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"GENETICS AND HUMAN AFFAIRS: A LITERATURE REVIEW"

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ABSTRACT:

Genetics is the field of biology that studies how traits are passed from parent to their offspring; blueprint of life. The basic components of genetics are DNA, genes, chromosomes and genetic inheritance. Genetics is built around molecules called DNA. DNA molecules hold all genetic information for an organism. Gene is a particular section of DNA that tells cell to perform a specific task. Heredity makes offspring looks like their parents. During reproduction, DNA is replicated and passed from parent to offspring. The environment that an organism lives in can also influences how genes are expressed.

Keywords: Hereditary, DNA, Genome, Sequencing, Alleles, Locus, Phenotypes.

INTRODUCTION:

Genetics: The branch of biology that deals with heredity, especially the mechanism of hereditary transmission and the variation of inherited characteristic among similar or related organisms.

Genetics tries to identify which traits are inherited, and explains how these traits are passed from generation to generation.[1,2,3]

Gregor Johann Mendel is called as the 'Father of genetics' and the study of principles of heredity laid by Mendel is called Mendelism.

AIMS AND OBJECTIVES:

Aims: To study and apply the concepts of modern transmission and molecular genetics.

Objectives:

1) To study the basic structure and function of gene.
2) To study the gene behaviour in context of a cell or organism.
3) To study the pattern of inheritance and gene distribution.
4) To study the genetic disorder and gene therapy.

Basic components of genetics:

1) Gene:

It is biological unit of heredity. It consists of continuous segment of DNA. Gene holds the information to build and maintain their cells and pass the genetic traits to offspring.

In eukaryotes, gene occupies specific position on chromosome called as locus[4,5].

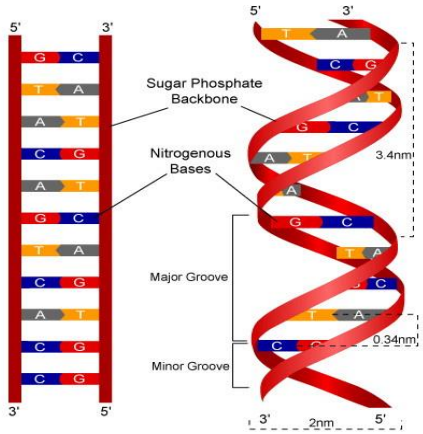

Fig. No.1: Structure of gene

2) Genome:

It is the collection of genetic information.

3) Chromosome:

The DNA molecule is packed into thread like structure called as chromosome. It is storage unit of gene.

Chromatin- DNA, RNA & protein that make up chromosome.

Chromatids- One of two identical parts of chromosome.

Centromere- It is point where two identical chromatids meets each other.

In human being;

Autosomes: 22 pairs

Sex chromosome: 1 pair[6,7,8].

4) Nucleotides:

Group of molecules that when linked together, forms the building blocks of DNA and RNA. Nucleotides are composed od phosphate group, the bases i.e.

Adenosine, cytosine, guanine, thymine and pentose sugar.

In case of RNA, thymine is replaced by Uracil.

5) **Codon**:

Series of three adjacent bases in one polynucleotide chain of a DNA or RNA molecule which codes the specific amino acid.

6) **Genetic code:**

The sequence of nucleotides in a DNA or RNA molecule that determines the sequence of amino acid in the synthesis of proteins.

7) **Allele**:

A variation in DNA sequence at locus is called as an allele.

Diploid organism contains two alleles of each locus (gene)

Homozygous: Alleles can be identical.

Heterozygous: Allele can be different.

Hemizygous: If only one allele is present.

8) **Genotypes**:

Set of alleles present in the genome of an organism.

9) **Phenotypes**:

It is result of gene expression.

DNA are transferred to RNA.

mRNA is translated into protein, tRNA and rRNA determine the physical characteristic of an organism.[9,10]

Human affairs:

Genetics holds a special place in human affair. Not only it is relevant in the sense that other scientific disciplines are but also has much to tell us about the nature of our humanity and in this sense the biological science[11].

Genetics and health:

Congenital disease- Disease which present at birth. Hereditary or familial disease- Disease which transmitted from parents to offspring[12].

Medical genetics:

A) Monogenic/Mendelian disorders

B) Chromosomal aberrations

C) Polygenic disorder

A) Monogenic/Mendelian disorders:

1) **Autosomal recessive:**

The disease appears in offspring of unaffected parents.

e.g. Sickle cell anaemia, Phenylketonuria, Cystic fibrosis.

2) **Autosomal dominant:**

Affected male and females appears in each generation of pedigree.

Affected mother and father transmits the phenotype to both son and daughter.

e.g. Neurofibromatosis, Adult polycystic kidney disease, Huntington disease.

3) X-linked dominant disease:

Affected male passes the disorder to all daughters but none of their sons.

Affected heterozygous females married to unaffected males passes the condition to half their sons and daughters.

e.g. Fragile X syndrome, Vitamin D resistant rickets.

4) X- linked recessive:

Many ore male than females show the disorder.

All daughters of an affected male are carriers.

None of the sons of an affected male shows the disorder or are carrier.

e.g. Haemophilia, Colour blindness.

5) Co dominant inheritance:

Two different alleles of gene can be expressed and each allele makes a slightly different protein.

Both alleles influence the genetic trait or determines the characteristics of the genetic condition

e.g. ABO locus.

6) Mitochondrial inheritance:

This type of inheritance applies to gene in mitochondrial DNA.

Mitochondrial disorders can appear in every generation of a family and can affect both male and females, but fathers do not pass mitochondrial traits to their children[13,14,15].

e.g. Leber's hereditary optic neuropathy (LHON)

B) Chromosomal aberration:

Chromosomal aberrations are abnormalities in structure or number of chromosomes and are often responsible for genetic disorders.

There is alteration in the number or structure of chromosomes.

a) Numerical abnormalities-

i) Euploidy

ii) Trisomy (2n+1): Klinefelter's syndrome

iii) Monosomy (2n-1): Turner's syndrome

b) Structural abnormalities:

Brakeage followed by loss or rearrangement deletion, translocation

C) Polygenic disorder:

A genetic disorder that is caused by the combined action of more than one gene[16].

e.g. Asthma

Auto-immune disease

Cancers

Cleft palate

Diabetes

Hypertension

Heart disease

Inflammatory bowel disease

Mental retardation

Mood disorder

Obesity

Infertility

Chromosomal mutation:

Mutation is any hereditary change in the genetic male up of an individual other than that which may be caused by simple recombination of genes[17,18].

Types:

According to their occurrence:

1) **Somatic mutation:**

Mutation occur in body or somatic cell.

2) **Gametic mutation:**

Mutation occur in body or reproductive cell.

According to size:

1) **Point mutation**: Alteration of one or two nucleotide pair.

It includes:

i) Deletion mutation

ii) Insertion or addition mutation

iii) Substitution mutation

2) **Chromosomal mutation:** There is change in structural or number of individual chromosomes.

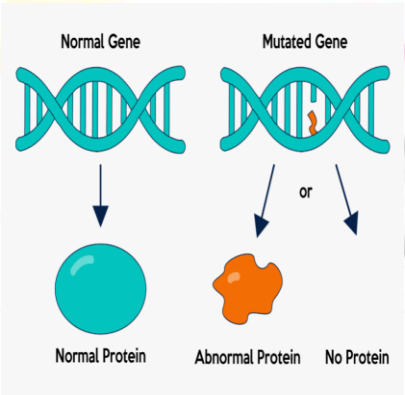

Fig No.2: Chromosomal Mutation

Human Genome Project (HGP):

HGP primarily found by the US government, department of energy, genome centre[19,20,21].

Goals of HGP:

1) To develop a genetic linkage map of human genome by identifying thousands of genetic materials and mapping them in the genome.
2) To obtain physical pam of human genome by cloning genomic DNA into cosmids.
3) To sequence the entire human genome.

Gene therapy:

- Introduction of normal gene into cells, that contains defective gene to reconstitute a missing protein product.
- Gene therapy used to correct the deficient phenotype so that sufficient amount of normal gene product are synthesized to improve a genetic disorder.
- Modifications of cell by transferring desired gene sequence into the genome.
- Two types of gene therapy:
  i) Somatic gene therapy
  ii) Germ line gene therapy[22]

DISCUSSION:

Genetic information can prove innocence and helps to identify and convict the guilty. To understand the genetics, it's important to know something about cells,

chromosomes and DNA. The cells are basic building blogs of life. A human body contains millions and millions of cells & Genetics is the only way to understand the hereditary disease[23].

CONCLUSION:

Genetics is the study of how living things receive common traits from previous generation. These traits ate described by genetic information. Human genetic is concerned with inheritance of human traits and their relation to human health, it deals with hereditary disorders and provide key to their prevention and control[24,25].

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