BIO301 _ Subjective Final Terms (Complete)
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1)DrawPunnet Square for a Cross between Homozygous Short Hairs(H) and Homozygous long hairs And Give their Genotype and Phenotype

	H	**H**
h	**Hh**	Hh
h	**Hh**	Hh

Genotype : Hh (100 percent) in F1

Phenotype : Short haired 100 percent (heterozygous dominant)

2)Difference between DNA & cDNA

DNA and cDNA are two types of nucleic acids. The DNA is the genetic material of most of the organisms. It is arranged inside the nucleus in eukaryotes. ... The main difference between DNA and cDNA is **that DNA is composed of both coding and non-coding sequences whereas cDNA only contains the coding sequences**

3)SNP

A single nucleotide polymorphism, or SNP (pronounced "snip"), is a variation at a single position in a DNA sequence among individuals. ... If a SNP occurs within a gene, then the **gene is described as having more than one allele**.

4)MUTATION AT END OF INTRON

The number of the first nucleotide of the following exon, a minus sign and the position upstream in the intron, like c.89-1G>T

5)Northern Blotting

A northern blot is **a laboratory method used to detect specific RNA molecules among a mixture of RNA**. Northern blotting can be used to analyze a sample of RNA from a particular tissue or cell type in order to measure the RNA expression of particular genes.

5)Southern Blotting

A Southern blot is **a laboratory method used to detect specific DNA molecules from among a many other DNA molecules**. The technique was named after its inventor, Edward Southern.

6)Western Blotting

A western blot is **a laboratory method used to detect specific protein molecules from among a mixture of proteins**. ... Next, the protein molecules are separated according to their sizes using a method called gel electrophoresis. Following separation, the proteins are transferred from the gel onto a blotting membrane

7)PCR Principle

Its principle is **based on the use of DNA polymerase** which is an in vitro replication of specific DNA sequences. This method can generate tens of billions of copies of a particular DNA fragment (the sequence of interest, DNA of interest, or target DNA) from a DNA extract (DNA template).

8)DIAGNOSTIC TESTS FOR GENETIC DISORDERS

PHYSICAL ANALYSIS

Physical analysis for affected individual.

ULTRASONOGRAPHY OF FETUS

Ultrasonography of fetus determines malformations of head, internal organs, extremities.

ANALYSIS OF AMNIOTIC FLUIDS

Amniotic fluid analysis to determine genetic and chromosomal disorders after 14 wks gestation.

More than 200 various genetic diseases easily be detected.

MATERNAL BLOOD ANALYSIS

Maternal blood analysis to observe abnormal fetus.

9)Short tandem repeat sequences (STR)

STR is simple repeats of nucleotide sequences.

Mononucleotide, dinucleotide .

Used in linkage analysis Used in DNA forensics.

10)Complications to basic inheritance pattern

Complications in basic inheritance pattern Single

genes -Non-mendelian inheritance .

Differences in gene actions can generate more complex inheritance patterns. Single gene can give codominance, incomplete dominance, overdominance.

Complications to inheritance patterns

Multiple genes - epistasis, polygenic traits Genes & the environment -

sexinfluenced traits, incomplete penetrance Complications

- A common recessive inheritnace can give dominant pattern
- Autosomal dominant inheritance with non-penetrance • Autosomal dominant inheritance with variable expression.
- Maternal inheritance of mt DNA
- Maternal inheritance of mitochondrial DNA •

11) Mitochondrial DNA Inheritence

(MTDNA) is inherited through ovum not sperm Mother

could pass it to all children.

- Father will pass it to none of the children.

Mitochondrial DNA

 More than one copy of mtDNA is passed.

 Mutations are common.

 More than one type of mtDNA genome is passe

Properties of Mitochondrial inheritance

 Heteroplasy

 Variable expression

 Pleiotropy

 Reduced penetrance

 Functional somatic mosaic

Common mitochondrial diseases

 Myoclonic epilepsy

 Mitochondrial recessive ataxia

 Leber hereditary optic neuropathy

 Sensory ataxia neuropathy

12)Why Seeds in F1 Generation are yellow ,when True Breeding Yellow is Crossed with True Breeding Green Plant?

F1 Generation consists of 100 percent Yellow coloured offsprings because of the Dominance effect ,as the yellow colour allele is dominant over Green Colour Allele .So yellow Colour is evident in F1 Generation ,though all offsprings are heterozygous.

13)Quantification of DNA and RNA (Nucleic Acids)

Various techniques are employees for quantitative analysis of DNA .DNA Quantification gives about DNA fragments length usually in kb units.

These may include

- SDS_PAGE (Sodium Dodecyle Sulphate _Polyacrylamide Gel Electrophoresis
- UV Spectrophotometry
- Organic Method of Phenol Cloroform

Etc

14)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

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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

15)Polymorphism - why to study

Essential to study inheritance patterns. Map phenotypes and anchor genes to the genetic map by co- segregation analysis. Determine change in function: resistant/sensitive populations genetically determined variability among humans is due to a difference in 0.1% of the genomic sequence.
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16)Types of polymorphism

Types of Polymorphic DNA Sequences

 RFLP: Restriction Fragment Length Polymorphism

 VNTR: Variable Number Tandem Repeats (8 to > 50 base pairs)

Types of Polymorphic DNA Sequences

 STR: Short Tandem Repeats (1– 8 base pairs).

 SNP: Single Nucleotide Polymorphisms

17)Structural Abnormalities

Chromosome breakage & subsequent reunion in a different configuration

Types based on genetic material

 Balanced – chromosome complement is complete.

 Unbalanced – when there is incorrect amount of genetic material.

Types

 Translocations

 Deletions

 Insertions Inversions

 Ring chromosome

 Isochromosomes

18)Epistasis

Alleles at one gene locus can hide or prevent expression of alleles at second gene locus.

Example

Labrador retrievers one gene locus affects coat color by controlling pigment eumelanin deposited in the fur.

A dominant allele B for black, recessive allele b brown coat, a second gene (e) locus controls eumelanin deposited in fur. Dogs homozygous recessive at this locus (ee) will have yellow fur no matter which alleles are at first locus. Alleles at one gene locus can hide or prevent expression of alleles at second gene locus is called as epistatsis.

19)Pleiotropy

Pleiotropy

 The ability of a gene to affect an organism in many ways is called pleiotropy.

 Pleiotropy refers to an allele which has more than one effect on the phenotype.

 This phenomenon occurs when a single gene locus produces more than one traits.

Pleiotropic effects in humans

Example:

Sickle cell anemia

Cystic fibrosis

Huntington disease

 One gene – more than one phenotypes

19)What is Epigenetics

Epigenetics literally means ‘above’ the genetics. Epigenetic trait is a stable and heritable phenotype resulting from changes in chromosome without alterations in the DNA sequence.

Causes of Epigenetics

DNA Methylation is epigenetics, not changes in DNA sequence

DNA methylations are regarded as epigenetics and not genetic changes in sequence of DNA.

Molecular Mechanism of Epigenetics Two primary mechanisms identified. Methylation of cytosine

Posttranslational modification to histone proteins; includes acetylation,

phosphorylation etc. Molecular Mechanisms

Histones modifications

Small interfering RNA – mechanism causing epigenetics A third mechanism involves expression of small interfering RNAs (siRNA).

20)Total RNA extraction using Trizol reagent

1. Homogenization
2. **PHASE SEPARATION.** Incubate the homogenized samples for 5 minutes at 15 to 30°C to permit the complete dissociation of nucleoprotein complexes. Add 0.2 ml of chloroform per 1 ml of TRIZOL Reagent. Cap sample tubes securely. Shake tubes vigorously by hand for 15 seconds and incubate them at 15 to 30°C for 2 to 3 minutes. Centrifuge the samples at no more than 12,000xg for 15 minutes at 2 to 8°C. Following centrifugation, the mixture separates into a lower red, phenol-chloroform phase, an interphase, and a colorless upper aqueous phase. RNA remains exclusively in the aqueous phase. The volume of the aqueous phase is about 60% of the volume of TRIZOL Reagent used for homogenization.
3. **RNA PRECIPITATION.** Transfer the aqueous phase to a fresh tube, and save the organic phase if isolation of DNA or protein is desired. Precipitate the RNA from the aqueous phase by mixing with isopropyl alcohol. Use 0.5 ml of isopropyl alcohol per 1 ml of TRIZOL Reagent used for the initial homogenization. Incubate samples at 15 to 30°C for 10 minutes and centrifuge at no more than 12,000xg for 10 minutes at 2 to 8°C. The RNA precipitate, often invisible before centrifugation, forms a gel-like pellet on the side and bottom of the tube.
4. **RNA WASH.** Remove the supernatant. Wash the RNA pellet once with 75% ethanol, adding at least 1 ml of 75% ethanol per 1 ml of TRIZOL Reagent used for the initial homogenization. Mix the sample by vortexing

and centrifuge at no more than 7,500xg for 5 minutes at 2 to 8°C. 5. **REDISSOLVING THE RNA.** At the end of the procedure, briefly dry the RNA pellet (air-dry or vacuum-dry for 5-10 minutes). Do not dry the RNA by centrifugation under vacuum. It is important not to let the RNA pellet dry completely as this will greatly decrease its solubility. Partially dissolved

RNA

6. samples have an A260/280 ratio < 1.6. Dissolve RNA in RNase-free water or 0.5% SDS solution by passing the solution a few times through a pipette tip, and incubating for 10 minutes at 55 to 60°C. (Avoid SDS when RNA will be used in subsequent enzymatic reactions.) RNA can also be redissolved in 100% formamide (deionized) and stored at -70°C (5).

21)Somatic and Germline Mutation

Somatic and Germline mutation

Somatic mutation: Arise in the somatic cells. Passed on to other cells through the process of mitosis.Effect of these mutations depends on the type of the cell in which they occur.

Germline mutation: They occur in the cells that produce gametes. This type of mutation usually passed on to future generation. In multicellular organisms, the term mutation is generally used for germ line mutations.

22)Effect of Alteration of Single Nucleotide

Even a single Nucleotide alteration is is important because it can cause changes in type of protein ,encoded for.

23)Mutations at Chromosome level

Variationn in Chromosome Number

Aneuploidy

Hyperploidy, Hypoploidy

Euploidy

Monoploidy, Diploidy, Polyploidy

 Variation in chromosome structure

Deletions

Translocations

Duplications

Inversions

Mutations and effects

24)Major types of mutations

 Points Mutations

 Variations in chromosomes number

 Variations in chromosomes structure

25)**Point mutations:**

• Point mutations affect single sites on DNA.

• Substitution of one base for another.

• Deletion/addition of a single base or small number of bases.

Transition: If purine (A/G) or pyrimidine (T/C) substitutes for itself = transition substitution

Transversion: If purine substitutes for pyrimidine or vice versa = transversion substitution

Results of point mutations

 Silent mutations

 Missense mutations

 Nonsense mutations

Silent Mutation: These types of mutations have affected the DNA sequence but not the protein.

They have no effect on the organism's phenotype. Since the genetic code is degenerate, several codons produce the same amino acid. Third base changes often have no effect on the amino acid sequence of the protein.

Missense mutation: Missense mutations substitute one amino acid for another. Some missense mutations have very large effects. Some have minimal or no effect. It depends on where the mutation occurs in the protein's structure and how big a change in the type of amino acid it is.

Missense mutation produces a change in amino acid sequence in protein product (e.g. Histidine in for

Arginine). May change function of protein or may not!

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 non- functional proteins. Nonsense mutation: Nonsense mutations convert an amino acid into a stop codon. The effect is to

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26)Positional cloning

- Disease gene locus is mapped with the help of marker.
- Genes in candidate region are screened by sequencing to identify mutation.
- Protein of identified gene is studied.

27)Facial Hair And Eye Colur Dominant Allele colours

The allele genes come in the form of brown, blue, or green, with brown being dominant, followed by green, and blue being the least dominant or what is called recessive. ... **Blue will always be recessive**. If both parents have a blue allele, it is likely that the child will have blue eyes.

scientists have found that a marker in the *LNX1* gene on chromosome 4 plays a role in facial hair growth. Specifically, variations in this gene seem to influence how thickly (or sparsely) your facial hair grows.

Other genes are likely involved as well. And as scientists learn more about the genetics of beard growth, they may find a better predictor for facial hair thickness.

28)VNTR

 VNTR are short nucleotide sequences organized as a tandem repeats on genomes.

 Found on many chromosomes

 Show variations in length between individuals.

Size of VNTR

 The size of repeat is about 7 to few tens of base pairs long.

Each variant acts as inherited allele

 Each variant acts as an inherited allele, allowing them to be used for personal or parental identification.

VNTR

 The rectangular blocks represent each of the repeated DNA sequences.

 The repeats are tandem - they are clustered together and oriented in the same direction.

VNTR - Methodology

- Restriction enzymes are used to cut the VNTR blocks from its specific sites and analyzed by RFLP.

- Blocks (VNTR) are amplified by polymerase chain reaction.
- Amplified products are run on gel electrophoresis which also determined its length.

Importance of VNTR

VNTRs are an important source of RFLP genetic markers used in linkage analysis (mapping) of genomes. When removed from surrounding DNA by the PCR or RFLP methods, and their size determined by gel electrophoresis or Southern blotting. As a result, they produce a pattern of bands unique to each individual.

Basic genetic principles during VNTR data analysis

Identity Matching: VNTR alleles from a specific location must match. If two samples are from the same individual - same allele patter

Inheritance Matching: Matching an individual with his/her parents or children, a person must have an allele that matches one from each parent.

Applications of VNTR

- Microbiology
- DNA fingerprinting
- Genetic diversity
- Forensics
- Mapping of genomes
- Breeding patterns of wild or domesticated animals

29)Mutation nomenclature

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 gene nomenclature committee) gene symbols should be used (www.genenames.org)

 DNA reference sequence used should preferably be from the RefSeq database, listing both

database accession and version number (like NM_004006.2)

Salient feature of Mutation nomenclature

There is no nucleotide 0 (zero).

 Nucleotide 1 is A of the ATG-translation initiation codon. Salient feature of Mutation nomenclature

There is no nucleotide 0 (zero).

 The nucleotide 5' of the ATG-translation initiation codon is -1, the previous -2, etc. e.g. in 5’

UTR c.-12G>A

 The nucleotide 3' of the translation stop codon is *1, the next *2, etc. e.g. in 3’ UTR c.*70T>A

 Nucleotide numbering is purely arbitrary and starts with 1 at the first nucleotide of the database reference file.

 No +, - or other signs are used.

 The sequence should include all nucleotides covering the sequence (gene) of interest and should start well 5' of

the promoter of a gene.

30)Heterogenous Dominant trait

Heterogenous Dominant trait is one which has both dominant and recessive alleles for that trait but only dominant succeeeds to express itself over the recessive .

Eg Brown Eyecolour ,Black hair colour ,both these colours are dominant over other shades .

31)Frequency of autosomal dominant disorders

Frequency of autosomal dominant disorders varies between 3 – 9.5 per 1000 individuals

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32)RAPD and AFLP

Random Amplified Polymorphic DNA

&

Amplified Fragment Length Polymorphism

Random Amplified Polymorphic DNA

- PCR based marker with 10-12 base pairs
- Random amplification of several fragments
- Amplified fragments run in agarose gel detected by ethidium bromide.
- Unstable amplification leads to poor repeatability

Amplified Fragment Length Polymorphism

- Restriction endonuclease digestion of DNA
- Ligation of adaptors
- Amplification of ligated fragments
- Separation of the amplified fragments via electrophoresis and visualization
- AFLPs have stable amplification and good repeatability.

33)pedigree drawing/ analysis

A complete, detailed family history is called a pedigree. Pedigrees are used to determine the pattern of inheritance of a genetic disease within a family.

34)Genetic disorders

- A genetic disease or disorder appear as a result of mutations in an individual's DNA.

 Change in DNA sequence may be small or large like deletions/addition of entire chromosome.

Classification of Genetic Disorders

Fully penetrant and low penetrant

 Fully penetrant, where other genes and environmental factors have no effect.

 Low-penetrance, genes play a small part, along with other environmental factors.

Classification of genetic disorders

 Multifactorial disorders

 Single gene disorders

 Chromosomal disorders

 Mitochondrial disorders

 Somatic mutations (cancers)

Inheritance patterns of genetic disorders

Autosomal Dominant: Affected individuals are heterozygote. One allele is mutated, while other one is normal.

Autosomal recessive: Homozygous, when both alleles of the gene are mutated.

X-linked recessive: Mutated alleles are present on either of the sex chromosomes.

Multifactorial: Genes and environment both contribute.

Multifactorial frequent, single gene disorders less frequent: Multifactorial are common. Single

gene less common like dominant/recessive pedigree patterns

35)Multifactorial diseases:

Genetic disorders controlled by many genes plus the effects of the environment.

Multifactorial diseases

- Asthma
- Cancers
- Ciliopathies
- Cleft palate
- Diabetes
- Heart diseases
- hypertension inflammatory
- bowel disease intellectual
- disability mood disorder
- Obesity infertility
-

36)Types of Penetrance

Complete Penetrance The allele is said to have complete penetrance if all individuals who have the disease-causing mutation have clinical symptoms of the disease.

Highly Penetrant If an allele is highly penetrant, and then the trait it produces will almost always be apparent in an individual carrying the allele.

Incomplete or reduced Penetrance is said to be reduced or incomplete when some individuals fail to express the trait, even though they carry the allele. Incomplete or reduced Penetrance

Low Penetrance An allele with low penetrance will only sometimes produce the symptom or trait with which it has been associated at a detectable

level. In low penetrance, it is difficult to distinguish environmental from genetic factors.

Determine Penetrance is difficult – if age related Penetrance can be difficult to determine reliably, even for genetic diseases that are caused by a single polymorphic allele.

For many hereditary diseases, the onset of symptoms is age related.

- Penetrance is expressed as frequency
- Multiple endocrine neoplasia 1 (MEN 1), a hereditary disorder
- Age related – 7% at age 10, 100% at age 60.

37)Loss of Mutation

If a mutation occurs in that allele, the function for which it encodes is also lost. The general term for these mutations is loss-of-function mutations. The degree to which the function is lost can vary. If the function is entirely lost, the mutation is called a **null mutation**.

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

38)Function of Trizol in RNA Extraxtion

TRIzol® Reagent **maintains the integrity of the RNA** due to highly effective inhibition of RNase activity while disrupting cells and dissolving cell components during sample homogenization.

39)The Role of $MgCl_2$ in PCR reaction

is **to enhance the DNA amplification by boosting the activity of Taq DNA polymerase**. The polymerase chain reaction is one of the important experimental procedure in downstream genomic and genetic research. It generates millions of amplicon for the gene of our interest.

40)Role of EDTA IN DNA Extraction

The EDTA works as **a chelating agent** in the DNA extraction. It chelates the metal ion present into the enzymes and as we all know that the metal ions are the cofactor which increases the activity of the enzyme. By chelating the metal ions, it deactivates the enzyme, therefore, reduces the activity of DNase and Rnase

41) **short range Applications of molecular markers –**

- Parentage determination
- Genetic distance estimation
- Determination of twin zygosity
- Sexing of pre-implanted embryos
- Identification of disease carriers

Applications of Molecular Markers –Long Range

- Gene mapping & mapping of QTL by linkage
- Markers assisted selection of breeds

42)Reagents Used in PCR

In general, a complete PCR reaction requires five basic PCR reagents; **DNA/RNA template, DNA polymerase, primers (forward and reverse), deoxynucleotide triphosphates (dNTPs) and PCR buffers**.

43)Types of Mutation

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

44)Mutations alter Gene Function

How mutations alter gene function

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.
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- Dominant negative-antagonizes normal product
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Predicting that a gene product won’t do the job

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- Is it non-conservative
- Is AA conserved in evolution

45)Exons and Introns

The protein coding regions regions of RNA are called Exons ,while non coding regions of RNA are called Introns .

46)Mutation and it's Effects

: In some disciplines the term "mutation" is used to indicate "a change" while in other disciplines it is used to indicate "a disease-causing change".

Sequence/ allelic variant some geneticists do not use term mutation. They use neutral terms like

"sequence variant", "alteration" and "allelic variant "Mutations occurs at a frequency of about 1 in every 1 billion base pairs. Every individual has about 6 mutations in each cell in their body.

Effect of mutation:

- Mutations may be harmful.
- Mutations may be beneficial.
- Mutations may have no effect on the organism..

Mutations are a major source of genetic variation in a population increasing biodiversity. Some variations may help to survive better and longer. Mutations in gametes (egg & sperm) are passed onto offspring. Mutations in body cells only affect the organism in which they occur and are not passed onto offspring.

47)Aneuploidy

Nullisomy

Monosomy

Trisomy

Tetrasomy

Monosomy:

Loss of a single chromosome

Monosomy of autosomes is lethal

Turner syndrome XO i.e. loss of sex chromosome.

Causes of monosomy: followings are the causes of monosomy

 Non disjunction – one gamete receives two copies of homologous chromosomes and other will have no copy.

 Loss of chromosome. As it move towards pole of cell during anaphase.

Trisomy is a type of polysomy in which there are three copies of a particular chromosome, instead

of the normal two. Trisomy is a type of aneuploidy (an abnormal number of chromosomes).

Trisomy -Autosomes and sex chromosome

 Autosomal trisomy

 Sex-chromosome trisomy

 Trisomy of chromosome 21, which is found in Down syndrome, is called trisomy 21.

Trisomy Types

 Primary trisomy - an entire extra chromosome has been copied.

 Partial trisomy - an extra copy of part of a chromosome.

 Secondary trisomy - the extra chromosome has duplicated arms.

 Tertiary trisomy - extra chromosome is made up of copies of arms from two other chromosomes. Common trisomy in humans

 Trisomy 21

 Trisomy 18

 Trisomy 13

 Klinefelter's syndrome (47 XXY)

Trisomy 21 Down syndrome Trisomy 21 1 in 700 live births Associated with increased maternal age

Polyploidy:

Multiples of haploid number

• Triploidy or tetraploidy etc

• Fetus does not survive

Two major types

 Autopolyploidy

 Allopolyploidy

Types of polyploidy

 Triploid (3x) watermelon

 Tetraploid (4x) - cotton

 Pentaploid (5x) - Kenai Birch

 Hexaploid (6x) – wheat

48)Expressivity

Extent to which allele is expressed at phenotypic level affected by genetic background and environment.

Difference likelihood of the gene generating its associated phenotype.

Influence of an expressed gene at the level of particular individuals.

Expressivity

The term is analogous to the severity of a condition in clinical medicine.

Variable expressivity when phenotype is expressed to a different degree among individuals with the same genotype.

49)Organic Method of DNA Extraction

- Cells which are to be studied need to be collected.
- Breaking the cell membranes open to expose the DNA along with the cytoplasm within (cell lysis).

    - Lipids from the cell membrane and the nucleus are broken down with detergents and surfactants.
    - Breaking down proteins by adding a protease (optional).
    - Breaking down RNA by adding an RNase (optional).
- The solution is treated with a concentrated salt solution (saline) to make debris such as broken proteins, lipids and RNA clump together.
- Centrifugation of the solution, which separates the clumped cellular debris from the DNA.
- DNA purification from detergents, proteins, salts and reagents used during the cell lysis step. The most commonly used procedures are:
    - Ethanol precipitation usually by ice-cold ethanol or isopropanol. Since DNA is insoluble in these alcohols, it will aggregate together, giving a *pellet* upon centrifugation. Precipitation of DNA is improved by increasing of ionic strength, usually by adding sodium acetate.
    - Phenol–chloroform extraction in which phenol denatures proteins in the sample. After centrifugation of the sample, denatured proteins stay in the organic phase while the aqueous phase containing nucleic acid is mixed with chloroform to remove phenol residues from the solution. ◦ Minicolumn purification that relies on the fact that the nucleic acids may bind (adsorption) to the solid phase (silica or other) depending on the pH and the salt concentration of the buffer.

Cellular and histone proteins bound to the DNA can be removed either by adding a protease or by having precipitated the proteins with sodium or ammonium acetate, or extracted them with a phenolchloroform mixture prior to the DNA-precipitation.
After isolation, the DNA is dissolved in a slightly alkaline buffer, usually in a TE buffer, or in ultra-pure water.

50)Difference Between Non Organic and Organic Method of DNA Extraction

Non-Organic DNA Extraction. •h **Does not use organic reagents** such as. phenol orh chloroform. • Digested proteins are removed by salting. out with high concentrations of LiCl.

Organic extraction involves the **addition of and incubation in multiple different chemical solutions**; including a lysis step, a phenol chloroform extraction, an ethanol precipitation, and washing steps. Organic extraction is often used in laboratories because it is cheap, and it yields large quantities of pure DNA.

Applications of PCR

The applications of the polymerase chain reaction

The polymerase chain reaction (PCR) allows the in vitro amplification of DNA fragments starting with tiny amounts of biological sample and oligonucleotide primers derived from sequence data. Since the technique is fast and easy, PCR has taken the DNA-technology to the routine laboratoria. We present a survey of the following applications of PCR: 1) The amplification of gene fragments as fast alternative of cloning. 2) The modification of DNA fragments. 3) The sensitive detection of pathogenic microorganisms, if desired followed by an accurate genotyping. 4) DNA analysis of arachaeological specimens. 5) The detection of mutations relevant for inherited diseases, malignant transformation or tissue typing. 6) The analysis of genetic markers for forensic applications, for paternity testing and for the mapping of hereditary traits. 7) The species-specific amplification of DNA segments between interspersed-repeat elements. 8) The study of gene expression.

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MSc Zoology