The Genome of an Organism

The Genome of an Organism:

A Structural and Functional Determinant

By

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PREFACE

Genome of an Organism: Structural and Functional Determinant tells that bacteria, viruses, plants, yeast, fungi- none of them can spare themselves from the involvement of genetics in their structural organization and daily body functions, like reproduction, and their interactions with the environment. Genetics significantly extends its role to agriculture, livestock, and biotechnology. Genetics intrigue the body to modify its organs and immune system following the transfer of these alterations in developmental and demographic mechanisms. Understanding such phenomenon is a fundamental requirement of any undergraduate and graduate student in the field of Zoology. Keeping in mind the requirements of Zoology students, it was strived to compose such a comprehensive compendium to provide information on structural and functional role of genome in the body of living organism. This book will be equally helpful to undergraduate and graduate students in general and those majoring in genetics and its applications in our daily lives.

This book contains 7 chapters. Chapter 1 is on "Genome as the Unit of Selection." As the name implies, its total focus is on the role of the genome in evolution and the ultimate selection of an organism in the environment. Chapter 2 is on "The Genetics of Species Formation." This chapter explains the role of genetics in the process of the formation of new species. Chapter 3 is "Genetic Variation, Gene Flow and New Species." As evident from the chapter's name of the chapter, it relates different but closely related concepts of genetics, then mechanisms of gene flow from parents to offspring and among the individuals. Chapter 4 is on “Behavioral Genetics”. As the name implies, behavior and genetics are very closely related. Whatever behavior an animal adapts to survive in the changing and hostile environment becomes part of its genetics, which is then transferred to its offspring.

Moreover, in the early developmental stages, an organism's genetics determines the behavior of the developing organism. Chapter 5 is on "Genetics and Developmental Evolution." Evolutionary processes work at all an organism's stages. The same is the case with genetics, which starts at the very beginning of an organism and controls the various activities and

development of organisms throughout their lives. Hence, the chapter explains the role of genetics in the early developmental processes of an organism. Chapter 6 is on "Genetic Theory of Adaptation." Some people may think that the relationship of adaptation with genetics is that genetics plays a fundamental role in all organisms' adaptive activities and functions.

Animals have to initiate adaptation when the environment changes or when the animal moves from one niche to another. Genetics, therefore, not only helps but also intrigues and then follows and preserves all these changes that animals have developed in their genome for further transmission and to produce a viable population. Chapter 7 is on "Genetic Variations in Natural Populations." This chapter may have a challenging relationship with the central theme of this book. But pondering the title, it talks about variations, and variations develop, which becomes the cause of natural selection. However, whether these variations are harmful or helpful is a different topic. It is well established that gene flows among individuals and populations. Genes also inherit character from parents to offspring. So, genes are the fundamental unit of heredity.

The present book aims to give an up-to-date overview of the Genomic studies and role of genome in manipulation structure and function of an organism. Hence this compendium can be of great help advanced students, junior researchers, faculty, and scientists involved in ecology, agriculture, plant and animal sciences, environmental microbiology, molecular biology, biochemistry, biotechnology, and other areas involving gene and genomic studies. We are thankful to all the contributors for their interests, significant contributions, and cooperation that made this humble endeavor possible because, without their unending support, motivation, and encouragement during the accomplishment of this grueling task, this would never have been succeeded.

Masroor Elahi Babar

Muhammad Ashraf

FOREWORD

Recently, there has been an explosion of genome sequence data which increasing day by day. This up-to-date textbook comprehensively covers diversified attributes of genome in the structure, organization, and function of an organism. Developmental aspects of an organism, its adaption to new environment with subsequent favorable changes and ultimate transformation into new species have been described in detail. The book takes full advantage of the authors' experience working in post-genomic revolutions in the world with an introduction to the molecular and genetic aspects and their beneficial effects for the continuation of life as we know it.

This book titled, "Genome of an Organism: Structural and Functional Determinant," presented here, is of paramount importance because it will help to educate the next generation of students about the role of genome in building an organism following performance of varying bodily functions because genome determines the future line of development and working of an organism. The way of presentation of this book brings forth the unique perspectives genome and its functional qualities shedding light on a very complex and vital subject that could otherwise be intractable. This book might be unusual for an academic text, but it comprehensively covers academic and applied aspects gene and genetics. It is worth mentioning that environmental problems exist, and ecological changes are occurring persistently. Animals must cope with these changes and modify themselves physically and psychologically for survival and fitness. This is the place where this book comes in. It is both time-sensitive and evolving. Complete understanding how animal manipulates its body under changing circumstances may not exist despite very compressive investigative and written works. We have struggled very humbly to contribute in understanding and explaining such genome based mechanisms frequently occurring in the life of an organism.

I hope this work, when shared freely and widely, will be an educational milestone as humanity struggles to understand and solve the enormous problems regarding genetics and its various aspects and where it serves basic uplift in any aspect of life of an organism. We are sure that it provide an intellectual foundation allowing students to become the engines that

move and use genetics for a variety of applications in our daily life. Therefore, this compendium can help provide long-lasting guidelines in studying genetics and its related disciplines and fields if used and applied in its actual letter and spirit.

CHAPTER 1

MUTATION THEORY AND PHENOTYPIC EVOLUTION

Summary:

After a brief introduction to mutation theory and phenotypic evolution, it talks about the causes of mutation and its types. It further discusses the beneficial utility of mutation theory, its significance, and its contradictions. Then, it further elaborates on the role of mutation theory in the evolution of eukaryotes and how changes in the coding regions of genes affect the physiological features of animals. This chapter further discusses the changes in the regulatory regions of genes due to mutations and how they impact the evolution of morphological characters and the diversity of animals. It ends with highlights on existing and future views on animal evolution.

Introduction

Mutations are the random changes in the DNA base pairs. These changes are then inherited from parents to offspring. During the mutation process, bases are added to the existing setup or deleted or replaced by other bases. Evolutionary naturalists opine that most mutations are harmful or harmless, but they still stress that rare, favorable mutations have occurred, resulting in the evolution of all forms of life. Genes, the parts of DNA carriers of heredity material, sometimes also undergo random changes. These random changes express themselves in the genome of offspring. This genome is then transferred from generation to generation. Natural selection selects suitable organisms that can adapt, survive, and reproduce in the given environment. People choose the most suitable microorganisms based on their fitness and their differences and variations. The source of this variation is always mutation. Mutations may be induced and spontaneous, with known and unknown causes. The action of physical and chemical agents called mutagens on the DNA molecule is the leading cause of the mutation. Mutagens may be chemicals, viruses, X-rays,

cosmic rays, and ionizing radiations. Mutations in germ cells will cause genetic diseases in the offspring, while mutations in somatic cells can cause cancer. Some agents like arsenic, asbestos, benzidine, vinyl chloride, thorium dioxide, chromium compounds, mustard gas, and melphalan may be both mutagenic and carcinogenic (Fig. 1..1, Fig. 1.2, Fig. 1..3, and Fig. 1.4).

Fig. 1.1. Most shocking real mutations in animals (Source: www.animali-life-club)

Fig. 1.2. Mutations(Source:biologyreference.com)

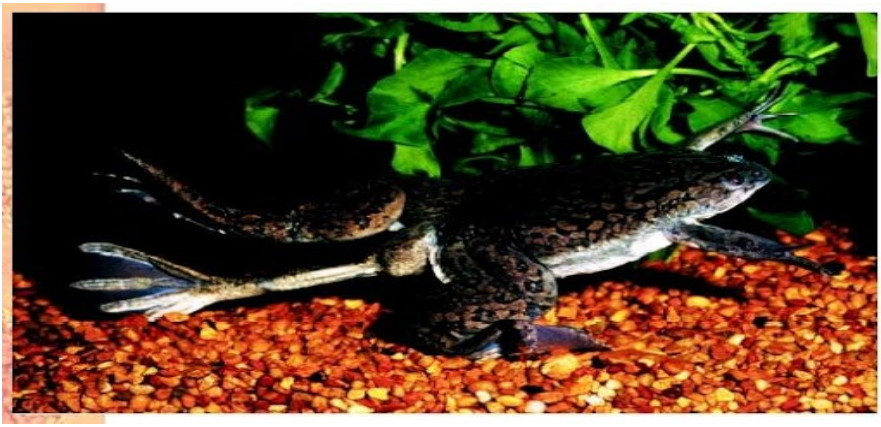

Fig. 1.3. Mutation (Source: biologyreference.com)

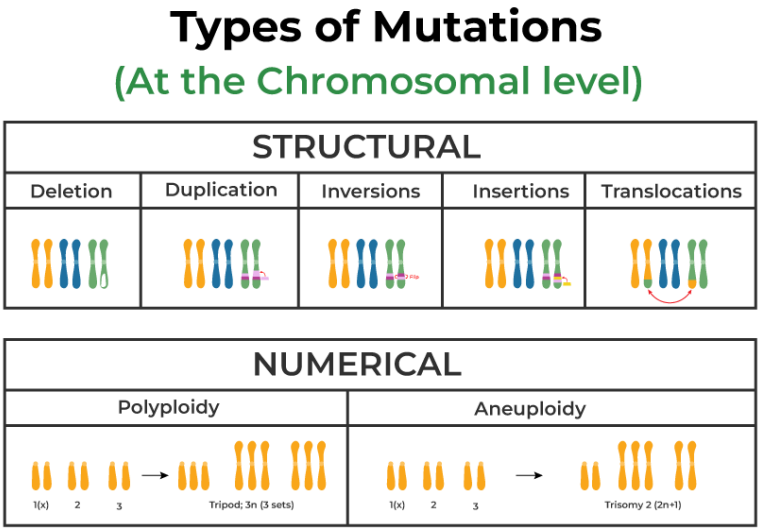

Fig. 1.4. Mutations (Source: Geeksfor Geeks Sonchhaya Education PVT.LTD., Noida Uttar Pradesh India)

Causes of Mutations and Their Types

Genes control the development and function of an organism. Mutations act on the encoded protein. They change the protein structure and make that protein express partially or not. As the mutation changes the DNA sequence, it damages a cell or an organism. The appearance of an individual, based on an animal's genotype, whether regular or mutated, is its phenotype. Animals may be haploid (carrying a single set of chromosomes), e.g., unicellular organisms, or diploid (having two copies of chromosomes), e.g., fruit flies, mice, humans, etc. Different forms of regular or mutated genes present on chromosomes are called alleles. Diploid organisms, due to double chromosomes, carry two copies of each gene. These genes may have identical or different alleles and may be homozygous or heterozygous for that particular gene. The mutation will be recessive in homozygous individuals for that particular gene, while the mutation will be dominant in heterozygous individuals.

Recessive mutations result in the loss of function of the encoded protein, while dominant mutations result in a gain of function. This notion is, however, always true. For example, in *Missense mutation*, one amino acid is replaced by another, which may not impact the protein's function. Nonetheless, this all depends on the conservative or non-conservative nature of change. Secondly, the role of amino acids in that change also matters. The function of *nonsense mutation* is to change an amino acid to a STOP codon, which ultimately terminates the process of protein synthesis (translation). Silent mutation either speeds up or slows down the process of mutation. Though there is no change in protein composition, phenotypic effects are apparent in this case. The last type of the four types is called frame-hift- mutation. In this case, many bases are inserted or deleted from the gene. A lot of amino acid changes take place in this mutation. Further to this, a premature STOP codon is inserted, and the process of protein synthesis halts forthwith (Fig. 1.5, Fig. 1.6, Fig. 1.7, Fig. 1.8, Fig. 1.9, Fig. 1.10, Fig. 1.11, and Fig. 1.12).

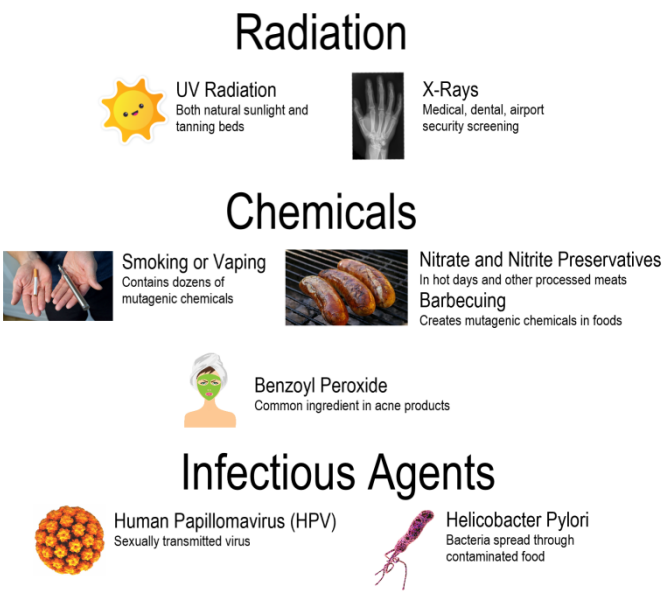

Fig. 1.5. Mutations, types, and causes (Source: Seeker, 2016)

a) Apo lipoprotein

All humans have a gene which are said to be apolipoprotein. The function of this gene is to transport cholesterol via the process of bloodstream. It is considered more beneficially due to the removal of cholestrol from arteries.

Apo lipoproteins are antioxidants that are used to prevent from swallowing. People have a gene named as PCSK9 that cause low expression of heart disease.

b) Increased bone density

The gene which caused bone density in human being are said to be low lipoprotein related to protein 5 (LRP5).

This variation occurs in a young person of Midwest family in car crash. Due to this crash the person bone not is break. X RAY analysis shows that the person belongs to those families which have stronger bones than average.

c) Malaria resistance

These variations occur in humans due to the hemoglobin variation that make it like sickle shape. When one copy present it cause the disease like malaria resistance but when there are two copies present it caused the disease like sickle cell anemia that is not actually by this mutation. The missing of one copy of that gene in human causes 29% chances to get malaria. On the other hand people with two copies of this gene respond to 93% of that disease.

d) Tetra chromatic vision

Different genes present in humans shows different color vision of light. The gene which has one kind of cone show blue color at chromosome at 7.The other two kinds shows the effect of both red and green on the X chromosome.

2) Beneficial mutation in organisms

Mutation that occurs in organism are said to be somatic or germ line. Germ line mutations are those mutations that are tans formed from parents to offspring through reproduction cells. Due to the long term effect of this mutation it is considered as the beneficial mutation. The examples of beneficial mutation in organism are as follow.

a) Nylonase: nylon bacteria

Nylonase are the most authentic example of beneficial mutation in bacteria. Nylonase eat the short molecule of nylon 6.Mutation occur in that bacteria due to the insertion of single nucleotide in genetic material. This mutation was found in 1940 due to the presence of nylon.

b) Gene mutation: Almond Trees

The species which have almond contain amygdalin. Amygdalin is a chemical that convert the cyanide into human body. A single gene mutation in this type of almond trees shows that there is no long production of amygladin.

c) Murry Gray: A Breed of Beef Cattle

Murry gray are known as cattle bread which can be obtained from cow species. In this case the specific cow produced more productive calves than others. Soon after the differences were identified and breed with positive characters were produced which ultimately became popular in all regions of Australia.

References

https://www.google.com.pk/?gws_rd=cr&ei=ZgvnWOeID4HyUOu9uJAB#q=mutation

https://www.google.com.pk/?gws_rd=cr&ei=ZgvnWOeID4HyUOu9uJAB#q=mutation+history

https://www.google.com.pk/?gws_rd=cr&ei=ZgvnWOeID4HyUOu9uJAB#q=mutation+causes

https://www.google.com.pk/?gws_rd=cr&ei=ZgvnWOeID4HyUOu9uJAB#q=mutation+types

http://www.biotechnologyforums.com

Fig. 1.6. Mutations, types, and causes (Source: slideshare.net)

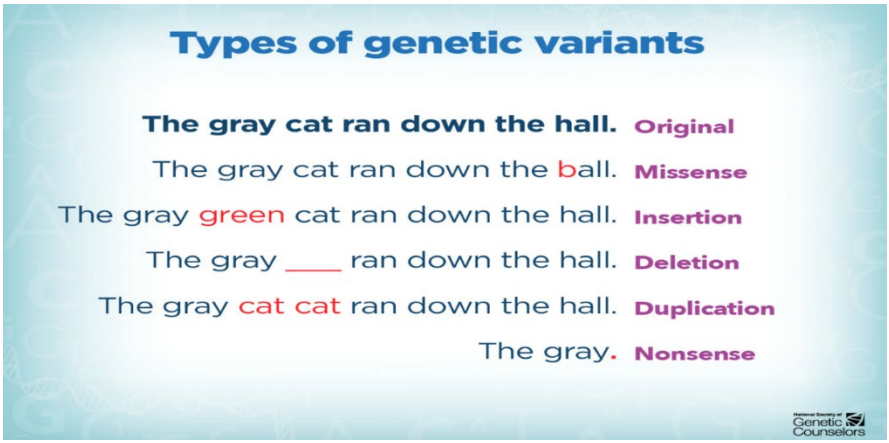

Fig. 1.7. Types of mutations (Source: parentprojectmd.org)

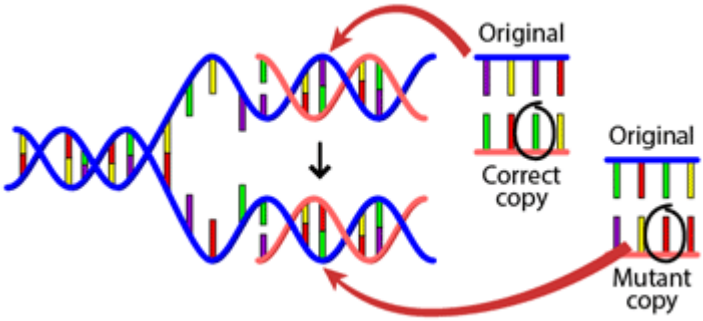

Fig. 1.8. Causes of mutation (Source: evolutition.berkely.edu)

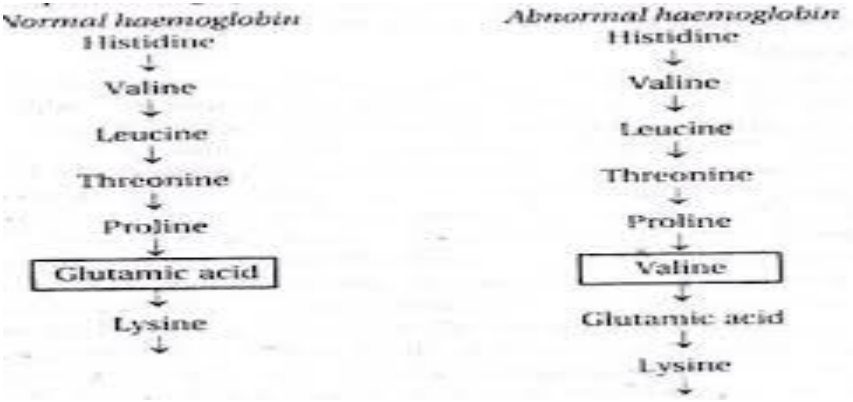

Fig. 1.9. Gene mutations, causes, examples and types (Source: biologydiscussion.com)

Gene Mutations

- This type of mutation involves a change in one or more of the nucleotides in a strand of DNA.
- There are four types of gene mutation:
- 1. Substitution of a nucleotide.
- 2. Inversion of two or more nucleotides.
- 3. Deletion of a nucleotide.
- 4. Insertion of a nucleotide.

Fig. 1.10. Types of gene mutations (Source: slideplayer.com)

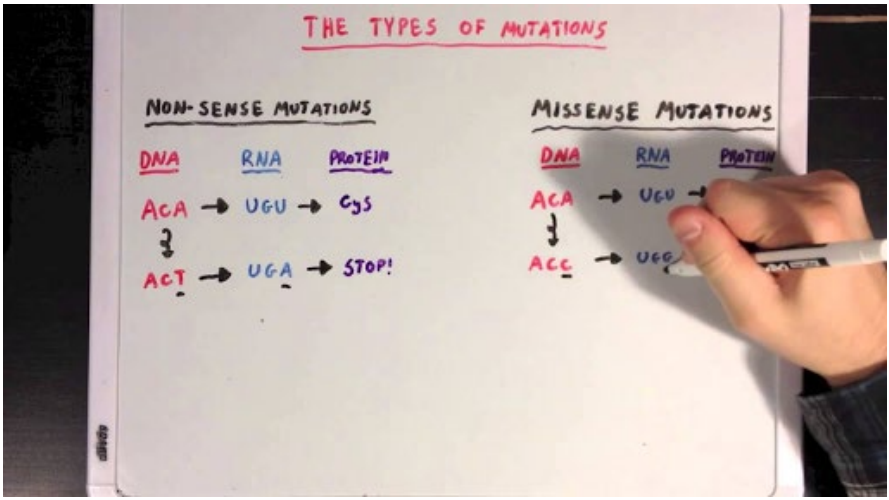

Fig. 1.11. The different types of mutations (Source: khan. academy.org)

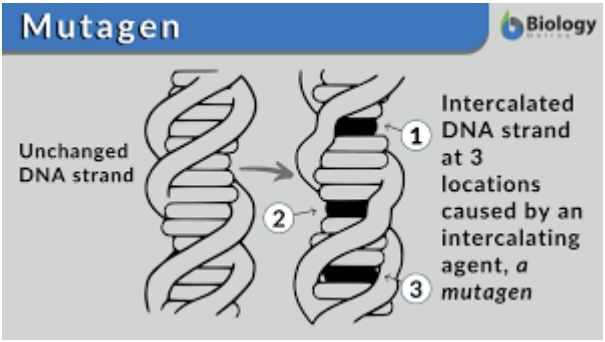

Fig. 1.12. Mutagen, definition, and examples (Source: biologyonline.com)

MUTATION THEORY

While working on evening primrose (*Oenothera lamackiana*), which escaped from a garden in an abandoned tomato field and produced many new varieties, De Vries (1886) named it mutation and later published his work as a mutation theory. He was of the view that such large scale and frequent changes were the reason for the origin of species instead of due to Darwinian gradualism. This theory sufficiently exerted its influence during the early decades of the twentieth century until scientists abandoned it

(Endersby, 2013). During this process, chromosomal abnormalities resulted in large-scale variations in primrose. Now the mutation is generally restricted to distinct changes in the sequence of DNA (Stableford, and Langford, 2018). Unlike Darwin, he opined speciation does not occur due to continuous variation but is the result of the appearance of sudden variations. Such sudden variations are, at this moment, called mutations. Following generations receive such mutation caused variations inherited from parents to offspring (Fig. 1.13, Fig. 1.14, Fig. 19.15, Fig. 19.16, Fig. 1.17, Fig. 1.18).

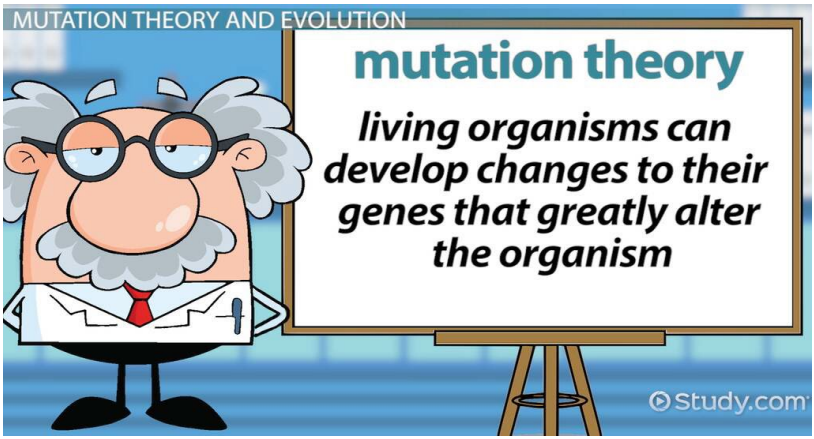

Fig1.13. Mutation theory (Source: study.com)

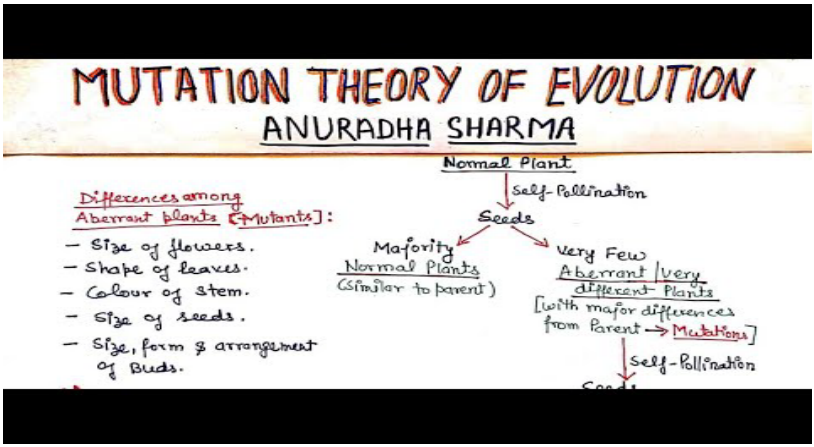

Fig.1.14. Mutation theory of evolution (Source: youtube.com)

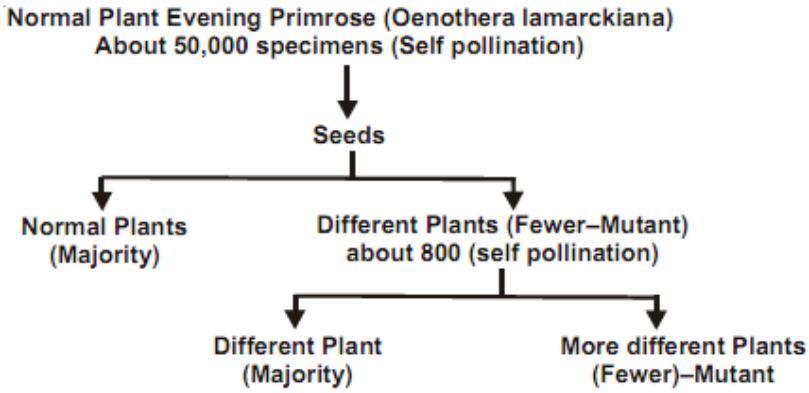

Fig. 1.15. Mutation theory (Source: expertsmind.com)

Fig.1.16. Mutation theory (Source: rajusbiology.com)

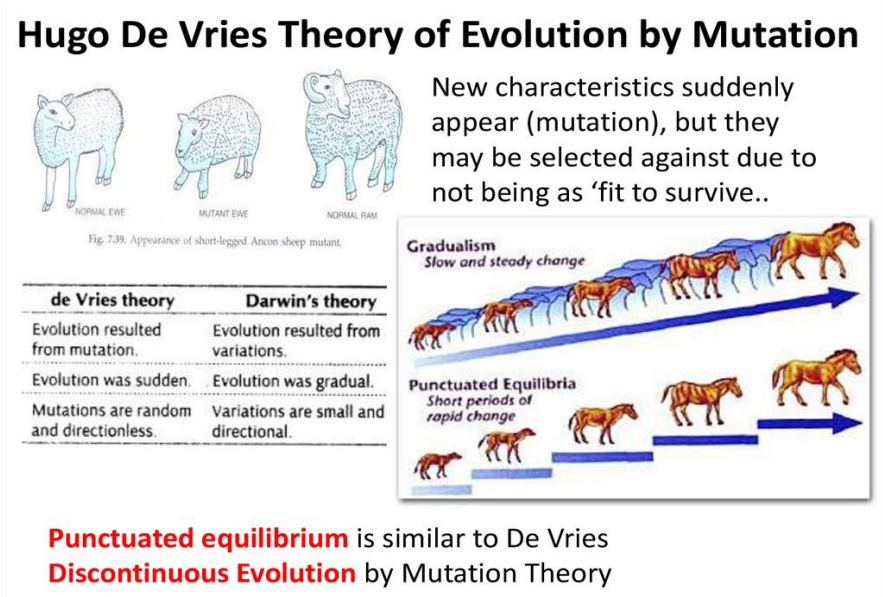

Fig. 1.17. Mutations and mutagenesis (Source: en.ppt.online.org)

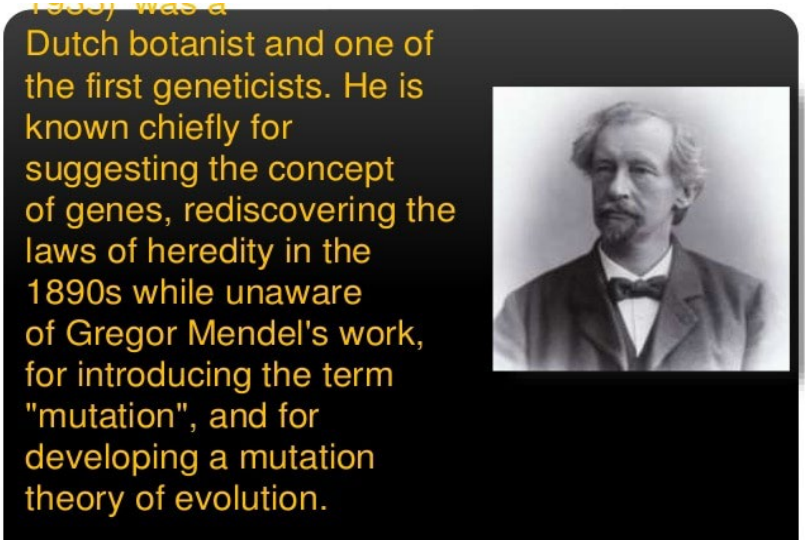

Fig. 1.18. Evolution through mutation (Source: slideshare.net)

Mutation Theory says that mutations are discontinuous variations that serve as raw material for evolution. All of a sudden, the appearance of mutations becomes operational immediately and is not a normal process in the life of an organism. Mutations are directionless and occur in one generation, and the offspring inherit them. Nature selects advantageous mutations and eliminates lethal ones, preventing less harmful and inoperable ones. Evolution is not a regular process. Instead, it works with numerous interruptions—irregular and sporadic process. The development of new species is the outcome of the accumulation of variations. As the mutations give rise to variations, a single mutation might result in the devolvement of new species (Fig. 1.19, Fig.1.20, Fig.1.21, and Fig. 1.22).

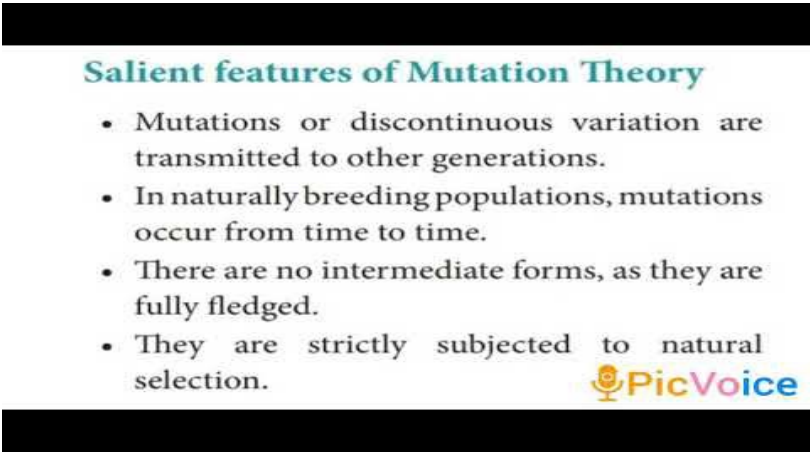

Fig. 1.19. Salient features of mutation theory (Source: youtube.com)

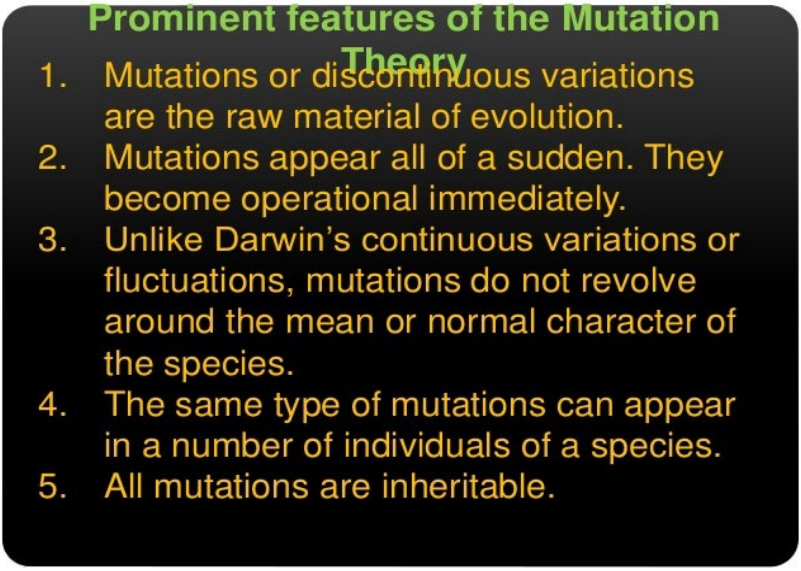

Fig. 1.20. Theory of evolution through mutation (Source: slideshare.com)

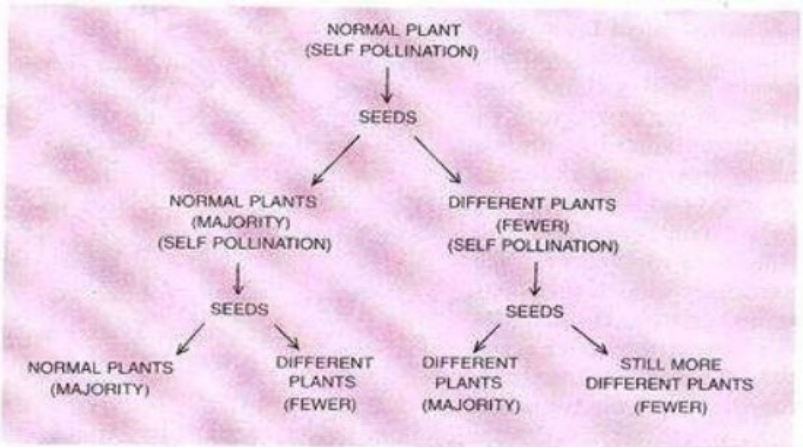

Fig. 1.21. Theory of evolution through mutation (Source: slideshare.net)

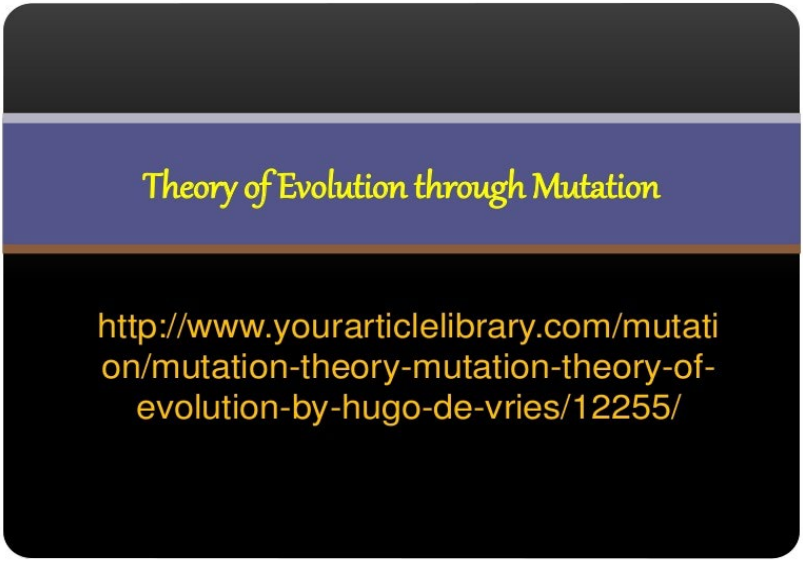

Fig. 1.22. Theory of evolution through mutation (Source: slideshare.net)

The usefulness of Mutation Theory

Mutations cause all variations and hence provide a raw material for evolution. Mutation can explain both progressive and retrogressive evolutions. The ratio of mutations is different from organism to organism and from organism part to part; hence, mutation theory can explain both forms of an individual, whether it is changed or unchanged. Previously, researchers observed several mutations and mutated organisms. Therefore, they are a source of new varieties. For example, the sudden appearance of Ancon Sheep (a short-legged variety) in 1791 Massachusetts, hornless Cattle from the homed cattle in 1889, hairless cats, and double-toed cats are a few examples of mutations. In addition to animals' development of delicious apples, Cicer gigas, Noval orange, and Red Sunflower are some examples in plants too (Fig. 1.23, Fig. 1.24, and Fig. 19.25).

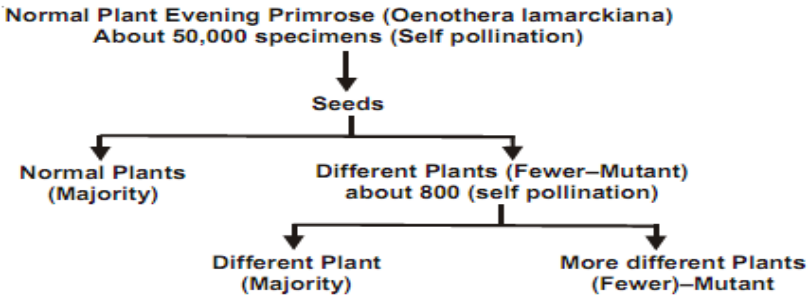

Fig. 1.23. Mutation theory (Source: expertsmind.com)

Fig. 1.24. Theory of evolution through mutation (Source: slideshare.com)

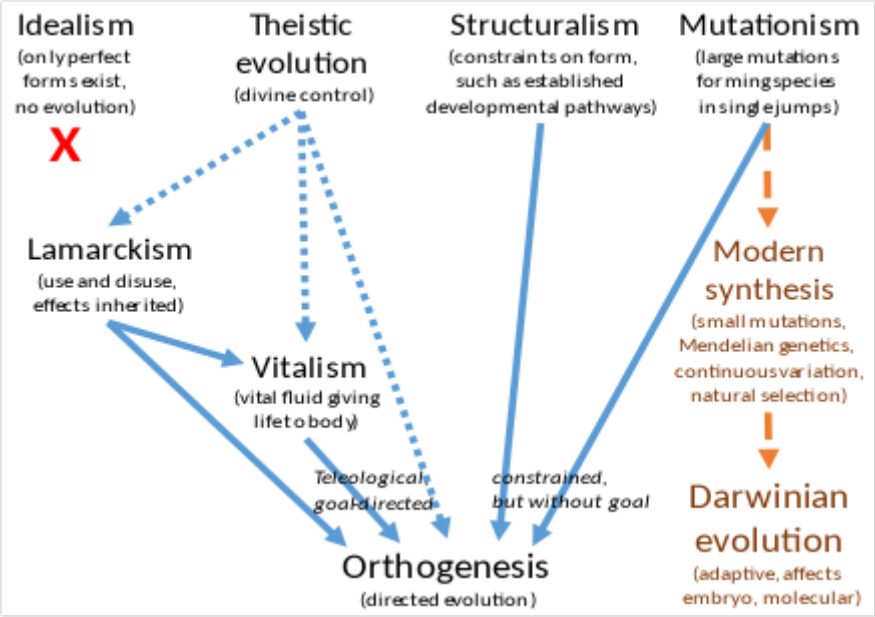

Fig. 1.25. Mutationism (Source: Philip, 2013)

Mutation Theory(contradictions): Some scientists reported that Hugo de Vries worked on *Oenothera lamarckiana* (plant) for his experimental studies. It was not a typical plant. This plant developed differently due to chromosomal aberrations and was in heterozygous form. They are further of the opinion that, in contradiction to Hugo de Vries, natural mutations commonly do not occur. The majority of mutations are not in favor of an organism. Dominant traits participate in evolution while mutations are generally recessive, hence the tiny contribution to evolution. Even mutations are not capable enough to explain the role of nature in evolution. Further phenomena such as pollination by insects, mutual dependence of flowers, and mimicry are hard to explain by Mutation Theory (Fig. 1.26, and Fig. 1.27).

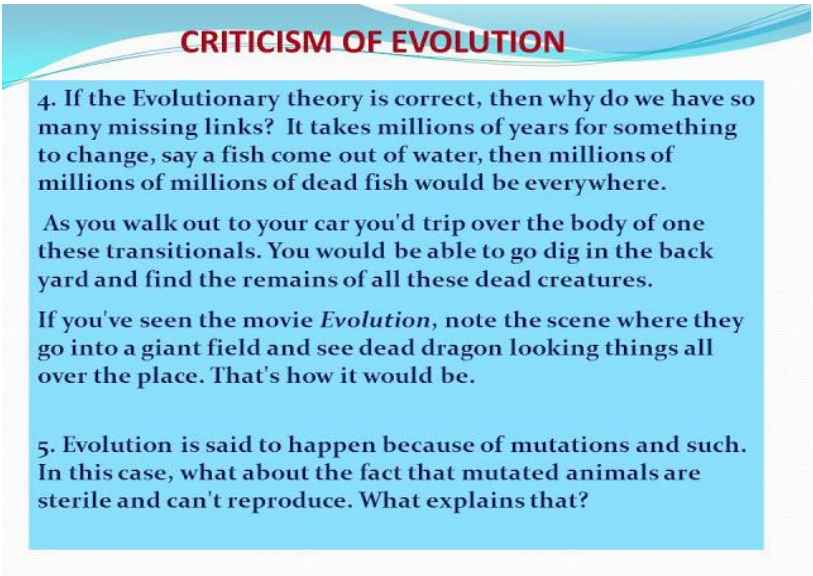

Fig.1.26. Criticism on evolution (Source: pinterest.com)

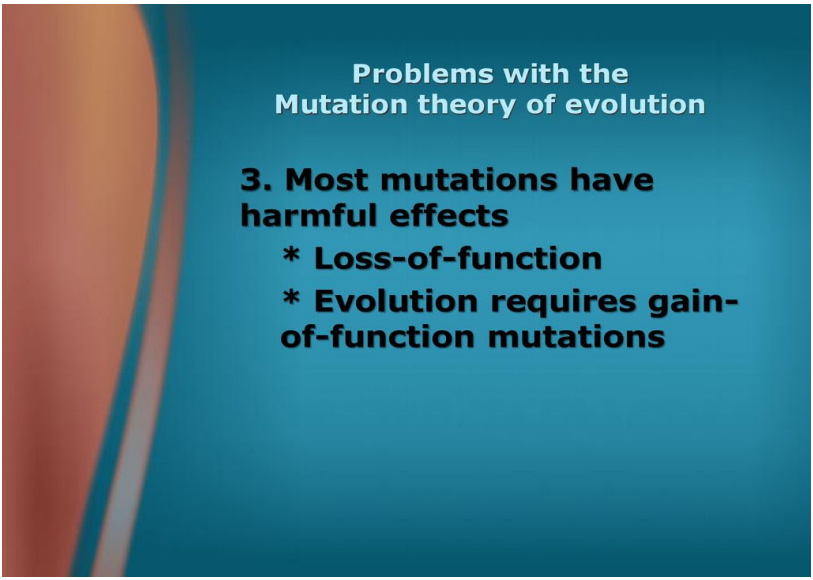

Fig. 1.27. Problems with Mutation Theory (Source: slideplayer.com)

Significance of Mutation Theory: There is still controversy about whether mutations are harmful or helpful. Chromosomal changes due to mutations play a pivotal role in evolutionary processes. If mutations occur in a single base and do not change the reading frame, they likely have no effect. Nonetheless, it is a general observation that sometimes mutations have no effect, and sometimes, they induce lethal consequences in an organism. TAC CCC GGG is a reasonably generic coding sequence. This code occurs in the human genome repeatedly. Following its transcription in mRNA, the process of protein synthesis starts. Different codons (GGG, GGA, GGC, GGU) encode different amino acids. Change at the third position of the codon has no effect. No alteration in a protein's sequence of amino acids will cause any phenotypic change in an organism. If mutations do occur, DNA repair mechanisms become active. They mend most of the changes before they become permanent. Even organisms have inherently built-up mechanisms which can eliminate permanently mutated somatic cells. Nonetheless, when a mutation causes changes in other nucleotides in the sequence, it dramatically affects the organism. For example, a change in the start codon prevents the initiation of the translation process. Conversion of glycine (simple amino acid) into the nonpolar (and relatively large) tryptophan (UGG codon) dramatically interrupts the function of the protein. If the stop codon changes, the translation process extends until the next in-frame codon.

In the past, scientists claimed that mutation is a DNA or RNA sequence change. Accordingly, mutation can be either germline or somatic in multicellular organisms. Germline mutations occur in the DNA of sex cells; potentially, they can be severe. Therefore, gametes produced during production are passed on to the next generation. Accordingly, the zygote will possess mutations, and each cell of the resulting organism will be affected. The appearance of phenotypic diseases due to these mutations is called heredity disease. Somatic mutations occur in body cells. Due to these mutations, some severe medical conditions may develop, but they are not inherited; hence, cancer is not called heredity (Fig.1.28, Fig. 1.29, Fig. 1.30, Fig. 1.31, Fig. 1.32, Fig. 1.33, and Fig. 1.34).