NAME :	WAQAR KHAN
ID:	14650
DEPERTMENT:	BS MICROBIOLOGY 41H
SEMESTER :	41H
COURSE TITLE:	FUNDAMENTEL GENETICS
FINAL:	TERMS ASSIGMENT (SUMMER 2020)
INSTRUCTOR:	MR. FAZLI ZAHIR MAIN
DATE :	2319/2020

Q No. 1: Fill in the BLANKS.

1) Gene that normally block cell cycle procession are known as **TUMOR SUPPRESSORS**

2) **NON DISJUNCTION** in the failure of chromosome to separate during meiosis.

3) one of the most important tumor suppressor is **TUMOR** *PROTEINE P53*

4) The overactive (cancer- promoting) from of these gene are called **ONCOGENES** while the normal. Not – yet – mutated forms are called **PROTO_ONCOGENES**

Q.2: Write short note on the following

1) Heredity AND VERIATION

ANs: Heredity may be define as the transmission of characters from one generation to successive generations or from parents to their offspring.

Example:

Heredity is a large proportion of genes in baker's yeast are also present in humans. This similarity in genetic makeup between organisms that have such.

Variation:

ANS: The visible differences between the parents and the offspring or between the off spring or between the offspring of the same parents is called variation.

Example:

A person's skin colour, hair colour dimples, freckles, and blood type are all examples of genetic variations that can occur in a human population.

4). Chromosomal theory of inheritance:

ANS: The Boveri-Sutton chromosome theory also known as the chromosome theory of inheritance or the Sutton Boveri theory is a fundamental unifying theory of genetics which identifies chromosomes as the carriers of genetic material.

The chromosome theory of inheritance, or the idea that genes are located on chromosomes, was proposed based on experiments by Thomas Hunt Morgan using Drosophila melanogaster, or fruit flies.

Key points of the theory;

Thomas Hunt Morgan, who studied fruit flies, provided the first strong confirmation of the chromosome theory.

Morgan discovered a mutation that affected fly eye colour. He observed that the mutation was inherited differently by male and female flies.

Based on the inheritance pattern, Morgan concluded that the eye colour gene must be located on the X chromosome.

2). Sex chromosome linked disease in human:

ANS: Disease like haemophilia, colour blindness, male pattern of baldness are sex linked disease.

Where colour blindness and haemophilia are X-linked diseases, male pattern of baldness is a Y-linked one.

This indicates that the X-linked disease will express themselves in a male whereas the female is always a carrier until both the genes are recessive in the female.

Male pattern of baldness being a Y-linked trait expresses itself only in the males while females are never affected by it.

3). Biological significance of Mendel's Laws:

ANS: "Mendel's work remained buried for about three decades, but after its rediscovery, the laws are being used for the various branches of breeding.

These are use for improving the varieties of fowls and their eggs; in obtaining rust-resistant and disease-resistant varieties of grains. Various new breeds of horses and dogs are obtained by cross breeding experiments.

The science of Eugenics is the outcome of Mendelism which deals with the bet treatment of human race".

Q.NO3: Discuss in detail sex chromosomes and X linked inheritance? ANS:

Introduction:

If you're a human being which seems like a good bet, most of your chromosomes come in homologous pair.

The two chromosomes of a homologous pair contain the same basic information- that is the same genes in the same order- but may carry different versions of those genes.

Key points:

In human and other mammals, biological sex is determined by a pair of sex chromosomes: XY in males and XX in females.

Genes on the X chromosome are said to be x-linked. X-linked genes have distinctive inheritance patterns because they are present in different numbers in female(xx) and males(xy)

X-linked human genetic disorders are much more common in males than in females due to the Y-linked inheritance pattern.

Sex chromosome in human:

Human X and Y chromosomes determine the biological sex of a person, with XX specifying female and XY specifying male. Although the Y chromosome contains a small region of similarity to the X chromosome so that they can pair during meiosis, the Y chromosome is much shorter and contain many fewer genes.

Sex chromosomes in human:

The human Y chromosome plays a key role in determining the sex of a developing embryo. This is mostly due to a gene called SRY "Sex determining region of Y".

SRY is found on the y chromosome and encodes a protein that turns on other genes required for male development.

XX embryos don't have SRY, so they develop as female.

XY embryos do have SRY, so they develop as male.

X Linked genes:

When a gene being is present on the X chromosome, but not on the Y chromosome, it is said to be x-linked.

X-linked genes have different inheritance patterns than genes on nonsex chromosomes.

That's because these genes are present in different copy numbers in males and females.

Example: Drosophila

X-linked genetic disorders:

The same principles we see at work in fruit flies can be applied to human genetics. In human, the alleles for certain conditions including some forms of colour blindness, hemophilia, and muscular dystrophy are X-linked.

These disease are much more common in men than they are in women due to their X-linked inheritance pattern .

Example:

A mother is heterozygous for disease-causing allele. Woman who are heterozygous for disease alleles are said to be carriers, and they usually don't display an y symptoms themselves.

Q No. 4: What is mitochondrial inheritance? How is non –nuclear DNA different from mitochondrial DNA? ANS:

Mitochondrial inheritance is also define in the DNA is also found in the mitochondria present in most plant and animal cells, as well as in the chloroplasts DNA are inherited.

The DNA molecule found in mitochondria are small and circular, much like the typical bacterium.

There are usually many copies of DNA in a single mitochondrion.

It is called the powerhouse of the cells.

EXAMPLES: They are children get mitochondrial DNA from their mother but not the from of Father.

HOW IS NON – NUCLEAR DNA DIFFERENT?

ANS: Here are some ways that mitochondrial DNA differ the found in the nucleus.

HIGH COPY NUMBER: A mitochondrion has multiple copies of its DNA and a typical cell has many mitochondria. As a result, cells usually have many copies- often thousand- of mitochondrial.

RANDOM SEGREGATION: Mitochondria is randomly distributed to daughter cells during mitosis and meiosis.

When the cells divides, the organelles that happen to be on opposite sides of the cleavage furrow or cell plate will end up in different daughter cells.

SINGLE- PARENT INHERITANCE: Non – nuclear DNA is often inherited uniparentally, meaning that offspring get DNA only from the Male and female parent, not both in humans.

For Example: Children get mitochondrial DNA from their mother but not their father.

MITOCHONDRIAL MUTATIONS AND HUMAN DISEASES

Mutations in mitochondria DNA can lead to human genetic disorders.

For Example

They are large deletion in mitochondrial DNA cause a condition called Kearns- slayer syndrome. These deletion keep up mitochondria from doing their job of extracting energy. Kearns-sayre syndrome can cause symptoms such as weakness of the muscles, including those that control eyelid and eye movements, as well as degeneration of the retina and development of heart disease.

MITOCHANDRIAL MUTATION IN HUMAN DISEASES

A person with a disease caused by a mitochondrial mutations may lack normal mitochondria and have only abnormal, mutations- bearing ones .in this case , an affected mother will always pass on mutation- bearing mitochondria to her children.

QNo.5: Explain in detail mutations and different types of mutations.

ANS: Mutation

DefinE: In the biology, a mutation is an alteration in the nucleotide sequences of the genome.

For Example: A butterfly may produce offspring with a new mutation.

OR

A mutation occurs when a DNA gene is damaged or changed in such away as to alter the genetic massage carried by that gene. A mutagen is an agent of substance that can bring about a permanent alteration to the physical composition of a DNA gene such that the genetic massage is changed. *For Example:* They are naturally occurring plants colour mutations. Photo credits: orange- forest Starr and Kim.

Mutation Helpful or Harmful.

Mutations happen regularly almost all mutations are neutral chemicals and UV radiation cause mutations many mutations are repaired by enzymes.

Some mutations can be harmful as well. Some type of skin cancers and leukaemia result from somatic mutations.

Some mutations may improve an organism survival beneficial.

Different types of mutations

Chromosome mutations

They are the five types in exist .

1) Deletion

Due to the breakage a piece of a chromosome is lost. **2) Inversion**

The chromosome segment breaks off segment flips around backwards segment heartaches.

3) Duplication

Occurs when a gene sequence is repeated.

4) Translocation

Involves two chromosomes that are not homologous part of one chromosome is transferred to another chromosomes.

5) Nondisjunction

Failure of chromosome to separate during meiosis causes gametes to have too many or too few chromosomes.

GENE MUTATIONS

Change in the nucleotide sequences of a gene many only involve a single nucleotide may be due to coping errors, chemical, viruses, etc.

They are the different types of gene mutations

POINT MUTATIONS

Change of the single nucleotide includes the Deletion, insertion, or substitution of one nucleotide in a gene .

Sickle cell disease is the result of one nucleotide substitution occurs in haemoglobin gene.

FREMESHFIT MUTATIONS

Inserting or detecting one or more nucleotide changes the " reading frame" like changing a sentence proteins built incorrectly. Frame shift a added. ATG GAA GCA CGT.