instead of two)

Congenital heart defects

Gastrointestinal issues, such as reflux and constipation

Increased risk for certain medical conditions, such as sleep apnea, hearing loss, and thyroid disorders

Autosomal Recessive Diseases:

Autosomal recessive diseases are genetic disorders that are caused by mutations in a gene located on one of the autosomes (non-sex chromosomes) that result in a loss or reduction of function of the encoded protein. In order to inherit an autosomal recessive disorder, an individual must inherit two copies of the mutated gene, one from each parent. This means that individuals who carry only one copy of the mutated gene (i.e., are heterozygous carriers) are typically unaffected but can pass on the mutation to their children.

Examples of autosomal recessive diseases include cystic fibrosis, sickle cell anemia, Tay-Sachs disease, and phenylketonuria (PKU).

Steps of DNA Purification:

The general steps involved in DNA purification from a biological sample include:

Cell lysis: Breaking open the cells to release the DNA.

Protein removal: Removing proteins and other cellular debris from the sample, typically through precipitation or filtration.

DNA isolation: Separating the DNA from other cellular components, such as RNA and proteins, typically through precipitation or centrifugation.

DNA purification: Cleaning and concentrating the DNA using various methods, such as column-based purification, ethanol

Precipitation, or silica-based purification.

Quality control: Assessing the quantity and quality of the purified DNA, typically through spectrophotometry or gel electrophoresis.

Base Analogs:

Base analogs are chemicals that can substitute for the normal nucleotide bases (A, T, G, C) during DNA replication or repair, leading to mutations in the DNA sequence. Base analogs can be incorporated into the DNA during replication if they are structurally similar enough to the normal nucleotides. Examples of base analogs include 5-bromouracil and 2-aminopurine.

Role of MgCl2 and EDTA Buffer:

MgCl2 is often added to PCR reactions as a cofactor for the Taq polymerase enzyme, which helps stabilize the enzyme and optimize its activity. The optimal concentration of MgCl2 varies depending on the reaction conditions and the specific Taq polymerase used, but is typically in the range of 1.5 to 4 mM.

EDTA buffer is often used in gel electrophoresis to prevent the degradation of DNA by nucleases. EDTA binds to divalent cations such as Mg2+ and Ca2+, which are cofactors for many nucleases, rendering them inactive. By chelating these cations, EDTA can protect the DNA from degradation during electrophoresis and subsequent staining or visualization. EDTA is typically added to the loading buffer or electrophoresis buffer at a concentration of 0.5 to 5 mM.

Common Recessive Disorders:

Examples of common autosomal recessive disorders include:

Cystic fibrosis

Sickle cell anemia

Tay-Sachs disease

Phenylketonuria (PKU)

Galactosemia

Congenital adrenal hyperpl

Five steps of organic mechanisms of lysis.

Organic mechanisms of lysis involve the use of organic solvents to disrupt the cell membrane and release cellular components. The general steps involved in organic mechanisms of lysis include:

Sample preparation: The biological sample is collected and prepared for lysis, typically through washing or centrifugation to remove excess buffer or cellular debris.

Organic solvent addition: An organic solvent, such as ethanol or methanol, is added to the sample to disrupt the cell membrane and denature proteins. The sample is then vortexed or mixed to ensure even distribution of the solvent.

Incubation: The sample is incubated at a specific temperature and time to allow the organic solvent to dissolve the lipids in the cell membrane and cause the cells to lyse. The optimal incubation conditions will vary depending on the specific protocol and sample type.

Centrifugation: The lysate is centrifuged to separate the insoluble cellular debris from the soluble components, such as nucleic acids or proteins. The supernatant, which contains the soluble components, is then transferred to a new tube for further processing.

Recovery: The soluble components can be recovered from the supernatant using various methods, such as precipitation, column-based purification, or extraction. The optimal recovery method will depend on the specific component of interest and downstream application.

Three mitochondial diseases.

Mitochondrial diseases are a group of genetic disorders that affect the function of the mitochondria, which are the powerhouses of the cell. Here are three examples of mitochondrial diseases:

Leigh Syndrome: This is a rare, inherited disorder that affects the central nervous system, causing progressive neurological deterioration. Symptoms can include weakness, movement disorders, seizures, developmental delay, and vision and hearing problems. The disease is caused by mutations in mitochondrial or nuclear DNA that affect the function of the mitochondria.

MELAS Syndrome: This stands for Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-like episodes. It is a rare genetic disorder that affects multiple organ systems, including the brain, muscles, and heart. Symptoms can include muscle weakness, recurrent headaches, seizures, hearing loss, and stroke-like episodes that can cause visual disturbances, confusion, and weakness on one side of the body. The disease is caused by mutations in mitochondrial DNA that affect the function of the mitochondria.

Leber's Hereditary Optic Neuropathy (LHON): This is an inherited disorder that primarily affects the eyes, causing sudden vision loss in young adults. It is caused by mutations in mitochondrial DNA that affect the function of the mitochondria in the cells of the optic nerve. Other symptoms can include painless vision loss, color vision abnormalities, and eye pain. There is currently no cure for LHON, but certain treatments may help slow the progression of the disease.

DNA quantification. Explain

DNA quantification is a process used to determine the concentration or amount of DNA present in a given sample. This can be important for a variety of applications, such as preparing samples for downstream analysis in research or clinical settings.

There are several methods for DNA quantification, including:

UV spectrophotometry: This method involves measuring the absorbance of DNA at a specific wavelength using a UV spectrophotometer. DNA has a characteristic absorbance peak at 260 nm, which can be used to determine the concentration of DNA in a sample. However, other substances present in the sample, such as RNA or proteins, can also absorb at this wavelength and interfere with the measurement.

Fluorometry: This method involves using a fluorescent dye that binds to DNA molecules and emits fluorescence when excited by a specific wavelength of light. The amount of fluorescence emitted is proportional to the amount of DNA present in the sample. Fluorometric assays can be more sensitive and specific than UV spectrophotometry, but may be affected by the presence of contaminants that can also bind to the dye.

Gel electrophoresis: This method involves running the DNA sample on an agarose gel and staining it with a dye such as ethidium bromide. The intensity of the DNA band can be compared to known standards of DNA of known concentrations to estimate the amount of DNA present in the sample.

Regardless of the method used, it is important to ensure that the DNA sample is free of contaminants such as proteins, salts, or other substances that may interfere with the quantification process. Additionally, it is important to use appropriate standards and controls to ensure accurate and reliable measurements.

Pleiotropy and its effects.

Pleiotropy is a phenomenon where a single gene or genetic variant can affect multiple seemingly unrelated traits or phenotypes. In other words, a single genetic factor can have multiple effects on an individual's phenotype.

Pleiotropic effects can have both positive and negative consequences. Some examples of pleiotropic effects are:

Sickle cell anemia: This is a genetic disorder caused by a mutation in the HBB gene that leads to the production of abnormal hemoglobin. This mutation is also associated with resistance to malaria, which has a positive effect, but sickle cell anemia itself can cause a number of negative effects such as chronic pain, organ damage, and increased risk of infection.

Marfan syndrome: This is a genetic disorder caused by mutations in the FBN1 gene, which codes for fibrillin, a protein that helps give connective tissue its elasticity. People with Marfan syndrome may have long limbs, a curved spine, and other skeletal abnormalities, but they also have a higher risk of cardiovascular problems such as aortic aneurysms.

Coat color in dogs: Some genes that control coat color in dogs have pleiotropic effects, such as the MC1R gene that controls the production of the pigment melanin. Mutations in this gene can lead to red or yellow coat colors, but they are also associated with increased risk of certain types of cancer.

Overall, pleiotropy underscores the complex interplay between genes and phenotype, and highlights the fact that genetic variation can have multiple effects on an individual's health and well-being. Understanding pleiotropic effects can be important for developing treatments and interventions for genetic disorders.

1.Non sense mutation.

2.prophase of mitosis.

3 Differentiate western, southern and northern blotting.

Non-sense mutation:

A nonsense mutation is a type of point mutation that occurs when a single nucleotide change in DNA leads to a premature stop codon in the mRNA. This stop codon signals the termination of translation, resulting in a truncated or nonfunctional protein. Nonsense mutations are associated with a number of genetic disorders, including cystic fibrosis and Duchenne muscular dystrophy.

Prophase of mitosis:

Prophase is the first stage of mitosis, the process by which cells divide to produce two genetically identical daughter cells. During prophase, the chromatin in the nucleus condenses into visible chromosomes, each consisting of two identical sister chromatids joined at the centromere. The nuclear envelope breaks down, and spindle fibers form from the centrosomes, which move to opposite poles of

the cell. The spindle fibers then attach to the chromosomes, preparing them for alignment and separation during later stages of mitosis.

Western, Southern, and Northern blotting:

Western, Southern, and Northern blotting are techniques used to detect specific nucleic acid or protein molecules in a sample.

Western blotting involves separating proteins in a sample by size using gel electrophoresis, transferring the proteins to a membrane, and then using specific antibodies to detect the presence and quantity of a particular protein of interest.

Southern blotting is used to detect the presence and quantity of specific DNA sequences in a sample. The technique involves digesting DNA with restriction enzymes, separating the fragments by size using gel electrophoresis, and then transferring the fragments to a membrane for detection using labeled DNA probes.

Northern blotting is used to detect specific RNA molecules in a sample. The technique involves separating RNA molecules by size using gel electrophoresis, transferring the RNA to a membrane, and then using labeled DNA probes to detect the presence and quantity of a particular RNA molecule of interest.

In summary, while all three techniques involve separating molecules by size and transferring them to a membrane for detection, they differ in the type of molecule being detected and the type of probe used for detection.

1 differentiate forward, reverse and neutral mutation.

2 characteristics of mitochondial inheritance.

Differentiate forward, reverse, and neutral mutation:

Forward mutation: A forward mutation is a type of mutation that changes the wild-type allele to a different allele. This can lead to a new trait or phenotype in an organism, as the new allele may have altered or lost the function of the original allele.

Reverse mutation: A reverse mutation, also known as a reversion mutation, is a type of mutation that changes a mutant allele back to the wild-type allele. This can occur when a mutation that caused a loss of function in a gene is reversed, leading to the restoration of the original gene function.

Neutral mutation: A neutral mutation is a type of mutation that does not affect the function of the gene or the phenotype of the organism. This can occur when a mutation affects a non-coding region of DNA or when a coding mutation results in the same amino acid being incorporated into the protein.

Characteristics of mitochondrial inheritance:

Mitochondrial inheritance is a unique form of genetic inheritance that is characterized by several key features:

Mitochondrial DNA (mtDNA) is inherited only from the mother. This is because the egg cell contributes the majority of the cytoplasm to the developing embryo, which includes the mitochondria.

mtDNA is circular and exists in multiple copies within each mitochondrion, and multiple mitochondria exist within each cell.

Mitochondrial disorders can be caused by mutations in mtDNA, which can affect the function of the mitochondria and lead to a wide range of symptoms, including muscle weakness, vision and hearing loss, and neurological problems.

Mitochondrial disorders can be inherited in a variety of ways, including through maternal inheritance, spontaneous mutations, and somatic mutations.

Mitochondrial disorders can have variable penetrance and expressivity, meaning that the severity of the symptoms and the age of onset can vary widely among affected individuals and families. This is due in part to the fact that different cells within an individual can have different amounts of mutated mtDNA, leading to differences in mitochondrial function and symptoms.

1.Autosome and sex chromosome.

2.Uses of PCR.

What is DNA Mutation deletion Insertion effect on protein mutation.

Autosome and sex chromosome:

Autosomes are the non-sex chromosomes in a cell, meaning that they do not determine an individual's sex. Humans have 22 pairs of autosomes, which are numbered from 1 to 22. Sex chromosomes, on the other hand, are the chromosomes that determine an individual's sex. In humans, sex chromosomes come in two types: X and Y. Females have two X chromosomes (XX), while males have one X and one Y chromosome (XY).

Uses of PCR:

PCR (polymerase chain reaction) is a powerful molecular biology technique that allows for the amplification of specific DNA sequences. Some common uses of PCR include:

DNA cloning and sequencing: PCR can be used to amplify a specific DNA sequence of interest, which can then be cloned or sequenced for further analysis.

Genetic testing: PCR can be used to detect the presence of specific DNA sequences that are associated with genetic disorders or diseases.

Forensic analysis: PCR can be used to amplify DNA from small or degraded samples, such as those found at a crime scene.

Disease diagnosis: PCR can be used to detect the presence of infectious agents, such as viruses or bacteria, in clinical samples.

Deletion and insertion mutations are types of frame-shift mutations that can have significant effects on the resulting protein. In a deletion mutation, one or more nucleotides are removed from the DNA sequence, leading to a shift in the reading frame and a truncated or nonfunctional protein. In an insertion mutation, one or more nucleotides are added to the DNA sequence, also leading to a shift in the reading frame and a truncated or nonfunctional protein. These mutations can have range of effects, from mild to severe, depending on the location and extent of the mutation and the function of the affected protein.

1Differ in ring chromosome and phalidelphia chromosomes.

2.difference in PFLP and AFLP.

Differences between ring chromosome and Philadelphia chromosome:

Ring chromosome and Philadelphia chromosome are both structural chromosomal abnormalities that can cause genetic disorders. The main differences between the two are:

Ring chromosome: This occurs when a piece of a chromosome breaks off and forms a ring-shaped structure that can lead to loss of genetic material. The symptoms of ring chromosome disorders can vary depending on which chromosome is affected and which genes are lost.

Philadelphia chromosome: This is a specific type of chromosome abnormality that is associated with chronic myelogenous leukemia (CML). It occurs when pieces of chromosomes 9 and 22 break off and swap places, creating an abnormal fusion gene called BCR-ABL1. This gene produces a protein that leads to uncontrolled cell growth and division, leading to cancer.

Differences between PFLP and AFLP:

PFLP (PCR-based fragment length polymorphism) and AFLP (amplified fragment length polymorphism) are both molecular marker techniques used to analyze genetic variation. The main differences between the two are:

Methodology: PFLP is based on the length variation of PCR products amplified from genomic DNA using primers that flank variable DNA sequences. AFLP, on the other hand, uses restriction enzymes to digest genomic DNA, followed by ligation of adaptors and PCR amplification of fragments selected by specific primers.

Sensitivity: PFLP is less sensitive than AFLP, as it can only detect DNA polymorphisms that result in fragment length differences of several base pairs. AFLP, on the other hand, can detect polymorphisms as small as a single base pair.

Throughput: PFLP is typically used for analyzing a small number of loci, while AFLP is capable of analyzing hundreds to thousands of loci simultaneously, making it a high-throughput method.

Cost: PFLP is relatively inexpensive and requires only basic equipment, while AFLP is more expensive and requires specialized equipment and reagents.

1Missense mutation and its effects.

2Continues and discontinues variations.

3 five basic complications of mendalian inheritance.

4Types of genetic disorders.

Missense mutation and its effects:

Missense mutation is a type of point mutation that results in the replacement of a single nucleotide in a DNA sequence, causing the substitution of one amino acid for another in the resulting protein. Depending on the location and nature of the amino acid change, missense mutations can have a wide range of effects on protein structure and function. In some cases, missense mutations may be silent, meaning they have no effect on protein function. In other cases, they can lead to a partial loss of function, altered function, or even gain of function, which can result in genetic disorders.

Continuous and discontinuous variations:

Continuous variation refers to a range of phenotypic variation in a population that follows a normal distribution, such as height or weight. Discontinuous variation, on the other hand, refers to traits that can be divided into distinct categories, such as blood type or eye color.

Five basic complications of Mendelian inheritance:

Mendelian inheritance is the way in which genetic traits are passed from one generation to the next according to the laws of Gregor Mendel. However, there are several complications that can arise in Mendelian inheritance, including:

Incomplete dominance: When the heterozygous phenotype is intermediate between the homozygous dominant and homozygous recessive phenotypes.

Codominance: When both alleles of a gene are expressed equally in the heterozygous phenotype.

Multiple alleles: When there are more than two alleles for a particular gene in a population.

Pleiotropy: When a single gene affects multiple traits.

Epistasis: When the expression of one gene is influenced by the presence of one or more other genes.

Types of genetic disorders:

Genetic disorders can be classified into several different categories, including:

Single-gene disorders: Disorders caused by mutations in a single gene, such as cystic fibrosis or sickle cell anemia.

Chromosomal disorders: Disorders caused by abnormalities in chromosome number or structure, such as Down syndrome or Turner syndrome.

Multifactorial disorders: Disorders caused by a combination of genetic and environmental factors, such as heart disease or diabetes.

Mitochondrial disorders: Disorders caused by mutations in the DNA of the mitochondria, which are organelles within cells that produce energy. Examples include Leber's hereditary optic neuropathy and mitochondrial encephalopathy.

1Epistasis

2Diagnosis test for genetic disorder

3 what is short range.

4 application of molecular markers

Epistasis:

Epistasis is a genetic interaction between two or more genes where the effect of one gene (the "modifier gene") masks or modifies the effect of another gene (the "target gene"). Epistasis can result in a wide range of phenotypic outcomes, including dominant or recessive expression, incomplete dominance, or gene interactions that result in new phenotypes.

Diagnosis tests for genetic disorders:

There are many different tests available for diagnosing genetic disorders. Some common tests include:

Karyotyping: A test that examines the number and structure of chromosomes in a sample of cells

Polymerase chain reaction (PCR): A technique used to amplify DNA segments for analysis

DNA sequencing: A method used to determine the precise sequence of nucleotides in a DNA molecule

Fluorescence in situ hybridization (FISH): A technique that uses fluorescent probes to visualize specific DNA sequences on chromosomes

Biochemical assays: Tests that measure the levels or activity of specific proteins or enzymes in blood or other bodily fluids

What is short range?

In genetics, "short-range" typically refers to genetic linkage between two genes that are physically close to each other on the same chromosome. When two genes are closely linked, they are less likely to be separated by recombination during meiosis, and are more likely to be inherited together as a "linkage group". Short-range linkage can be an important tool for genetic mapping and identifying the location of disease-causing genes.

Applications of molecular markers:

Molecular markers are DNA sequences that can be used to identify genetic variation within and between populations. Some common applications of molecular markers include:

Genetic mapping: The use of molecular markers to identify the location of genes on a chromosome

Population genetics: The study of genetic variation and evolutionary processes within and between populations

Forensic genetics: The use of DNA profiling to identify individuals in criminal investigations or paternity testing

Plant and animal breeding: The use of molecular markers to identify desirable traits in crops or livestock

Medical genetics: The use of molecular markers to diagnose and predict the risk of genetic diseases.

1Principle of PCR.

2 how mendal perform plant experiments and why?

3 addition mutation.

4 causes of non disjunction.

5 point mutation Transversion and transition.

Principle of PCR:

The polymerase chain reaction (PCR) is a laboratory technique used to amplify specific DNA sequences. The principle of PCR involves repeated cycles of DNA denaturation, primer annealing, and DNA extension by a thermostable DNA polymerase. This process results in exponential amplification of the target DNA sequence, allowing for detection and analysis of very small amounts of DNA.

How Mendel performed plant experiments and why?

Gregor Mendel was a monk and scientist who performed experiments with pea plants in the mid-1800s. He conducted hybridization experiments by cross-breeding different varieties of pea plants and observing the traits of their offspring. He carefully recorded the phenotypic ratios of the offspring and used statistical analysis to infer the underlying genetic principles. Mendel chose pea plants for his experiments because they are easy to cultivate, have a short generation time, and have easily observable traits that can be easily manipulated.

Addition mutation:

An addition mutation is a type of mutation that involves the insertion of one or more nucleotides into a DNA sequence. This can result in a frameshift mutation, in which the reading frame of the DNA sequence is altered and can lead to significant changes in the amino acid sequence of the resulting protein.

Causes of non-disjunction:

Non-disjunction is the failure of homologous chromosomes or sister chromatids to separate properly during meiosis or mitosis, resulting in an abnormal distribution of chromosomes in the daughter cells. The causes of non-disjunction can include errors in chromosome pairing, errors in spindle formation or function, and errors in the cohesion or separation of sister chromatids.

Point mutation: Transversion and transition:

Point mutations are single nucleotide changes in a DNA sequence. They can be further classified into two categories: transitions and transversions. Transitions are point mutations in which one purine (A or G) is replaced by the other purine, or one pyrimidine (C or T) is replaced by the other pyrimidine. Transversions are point mutations in which a purine is replaced by a pyrimidine or vice versa.

1.Name DNA quantification methods.

2.point mutation.

3. Polyploidy.

4.x-linked recessive diseases.

DNA quantification methods:

There are several methods for quantifying DNA, including:

Spectrophotometry: Measures the absorbance of DNA at a specific wavelength, which correlates with DNA concentration. The most common wavelength used is 260 nm.

Fluorometry: Uses fluorescent dyes that bind to DNA and emit fluorescence when excited with a specific wavelength of light.

Gel electrophoresis: Allows for estimation of DNA concentration based on band intensity relative to known standards.

Polymerase chain reaction (PCR): Can be used to amplify a specific DNA fragment and thereby estimate the concentration of that fragment in a sample.

Point mutation:

A point mutation is a type of mutation that involves a change in a single nucleotide base pair in the DNA sequence. Point mutations can be further classified into three types:

Silent mutations: Do not result in any change to the amino acid sequence of the resulting protein.

Missense mutations: Result in a different amino acid being incorporated into the protein sequence, which can alter protein function.

Nonsense mutations: Result in a premature stop codon in the protein sequence, which can lead to a truncated and nonfunctional protein.

Polyploidy:

Polyploidy is a condition in which an organism has more than two sets of chromosomes in its cells. Polyploidy can occur naturally or as a result of errors during cell division, such as failure of chromosomes to separate during meiosis. Polyploidy can have a wide range of effects on the phenotype of an organism, including increased size, altered morphology, and altered reproductive characteristics.

X-linked recessive diseases:

X-linked recessive diseases are genetic disorders caused by mutations in genes located on the X chromosome. Because females have two X chromosomes, they can be carriers of X-linked recessive mutations without showing symptoms. Males, who only have one X chromosome, are more likely to exhibit symptoms of X-linked recessive diseases if they inherit a mutant allele. Examples of X-linked recessive diseases include hemophilia, Duchenne muscular dystrophy, and red-green color blindness.

What are mutagens, what are chemical and physical mutagenes.

Mutagens are agents that can cause changes in the DNA sequence of an organism, leading to mutations. Mutations can have a wide range of effects, ranging from no effect to causing disease or altering the phenotype of an organism.

Chemical mutagens are compounds that can react with DNA or alter the cellular environment, leading to DNA damage or errors in DNA replication. Examples of chemical mutagens include alkylating agents, such as ethyl methanesulfonate (EMS) and nitrogen mustards; base analogs, such as 5-bromouracil and 2-aminopurine; and intercalating agents, such as ethidium bromide and acridine orange. Chemical mutagens can also include reactive oxygen species (ROS) and other free radicals that can damage DNA.

Physical mutagens are agents that can cause DNA damage or errors through physical means, such as radiation or temperature changes. Examples of physical mutagens include ionizing radiation, such as X-rays and gamma rays; ultraviolet (UV) radiation; and high-energy particles, such as cosmic rays and alpha particles. Physical mutagens can also include extreme temperatures, such as heat or cold, and high pressure.

Both chemical and physical mutagens can have a wide range of effects on DNA and can cause different types of mutations, including base substitutions, deletions, insertions, and rearrangements. The mutagenic potential of a particular agent depends on a variety of factors, including the type and dose of the agent, the target organism or cell type, and the specific DNA sequences that are affected.

Define suppressor mutation,intragenic and intergenic mutation.

Suppressor mutation: A suppressor mutation is a type of mutation that can reverse the effects of another mutation, often by restoring the original function of a gene or protein. A suppressor mutation can occur at the same site as the original mutation (intragenic suppressor) or at a different site (intergenic suppressor).

Intragenic mutation: An intragenic mutation is a type of mutation that occurs within a single gene. This type of mutation can result in changes to the amino acid sequence of the encoded protein, leading to altered or lost protein function. Intragenic mutations can include missense mutations, nonsense mutations, and frame-shift mutations, among others.

Intergenic mutation: An intergenic mutation is a type of mutation that occurs outside of a gene or in a non-coding region of DNA. Intergenic mutations can affect the regulation of gene expression or other functions, such as DNA replication or repair. Some examples of intergenic mutations include promoter mutations, enhancer mutations, and mutations in regulatory regions or non-coding RNAs. Intergenic mutations can also have effects on nearby genes, as they can alter the local chromatin environment or affect the binding of transcription factors or other regulatory proteins.

Continues and discontinues mutation.

Continuous mutation and discontinuous mutation are two different ways to describe the effect of a mutation on the resulting protein.

Continuous mutation: A continuous mutation is a type of mutation that results in a minor change to the amino acid sequence of a protein. This type of mutation is also known as a missense mutation. In a missense mutation, a single nucleotide change in the DNA sequence leads to the substitution of one amino acid for another in the resulting protein. The effect of a continuous mutation can be mild or negligible, as the substituted amino acid may have similar properties to the original amino acid.

Discontinuous mutation: A discontinuous mutation is a type of mutation that results in a major change to the amino acid sequence of a protein. This type of mutation is also known as a nonsense mutation or a frame-shift mutation. In a nonsense or frame-shift mutation, one or more nucleotides are added, deleted, or altered in the DNA sequence, leading to a shift in the reading frame and a truncated or nonfunctional protein. The effect of a discontinuous mutation is usually more severe than a continuous mutation, as it can result in the loss of protein function or the production of a nonfunctional protein.